B

B: Refers to back cross generation; the numbers of back crosses are indicated by subscripts, e.g., B_1. ►back cross

β: ►Error types, ►power of a test

7B2 (secretogranin V, chromogranin): A 25–29 kDa pro-hormone which is processed to a 18–21 kDa neuroendocrine chaperone (distantly related to chaperonins-60/10) in the secretory pathway. It is widely found in animals, and it is encoded in human chromosome 15q13-q14 as SGNE-1. It is an inhibitor/activator of the pro-hormone convertase PC2 enzyme but not of other PCs. ►Golgi apparatus, ►chaperonins; Umemura S et al 2001 Pathol Int 51:667.

B104: Refers to *Drosophila* retroposon, which is similar to copia, gypsy and others. ►Copia

β Barrel: The polypeptide chain of a membrane protein forms a folded up β sheet arranged in the shape of a barrel. ►Protein structure, ►membrane proteins, ►transmembrane β barrel detection in bacteria: http://cubic.bioc.columbia.edu/services/proftmb/; http://bioinformatics.bc.edu/clotelab/transFold/; http://tmbeta-genome.cbrc.jp/annotation/.

B1 B Cell: These are fetal and early infant B cells which may be found in excess in leukemias and autoimmune diseases. ►B lymphocyte, ►leukemias

B Box: This refers to a part of the internal control region of tRNA and some other genes. See also A box and internal control region of pol III genes. ►Trna, ►pol III

B.C.E. (before the common era): An archaeological concept for the designation of age of event(s) or artifacts based on different criteria such as carbon dating, old scriptures, etc.

B Cell: B lymphocyte, B lymphocyte receptor.

B Chromosome: This is the accessory (supernumerary) chromosome. They are generally heterochromatic and carry no major genes yet they may be present in several copies in many plants. B chromosomes have no homology to the regular chromosomal set (A chromosomes) and are prone to non-disjunction because their centromeres appear to be defective. If A-B translocations are constructed, the placement of genes to chromosomes, arms or even shorter regions may be facilitated. The principle of the use of A-B chromosome translocations is presented in the diagram here. The A chromosome or a translocated segment carries the dominant *A* allele and the B chromosome has no counterpart to it, therefore a null phenotype (*a*) appears in its absence. In the diagram the male has the translocation and the female is homozygous recessive for the *a* allele. In case there is no B chromosomal non-disjunction—when the chromosomal constitution is as diagramed—both the endosperm and the embryo express the dominant gene. In the case of non-disjunction the phenotypic effects depend on the constitution of the sperm, which fertilizes the diploid polar nucleus of the endosperm or the embryo, respectively (see Fig. B1).

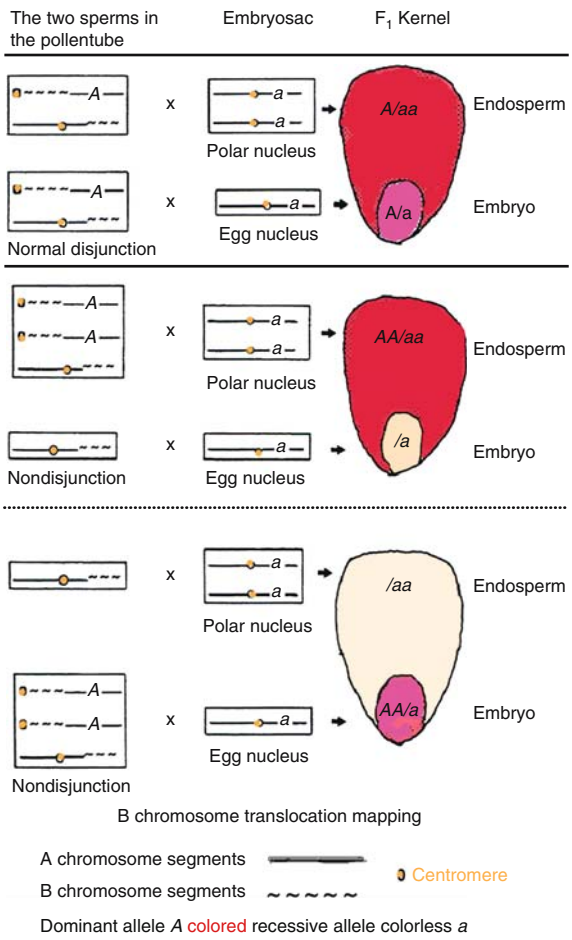

Figure B1. B chromosome translocation mapping

This type of difference reveals the approximate physical and genetic position of the locus. Had the dominant allele been outside the translocated segment, the recessive allele would not have been unmasked. The example given here describes the most favorable case when the consequence of the translocation can be identified without tissue-specificity.

B chromosomes have been reported in ~1300 species of plants and in ~500 species of invertebrate and vertebrate species of animals. The transmission of the B chromosomes varies in different species and it may be preferential in the female member (e.g., in grasshoppers) or in some wasps by the male. A total of 19 B chromosome sequences of maize have been isolated by microdissection and cloned and characterized according to their information content (Cheng Y-M, Lin B-Y 2003 Genetics 164:290). ►mapping genetic, ►trisomic analysis, ►translocation genetic, ►centromere, ►centromere silencing; Beckett JB 1982 J Hered 73:29; Page BT et al 2001 Genetics 159:291; Camacho JPM et al 2000 Philos Trans R Soc Lond B 355:163.

B DNA: ►DNA type, ►Z DNA

B Lymphocyte (B cell): This is responsible for humoral antibody synthesis and secretion. Pro-apoptotic proteins, BAX and BAK, limit the number of B cells. The deletion of *Bax* and *Bak* genes leads to autoimmune disease in mice (Takeuchi O et al 2005 Proc Natl Acad Sci USA 102:11272). There are two types of B cells, B1 and B2. T cells activate the common B2 cells. Its differentiation from hematopoietic cells (plasma cells) depends on transcription factors (XBP-1) of the bone marrow, cytokines and antigens, T_H cells and a series of non-receptor and receptor tyrosine kinases and phosphatases, proteins mediating the pathway shown in italics (see Fig. B2) in the diagram outlining the developmental pathway of B lymphocytes. The B1 cells also employ an RNA editing system for the diversification and amplification of their antigen receptors in contrast to T cells, which rely on the V(D)J recombination system, and appear independent of T cell activation. ►T cells, ►apoptosis, ►EBF, ►CpG motifs, ►CD40, ►TAPA-1, ►blood, ►immune reactions, ►germinal center, ►Blimp-1, ►BTK, ►surrogate chains, ►Pax, ►immunoglobulins, ►BASH, ►V(D)J, ►RNA editing, ►XBP; Fagarasan S, Honjo T 2000 Science 290:89; ►blood, ►bone marrow, ►thymus, ►immune system; Hardy RR, Hayakawa K 2001 Annu Rev Immunol 19:595; Reimold AM et al 2001 Nature [Lond] 412:300; Berland R, Wortis HH 2002 Annu Rev Immunol 20:253.

B Lymphocyte Receptor (B cell receptor, BCR): This is constructed from the membrane-bound immunoglobulin molecules IgM and IgD as receptors and after the attachment of the antigen they can also use IgG, IgA and IgE (see Fig. B3). The intracellularly linked IgA and IgB heterodimer that is the signaling portion of the receptor and the complex transmits the immunoglobulins recognized by the BCR. All the immunoglobulins are attached to the receptors associated with the IgA and IgB heterodimer through their heavy-chain terminal amino acids. This tail consists of 3 amino acids in IgM and IgD but it has 28 amino acids in IgG and IgE. In both, part of the A-B heterodimer includes an ITAM (immunoreceptor tyrosine-based activation motif). The latter is instrumental in activating the Sky and Lyn protein tyrosine kinases that mediate the switching to the IgG, IgA and IgE molecules when the appropriate antigen is presented. These tails are required for the endosomal targeting of the immunoglobulins. The B cell linker protein (BLNK) is also required for the normal development of B lymphocytes. ►B lymphocyte, ►BASH, ►immune system, ►ITAM, ►ITIM, ►BAP, ►endocytosis, ►agammaglobulinemia; Meffre E et al 2001 J Clin Invest 108:879; Vilches C, Parham P 2002 Annu Rev Immunol 20:217.

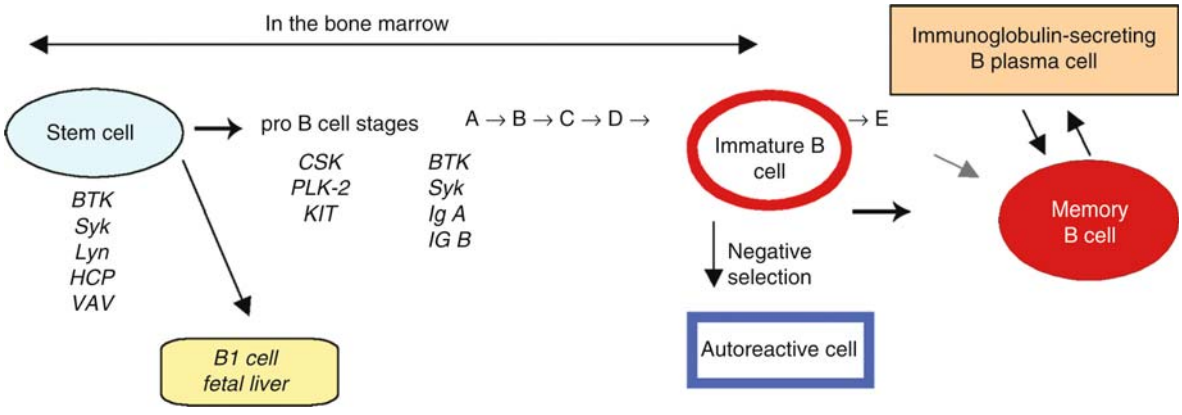

Figure B2. B lymphocyte-bone marrow. During the pro-B cell stages in the bone marrow first the IgH and then the IgL immunoglobulin chains are rearranged through signaling by the pre-BCR (B cell receptor). After expression of the BCR, some cells leave the bone marrow and mature into IgM and IgD cells that move between the peripheral lymphoid organs

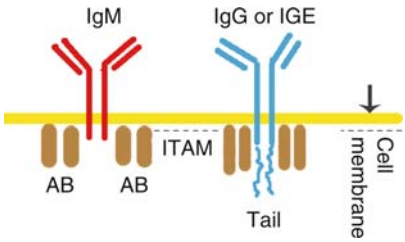

Figure B3. B lymphocyte receptor

β Oxidation: Refers to the degradation of fatty acids at the β carbon into acetyl-coenzyme A. ►fatty acids, ►acetyl CoA

B7 Protein: This is required for the activation of B cells. It is recognized by CD28 on the surface of the antigen-presenting cells. The other signal required for B cell activation is a foreign peptide antigen, associated with class II MHC molecule on the surface of an antigen-presenting cell. This is the same as BB 1 or CD80. ►CD40, ►antigen presenting cells, ►MHC, ►B cell, ►co-stimulator, ►ICOS; Yoshinaga SK et al 2000 Int Immunol 12:1439.

B1, B2 Repeats: These are highly dispersed SINE elements in the mouse genome. ►SINE

B2 RNA: This is a small (178-nucleotide) non-coding RNA in the mouse regulating RNA polymerase III and II (Allen TA et al 2004 Nature Struct Biol 11:816). ►RNA regulatory

β Sheet: Refers to the secondary structure of proteins where relaxed polypeptide chains run in a close parallel or an antiparallel arrangement. (See diagram on protein structure).

β Sheet Breaker Peptides (iAβ5): 4 amino acids (LVFF) in the 17–20 N-terminal domain of amyloid β protein (Aβ) may be substituted (particularly by proline) in this region and may alter the conformation of β sheets and reduce the formation of and disassemble the already formed amyloid fibrils characteristic of Alzheimer's disease (see Fig. B4). A charged amino acid may be added to increase solubility (Leu, Pro, Phe, Phe, Asp). Shorter or different peptides may have opposite effects. The iAβ5 molecules may provide an approach to the treatment of Alzheimer's disease. (Alzheimer's disease, Soto C et al 2000 Lancet 355:192).

Figure B4. β sheet breaker

Babelomics: This is the web server for the interpretation of microarray data as well as gene annotation and interaction networks: http://www.fatigoplus.org.

Baboon (*Papio*): ►Cercopithecidae, ►xenotransplantation

BAC (bacterial artificial chromosome): Refers to bacterial cloning vector (derived from F plasmid) that can accommodate up to 350 kb (most commonly 120–150 kb) DNA sequences and has a considerably lower error rate than the still larger capacity yeast artificial chromosome (YAC). BACs usually exist in a single copy per cell. *Random BAC*s are selected at random from a genomic library and are then shotgun-sequenced. Most BAC vectors lack selectable markers suitable for mammalian cell selection but can be retrofitted by employing the Cre/loxP site-specific recombination system. ►vectors, ►PAC, ►YAC, ►BIBAC, ►shotgun sequencing, ►genome projects, ►F plasmid, ►Cre/loxP, ►selectable marker, ►TAC; Wang Z et al 2001 Genome Res 11:137, http://www.nih. gov/science/models/bacsequencing/end_sequencing_project.html.

BACE (beta site APP-cleaving enzyme): The amyloid precursor protein (APP) cleavage enzyme—a β-secretase [a membrane-bound aspartyl protease]—is involved in the production of brain plaques in Alzheimer's disease. The transmembrane BACE splits APP into soluble β-sAPP and a membrane-attached carboxy-terminal fragment, CTF-β. The latter is expected to be the substrate for γ-secretase. Bace1 also controls the myelination of peripheral nerves (Willem M et al 2006 Science 314:664). BACE1 deficient mice do not generate Aβ peptide, responsible for Alzheimer plaques and appear to be normal. BACE is encoded at human chromosome 11q23.3. Its homolog BACE2 is in chromosome 21, near the region critical in Down's syndrome trisomy. Research on drug development for the treatment of Alzheimer's disease has a likely target in BACE. ►Alzheimer's disease, ►secretase, ►Down's syndrome, ►presenilin, ►GSK; Roberds SL et al 2001 Hum Mol Genet 10:1317, substrate binding; Gorfe AA, Catfisch A 2005 Structure 13:1487.

BACH (BRCA1-associated carboxy-terminal helicase): This is a DEAH family protein. Phosphorylated BACH helicase interacts with the C-terminal domain of BRCA1 (BRCT) and in the case of DNA damage it controls the G2→M transition in the cell cycle as a checkpoint. BRCT mediates the tumor-suppressor function of BRCA1. BACH1 seems to be identical to Fanconi anemia FANCJ, and is essential for homologous recombination (Litman R et al 2005 Cancer Cell 8:255). ►breast cancer, ►DEAH box proteins, ►cell cycle, ►checkpoint; Yu X et al 2003 Science 302:639.

B

Bach, Johann Sebastian: One of the greatest geniuses of classical music, Bach's (1685–1750) family included over 50 more or less renowned organists, cantors and musicians (see Fig. B5). Of the four surviving children from his first marriage to his second cousin Maria Barbara Bach, three were musicians (inbreeding coefficient 1/64 [three offspring died in infancy]). Five of the 13 children from his second marriage to unrelated singer Anna Magdalena Wilcken (assortative mating) survived and of these three were musically talented. This family tree reveals that musical ability may be controlled by relatively few genes, and the cultural environment may also play a major role. Recent studies have demonstrated that musical talent is correlated with stronger development of the left planum temporale, increased leftward asymmetry of the cortex. ►dysmelodia, ►musical talent, ►Mozart, ►Beethoven; Wolff C 2000 Johann Sebastian Bach: The learned musician, Norton, New York.

Figure B5. Bach and Bach morning prayer. Morning prayers in the family of Sebastian Bach, painted in 1870 by American artist Toby Edward Rosenthal in Europe. Members of the large family are either playing music or singing. From Music with Ease (http://www.music-with-ease.com); courtesy of Paul Wagner.

Bacillus: This is a rod-shape bacterium. (►*Bacillus subtilis*, ►*Bacillus thüringiensis*)

Figure B6. Bacillus

Bacillus Calmette-Guerin (BCG): This attenuated form of *Mycobacterium bovis* bacillus is used to vaccinate against the human *Mycobacterium tuberculosis* and may serve as a suitable vector for the *B. burgdorferi* and the HIV virus. BCG differs from the virulent *M.t.* by the deletion of ~91 open reading frames and several (~38) additions. BCG has also been introduced into liver cancer cells, skin tumors and other cancer cells with some beneficial effects on slowing down metastasis and/or delayed recurrence. Protection against tuberculosis has the highest correlation with the rapid accumulation of specific $CD8^+$ T cells in the infected tissues of challenged mice. Specific IFN-production by $CD4^+$ T cells reflected the load of *M. tuberculosis* rather than the strength of protection (Mittrücker H-W et al 2007 Proc Natl Acad Sci USA 104:12434. ►*Borrelia*, ►acquired immunodeficiency, ►mycobacteria, ►metastasis, ►T cell, ►interferon; Sassetti CM et al. 2001 Proc Natl Acad Sci USA 98:12712; Hsu T et al 2003 Proc Natl Acad Sci USA 100:12420.

Bacillus cereus: This opportunistic pathogen causes diarrhea and emetic syndromes. Its 4,559,996 bp sequenced genome is closely related to that of *Bacillus anthracis* and *Bacillus thüringiensis*. (►anthrax, ►*Bacillus thüringiensis*; Ivanova N et al 2003 Nature [Lond] 423:87)

Bacillus subtilis: A gram-positive, rod-shaped soil bacterium that lives on decayed organic material and is therefore harmless. Under conditions of starvation, most of the cell content, particularly the DNA moves to one end of the cell. This area, constituting about 10% of the cell, is walled off and becomes a spore. Before spore formation a checkpoint protein (DisA) scans the chromosome for DNA damage and halts spore formation until the damage is repaired (Bejerano-Sagie M et al 2006 Cell 125:679). The spore is extremely resistant to various environmental effects that can destroy vegetative cells. Under favorable conditions the spore regenerates the bacterium. Before the cells form spores, half of the bacterial population exports a killer protein,

SdpcC. The killer protein destroys the non-producer cells, which are then cannibalized by the producer cells. The exporter cells produce an immunity protein SdpI that protects these cells from the killer protein. SdpC stimulates SdpI production. SdpI synthesis is under the control of a two-gene operon which is controlled by the SdpR repressor (Ellermeier CD et al 2006 Cell 124:549). The size of its cells is similar to those of *E. coli*, and its genome of 4,214,810 bp (4,100 ORF) was completely sequenced by 1997 (Nature 390:248). Among the 4,100 genes only 271 appeared indispensable for growth of the bacterium when the genes were inactivated singly (Kobayashi K et al 2003 Proc Natl Acad Sci USA 100:4678). The DNA has a different base composition from that in *E. coli*, A + T/G + C ratio is 1.38 in the former and 0.91 in the latter indicating that *B. subtilis* has more A + T than *E. coli*. The genome includes many repeats in only half of the chromosome, at both sides of the replicational origin. For transcription it uses 18 different σ factors although its major RNA polymerase is similar to that of *E. coli* ($\alpha\alpha\beta\beta'\sigma$). The 43 kDa ($\sigma 43$) recognizes some of the consensus sequences of *E. coli* promoters. Its best known phage is SPO1 that is transcribed either by a phage RNA polymerase or by the host. It contains 4100 protein-coding genes with an average length of 890 bp. 78% of them start with ATG. 75% of the genes are transcribed in the direction of the replication. 53% of the genes occur only once (singlets) whereas the putative ATP binding transporter family paralogues appear to be 77 (14% of the genome). The DNA is not methylated. ►*E. coli*, ►forespore, ►endospore, ►sporulation, ►paralogous, ►fratricide; Hecker M, Engelmann S 2000 Int J Med Microbiol 290:123, http://genolist.pasteur.fr/.

Bacillus thüringiensis: This gram-positive bacterium produces the BT toxin. The toxin (delta endotoxin) is within the crystalline inclusion bodies produced during sporulation. In an alkaline environment (such as in the midgut of insects) the crystals dissolve and release proteins of M_r 65000 to 160000 that are cleaved by the proteolytic enzymes of the insects into highly toxic peptides. These toxins are most effective against *Lepidopteran* larvae (caterpillars) but some nematodes are susceptible to one or another form of the toxin (Wei J-Z et al 2003 Proc Natl Acad Sci USA 100:2760; Cappello M et al 2006 Proc Natl Acad Sci USA 103:15154). The most economical solution is to transform plants (tobacco, cotton, maize etc.) with the *Bt2* gene, which codes for the 1,115 amino acid residue pro-toxin protein. Actually, a smaller polypeptide, Mr 60K, and even smaller fragments are still fully active. The transgenic plants kill the invading caterpillars within a couple of days and remain practically immune to any damage. The activity of the transgene has been further enhanced by the use of high efficiency promoters in the T-DNA constructs. In some instances, however, transgenic cotton was overpowered by bollworms. There are differences in the spectra of the bacterial toxins produced by different strains of the *bacillus* and this provides an opportunity to extend the range of toxicity to other insect species. For the production of corn rootworm resistant transgenic plants, the toxin gene of *B. thüringiensis tenebrionis* has been used. Mutation in insect aminopeptidase receptors, in a cadherin superfamily gene and β-1,3-galactosyltransferase may impart resistance to the BT toxin. Glycolipids are the receptor to the BT toxin and the absence of the special glycolipid (vertebrates) or mutation in the receptor imparts resistance (Griffitts JS et al 2005 Science 307:922). *B. thüringiensis*-induced mortality depends on the presence of enteric bacteria in the gut of the target organisms (Broderick NA et al 2006 Proc Natl Acad Sci USA 103:15196).

The resistance of pests to BT is surprisingly rare under field conditions, only the diamondback moth (*Plutella xylostella* [L]) seems to be an exception (Fox JL 2003 Nature Biotechnol 21:958). The transfer of the BT toxin gene into commercial varieties may have some deleterious effects on the Monarch butterfly and other lepidopteran larvae, however under field conditions these adverse effects may not be very serious. Resistance genes to the BT toxin are located in different chromosomes of plants and experimental evidence shows that insects cannot simultaneously overcome the two-gene hurdle unless single and two-gene resistance crops are grown at the same time. In such a situation the insect can develop immunity against one than the other gene and eventually the plant resistance may be lost. However, when only two-gene resistant plants are grown the evolution of the insects may not be possible (Zhao J-Z et al 2005 Proc Natl Acad Sci USA 102:8426). The culture of the BT toxin transgenic cotton did not affect biodiversity as much as the use of broad-spectrum insecticides and secured higher yield in two years of large scale agricultural production (Cattaneo MG et al 2006 Proc Natl Acad Sci USA 103:7571). ►transformation of plants, ►promoter, ►*Photorhabdus luminescens;* pest eradication by genetic means, ►hookworm, ►Cry9C, ►cadherins, ►insect resistance in plant, ►GMO, ►*Bacillus cereus*, ►anthrax; Gahan LJ et al 2001 Science 293:857; Griffits JS et al 2001 Science 293:860; series of articles in Proc Natl Acad Sci USA 98:11908–11937 [2001]; membrane receptor: Pérez C et al 2005 Proc Natl Acad Sci USA 102:18303.

B

Back Cross: The F1 is crossed (mated) by either of its two parents (►test cross). Each back crossing reduces by 50% the genetic contribution of the non-recurrent parent; thus after (r) back crosses it will be (0.5)r. The percentage of individuals homozygous for the (n) loci of the recurrent parent in (r) number of back crosses = $[(2^r - 1)/2^r]^n$. The chance of eliminating a gene linked to a selected allele is determined by the intensity of linkage (p) and the number of back crosses (r) according to the formula $1 - (1 - p)^{r+1}$.

Back Mutation: This mutation (recessive) reverts to the wild type allele. ►reversion

Back Reaction: Refers to a property of RNA polymerase to move backward and cleave the synthesized RNA if a nucleotide needed for the forward (synthetic) reaction is not available. ►dead end complex

Backbone: An example is the sugar-phosphate chain of nucleic acids or the N–C chain of amino acids in a protein. To the backbone, side chains may be attached such as nucleotides or some other molecular groups in amino acids, except in glycine, which has no side chain. ►Watson and Crick model

Background, Genetic: Refers to the (residual) genetic constitution without considering particular loci or genes under special study. Knowledge of the genetic background may be of substantial importance because different sets of modifier genes may influence the expression of particular genes. ►modifier genes

Background Radiation: This is the natural radiation from cosmic or terrestrial sources. ►cosmic radiation, ►terrestrial radiation

Background Selection: The recurrence of deleterious mutations reduces the effective size of the population. The selection is directed against the chromosomal background carrying the particular allele(s). The balance between hitchhiking and background selection determines the extent of genetic variation in a population. ►hitchhiking, ►effective population size; Charlesworth D et al 1995 Genetics 141:1619.

Backtracking, Transcription: In this process the RNA polymerase (the elongation complex) slides backward on the DNA template by one or more nucleotides. In this case the RNAP may lose the 3′-end of the transcript and the complex may be temporarily inactivated thereby interrupting transcript elongation. Reactivation may require (prokaryote) GreA, GreB and TFIIS (eukaryote) and other protein factors (Mfd). Reinitiation may be facilitated by the use of multiple RNAP molecules. (►Gre, ►Mfd, ►TFIIS, ►reinitiation, ►Epshtein V; Nudler E 2003 Science 300:801).

Bactenein: antimicrobial peptides.

Bacteremia: Refers to bacterial infection of the blood.

Bacteria: This broad taxonomic group of microscopically visible (prokaryotic) organisms has DNA as the genetic material (nucleoid) that is not enclosed by a distinct membrane within the cell and may have various numbers of extrachromosomal elements, plasmids that constitute from about 2 to 20% of the DNA per cell. The size of the bacteria varies considerably from 0.2 to 750 µm. Bacteria are capable of protein synthesis and of independent metabolism even if they are parasitic or saprophytic. Their cell wall is mucopolysaccharide and protein. Their cells contain ribosomes (70S) but no mitochondria, plastids, endoplasmic reticulum or other compartments. They divide by fission in an exponential manner as long as nutrients and air are not in limited supply. The division rate of bacteria during exponential growth can be expressed as $N = 2^g \times N_0$ where N is the number of cells after g generation of growth and N_0 is the initial cell number. After the exponential phase, unless their increasing need is met, the growth either declines or may become stationary. The generation time of *E. coli* under standard conditions may be 20–25 minutes.

Bacteria can be classified into three main groups: *Archebacteriales*, *Eubacteriales* and *Actinomycetales*. *Eubacteriales* includes *Pseudomonadaceae* (*Pseudomonas aeruginosa*), *Azotobacteriaceae* (*Azotobacter vinelandii*), *Rhizobiaceae* (*Rhizobia, Agrobacteria*), *Micrococcaceae* (*Micrococcus pyogenes*), *Parvobacteriaceae* (*Haemophilus influenzae*), *Lactobacteriaceae* (*Diplococcus pneumoniae*, *Streptococcus faecalis*), *Enterobacteriaceae*, (*Aerobacter aerogenes, Escherichia coli, Salmonella typhimurium)*, *Bacilliaceae* (*Bacillus subtilis, B. thüringiensis*). *Actinomycetales* includes *Mycobacteriaceae* (*Mycobacterium phlei*, *Mycobacterium tuberculosis*) and *Streptomycetaceae* (*Streptomyces coelicolor, S. griseus*). There are many other types of classification systems. It is common to identify bacteria as gram-positive (indicating that they retain the deep red color of the gram stain [crystal-violet and iodine] after treatment with ethanol) or gram-negative that fail to retain it (and may appear colorless or just slightly pinkish). These properties depend on the composition and structure of the cell wall. Gram-positive bacteria are surrounded by peptidoglycan outside of the plasma membrane, and gram-negative cells have an outer membrane enveloping the peptidoglycan wall. The peptidoglycans are polymers of sugars and peptides and are cross-linked by pentaglycines that determine the shape of the cell wall and the bacterium. There are at least 10^{30} bacteria on the planet and at least as many bacteriophages. Probably less than 1% of the bacteria can be cultured in the laboratory and that makes their study

difficult (Amann RI et al 1995 Microbiol Rev 59:143). Closely related bacteria can be distinguished—even if the difference between two 16S RNAs is in only one nucleotide—by the use of quenched autoligation probes. ►conjugation, ►bacterial recombination frequency, ►bacterial transformation, ►recombination molecular mechanisms in prokaryotes, ►bacteria counting, ►quenched autoligation probe; bacterial names and standing nomenclature: http://www.bacterio.net, chromosome maps of several bacteria: http://wishart.biology.ualberta.ca/BacMap/.

Bacteria Counting: This is done either by counting the number of colonies formed or by determining cell density in a volume, using a photometer. In the first procedure an inoculum of a great dilution of a culture is seeded on a nutrient agar plate and incubated for a period of time (e.g., 2 days). Each colony thus formed represents the progeny of a single cell and the number of colonies indicates the number of *live* bacteria in the volume of the inoculum. Optical density obtained through the second procedure indicates the cell density that becomes meaningful only if information is available on the correlation between light absorption and cell number, determined earlier by the plating technique. If the plate was seeded by 2 mL of the culture diluted 10^7 times and 100 colonies are observed then the number of live cells is $(100/2) \times 10^7 = 5 \times 10^8$ cells/mL. ►bacteria, ►lawn bacterial

Bacterial Artificial Chromosome: ►BAC

Bacterial Recombination Frequency: In the case of conjugation transfer of genes recombination frequency is determined by the time in minutes since the beginning of mating. This procedure is useful for genes that are more than 2 to 3 minutes apart. It takes about 90–100 minutes at 37 °C to transfer from a Hfr donor bacterium to an F− recipient cell the entire genome (more than 4 million nucleotides). The efficiency of transfer depends on the nature of the Hfr strain used. Approximately $5{-}6 \times 10^4$ nucleotides are transferred per minute. Bacterial recombination does not permit the recovery of the reciprocal products of recombination and all detected crossover products are double cross-overs. If bacterial genes are closer than 2 to 3 minutes, then recombination mapping is used. For bacterial recombination, selectable (auxotrophic) markers are generally used so that the phenotypes can be easily recognized. The recipient strain carries genes, e.g., a and b^+, and the donor strain carries the alleles a^+ and b defining the interval where recombination is studied. In order to measure the number of successful matings, the donor strain also carries the prototrophic gene (c^+) and the recipient is marked by the auxotrophy allele (c) of the same locus. The c gene does not have to be very close to the interval studied:

$$\frac{a \quad b^+}{a^+ \quad b} \qquad \frac{c^+}{c}$$

The *frequency of recombination* (p) is than calculated:

$$p = \frac{\text{number of cells } a^+ b^+ \text{ constitution}}{\text{number of } c^+ \text{ cells}}$$

The a^+b^+ recombinants are the result of an exchange between a^+ and b^+ and also beyond the c^+ site as shown by the arrows: $a^+ \uparrow b^+ c^+ \downarrow$.

To determine *gene order by recombination* one must use at least three loci in a reciprocal manner (see Tables B1and B2):
Would the gene order be *a b c*:

Table B1. Gene order determination by recombination in bacteria

Donor	$a \downarrow b^+c^+\downarrow$	To obtain triple prototrophs **double** exchange is sufficient in both of the reciprocal crosses
Recipient	$a^+b\ c$	
Donor	$\downarrow a^+\downarrow b\ c$	
Recipient	$a\ b^+c^+$	

Would the gene sequence be *b a c*:

Donor	$\downarrow b^+\downarrow a\ c^+\downarrow$	In order to obtain triple prototroph recipients, the number of exchanges must be at least **quadruple** as shown by the arrows
Recipient	$b\ a^+\uparrow c$	
Donor	$b \downarrow a^+\downarrow c$	In the reciprocal cross only **double** recombination is required to produce $b^+a^+c^+$ prototrophs
Recipient	$b^+a\ c^+$	

Thus, depending whether the gene order is *abc* or *bac* one can tell from the frequency of prototrophs in the reciprocal crosses. The higher numbers of exchanges are less frequent.

Recombination frequency in bacteria within very short intervals, such as between alleles within a gene can also be determined by transduction. If the constitution of the donor DNA is $a^+\ b^+$ and the recipient is $a\ b$, *the frequency of transduction* (recombination) is:

$$\frac{[a^+b] + [ab^+]}{[a^+b] + [ab^+] + [a^+b^+]}$$

Gene order in bacteria can be determined by a three-point transformation test as illustrated by a hypothetical experiment described here when the donor DNA is

$a^+ b^+ d^+$ and the recipient is $a^- b^- d^-$, and the reciprocal products of recombination are not recovered:

Table B2. Gene order determination by transformation in bacteria

Genes	Genotypes of Transformants						
a	+	−	−	−	+	+	+
b	+	+	−	+	−	−	+
d	+	+	+	−	−	+	−
Number of cells	12000	3400	700	400	2500	100	1200

Recombination for a particular interval is calculated by the number of recombinants in the interval(s) divided by the total number of cells transformed in that interval. There are 7 classes of cells. Recombination is calculated in three steps: in the *ab*, *bc* and *ad* intervals.

In the ***ab*** interval we have here

$$\frac{[3400 + 400 + 2500 + 100]}{[1200 + 3400 + 400 + 2500 + 100 + 1200]} \approx 0.33$$

in the ***bd*** interval

$$\frac{[700 + 400 + 100 + 1200]}{[12000 + 3400 + 700 + 400 + 100 + 1200]} \approx 0.14$$

in the ***ad*** region

$$\frac{[3400 + 700 + 400 + 2500 + 100 + 1200]}{[12000 + 3400 + 700 + 400 + 2500 + 100 + 1200]} \approx 0.41$$

Although the frequency of recombination in three-point transformation test is never exactly additive, it is clear that the *a*-*d* distance is the longest and thus the conclusion that the gene order is *abd* appears to be reasonable. Recombination may affect the population structure and it permits evolutionary conclusion. ►physical mapping, ►crossing over, ►mapping genetic, ►conjugation, ►transduction, ►bacterial transformation, ►generalized transduction; Lederberg J 1987 Annu Rev Genet 21:23; Feil EJ, Spratt BG 2001 Annu Rev Microbiol 55:561.

Bacterial Transformation: Refers to genetic alteration brought about by the uptake and integration of exogenous DNA in the cell that is capable of expression. The exogenous DNA is generally supplied at a concentration of 5 to 10 μg/mL to transformation competent cells. Competence is a physiological state when the cells are ready to accept and integrate the exogenous DNA. Competence is maximal in the middle of the logarithmic growth phase. The donor DNA may synapse with the recipient bacterial chromosome and naked DNA generally replaces a segment of the bacterial genetic material rather than adding to it. The entire length of the donor DNA may not be integrated into the host and the superfluous material is degraded. The integrated DNA may form a permanent part of the bacterium's chromosomal genetic material. During integration only one or both strands of the donor DNA may be integrated. Transformation may be regarded as one of the mechanisms of recombination and can be used for determining the gene order in the bacterial chromosome. The frequency of transformation in prokaryotes is generally less than 1% and it is usually within the range of 10^{-3} to 10^{-5}. Transformation of bacterial protoplasts (spheroplasts) may occur at a higher frequency. Transformation may also denote the transfer and expression of plasmid DNA in the cell. These plasmids may remain as autonomous elements within the bacteria. Transformation with the aid of plasmids is much more efficient. Moreover, competence can greatly be enhanced by some divalent cations and by other means. ►transformation genetic, ►competence of bacteria, ►bacterial recombination frequency; vectors, Hotchkiss RD, Gabor M 1970 Annu Rev Genet 4:193; Oishi M, Cosloy SD 1972 Biochem Biophys Res Commun 49:1568.

Bacteriocin: Denotes natural bacterial products that may kill sensitive bacteria. ►colicins, ►pyocin, ►pesticin; Riley MA 1998 Annu Rev Genet 32:255; bacteriocin identifying tool: http://bioinformatics.biol.rug.nl/websoftware/bagel/bagel_start.php.

Bacteriocyte: Refers to special cells of eukaryotes harboring bacterial symbionts. (Spaulding AW, Dohle CD 1998 Mol Biol Evol 15:1506; Nakabachi A et al 2005 Proc Natl Acad Sci USA 102:5477).

Bacteriome: This is a cytoplasmic polyploid organ of insects harboring one or more species of bacteria. The exact function of the bacteriome is unknown; it probably synthesizes useful nutrients for the host. (Von Dohlen CD et al 2001 Nature [Lond] 412:433; Normark BB 2004 PloS Biol 2:0298).

Bacteriophages: These are viruses infecting bacteria (see Table B3). (►phage, ►phage life cycle, ►phage morphogenesis, ►filamentous phages, ►lambda phage, ►T4, ►T7, ►φX174, ►MS2, ►Mu bacteriophage, ►icosahedral, ►virulence, ►temperate phage, ►development, ►phage therapy; Knipe DM, Howley PM (eds) 2001 Fundamental Virology Lippincott Williams & Wilkins, Philadelphia, PA); Brüssow H, Hendrix RW 2002 Cell 108:13; Campbell A 2003 Nature Rev Genet 4:471, http://www.phage.org.

Table B3 Major types of bacteriophages

Phage	Type	Host	Da × 10^6	Morphology
MS2, f2, R17	RNA, ss, virulent	*E. coli*	1	icosahedral
φ6	RNA, ds, virulent	*Pseudomonas*	3.3, 4.6, 7.5	icosahedral
φX174, G4, St-1	DNA, ss, virulent	*E. coli*	1.8	icosahedral-tail
M13, fd, f1	DNA, ss, virulent	*E. coli*	2.1	filamentous
P22	DNA, ds, temperate	*Salmonella*	26	icosahedral-tail
SPO1	DNA, ds, virulent	*Bacillus subtilis*	91	icosahedral-tail
T7	DNA, ds, virulent	*E. coli*	26	octahedral-tail
lambda	DNA, ds, temperate	*E. coli*	31	icosahedral-tail
P1, P7	DNA, ds, temperate	*E. coli*	59	head-tail
T5	DNA, ds, virulent	*E. coli*	75	octahedral-tail
T2, T4, T6	DNA, ds, virulent	*E. coli*	108	oblong head-tail

ss = single-stranded, ds = double-stranded

The major types and characteristics of bacteriophages are presented here:

Bacterioplankton: This includes prokaryotes and plays a major role in biogeochemical processes in seawater. *Silicibacter pomeroyi* represents 10–20% of bacterioplankton; it has a chromosome of 4109611 bp and a megaplasmid of 491611 bp in its sequenced genome (Moran MA et al 2004 Nature [Lond] 432:910).

Bacteriorhodopsin: This is a light receptor protein in the plasma membrane of some bacteria; it pumps protons upon illumination. ►rhodopsin

Bacteriostasis: This prevents the reproduction of bacteria without destroying them. In the long run, however, this may lead to their destruction. Many antibiotics have such an effect. ►antibiotics

Bacteroid: These are specialized, modified forms of bacteria such as the ones found in the root nodules where they act in the fashion of intracellular "organelles" in nitrogen fixation. ►nitrogen fixation; Li Y et al 2002 Microbiology 148:1959.

Bacteroides fragilis: This is an obligate anaerobic, opportunistic pathogen of the human colon. Its circular chromosome is 5,205,140 base pairs with an estimated 4,274 genes; it harbors a plasmid too. Its genome, like that of *B. thetaiotaomicron*, has many inversions and rearrangements, which affect gene expression. Several species of bacteroides are part of the indigenous intestinal flora and may be represented by 10^{11} to 10^{12} cells/g feces. They constitute 15–20% of gingival flora and 8–16% of dental plaques. It is somewhat of a paradox that the host does not marshal the immune defense against them. These organisms apparently decorate their surface polysaccharides with L-fucose and mimicking host polysaccharides thus acquiring an evasive tool in their competitive environment (Coyne MJ et al 2005 Science 307:1778). The pathogenic bacteria apparently do not have this self-defense against the host. (►fucose; Cardeño-Tárraga AM et al 2005 Science 307:1463).

Bacteroides thetaiotaomicron: This is a gram-negative anaerobic bacterium with a 6.26 Mb sequenced genome. It is a predominant member of the human and mouse small intestinal and colon microbiome. It plays an important role in the metabolism of dietary polysaccharides, which are not digestible by human enzymes. (►microbiome; Kuwahara T et al 2004 Proc Natl Acad Sci USA 101:14919).

Bactigs: Contigs of BACs. (►contig, ►BAC)

Bacto Yeast Extract: This water-soluble fraction of autolyzed yeast contains vitamin B complex.

Bacto-Tryptone: A peptone, rich in indole (tryptophan), is used for bacterial cultures and the classification of bacteria on the basis of activity.

Baculoviruses: These are large (130 kbp) double-strand DNA viruses used for the construction of insect transformation vectors. Baculoviruses do not efficiently transform mammalian or plant cells. The baculovirus vectors accommodate large amounts of DNA and the foreign DNA replacing the polyhedrin gene is expressed under a powerful polyhedrin promoter. The majority of the proteins within the insect remain soluble. The extracellularly present virus particles appearing *late* in the infection are called

non-occluded virus. The occluded virus particles occur in the cell nuclei and appear *very late* in the infection phase. The polyhedra viral protein coating is responsible for the occlusion. (►polyhedrosis, ►AcNBPV, ►transformation, ►viral vectors; insect viruses, Grabherr R et al 2001 Trends Biotechnol 19[6]:231; Herniou EA et al 2003 Annu Rev Entomol 48:211).

Baculum: This is a bony structural element above the urethra in the penis of many species (e.g., rodents, carnivores, primates) but not in humans.

Bad: Refers to an apoptosis-promoting protein when phosphorylated by MAPK/RSK or Akt or a c-AMP-dependent protein kinase. When it is dephosphorylated (by calcineurin) it may interfere with the apoptosis suppression of Bc*l* proteins and that may lead to carcinogenesis. Also, when RSK phosphorylates CREB, cell survival is facilitated. The combined inhibition of EGFR (epidermal growth factor receptor) and PI3K (phosphatidylinositol kinase)—regulated by BAD—synergistically favors apoptosis (She Q-B et al 2005 Cancer Cell 8:287). ►apoptosis, ►survivin, ►CaM-KK, ►Bc*l*, ►MAPK, ►RSK, ►CREB, ►Akt; Konishi Y et al 2002 Mol Cell 9:1005; Ranger AM et al 2003 Proc Natl Acad Sci USA 100:19324.

Badger (*Taxidea taxus*): 2n = 32.

Badnaviruses: These are double-stranded DNA viruses of plants (►pararetrovirus).

Baf Complex: This is similar to SWI/SNF proteins and it regulates chromatin remodeling. The Baf60c subunit (60-kDa, encoded by mouse gene *Smarcd3*) is essential specifically for the expression of the differentiation of the heart and somites of the early embryo (Lickert H et al 2004 Nature [Lond] 432:107). ►SWI/SNF; chromatin remodeling, Liu R et al 2001 Cell 106:309.

BAFF (B cell activating factor): TNF receptor ligand, which among other proteins regulates B cell proliferation and differentiation. Autoreactive B cell survival is regulated by BAFF-dependent protein kinase, PKCδ, which phosphorylates serine 14 in histone H2B (Mecklenbräuker I et al 2004 Nature [Lond] 431:456). ►Blys, ►TNFR, ►APRIL, ►NF-κB, Thompson JS et al 2001 Science 293:2108; Schiemann B et al 2001 Science 293:2111.

BAG1: (Bcl2-associated athanogene, 9p12): This is part of an anti-apoptotic complex and affects cell division, cell migration and differentiation. The BAG family proteins recruit molecular chaperones and thus play a role in regulating protein conformation. ►BCL, ►athanogene; Takayama S, Reed JC 2001 Nature Cell Biol 3:E237.

BAIT: ►Two-hybrid system

BAK: A member of the Bc*l* protein family comprising BH1, BH2 and BH3 domains that after a conformational change induced by other members of the Bc*l* family of proteins containing only BH3 domain, promotes apoptosis by opening the permeability transition pore complex channel in the mitochondrial membrane. The mitochondrial outer-membrane protein VDAC2 inhibits BAK and apoptosis. ►Bc*l*, ►BID, ►Bim, ►apoptosis, ►porin; Wei MC et al 2000 Genes Dev 14:2060; Korsmeyer SJ et al 2000 Cell Death Differ 7:1166; Cheng EH et al 2001 Mol Cell 8:705; Cheng E H-Y et al 2003 Science 301:513.

Bal 31: The exonuclease removes simultaneously nucleotides from the 3′ as well the 5′ ends and thus it can be used for mapping functional sites in a DNA (see Table B4): The fragments can then be separated by electrophoresis and assayed after transformation. ►deletions unidirectional, ►exonuclease electrophoresis; Wei C-F et al 1983 J Biol Chem 258:13506.

Table B4. Bal 31 exonuclease

0 Time	*a b c d e*	Original DNA
After 1 time unit	*b c d*	Digest
After 2 time units	*c*	Digest

Balance of Alleles: This population model assumes that at the majority of loci several different alleles are present and these are maintained in a dynamic equilibrium by the continuous but variable selective forces. ►balanced polymorphism, ►Hardy-Weinberg theorem, ►fitness, ►selection

Balanced Lethals: These are genetic stocks heterozygous for two or more non-allelic linked recessive lethal genes. Since both homozygotes die, only heterozygotes survive that are phenotypically wild type or in some cases exhibit mutant phenotype and continue to produce both types of lethals. Such stocks can be maintained indefinitely as long as recombination between the linked loci can be prevented.

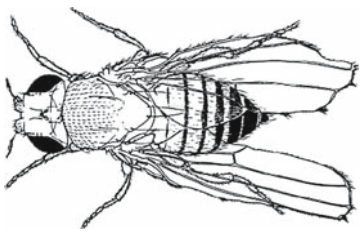

Figure B7. *Beaded* (From Bridges CB, Morgan TH 1923 Carnegie Inst Wash 327:152)

For balancing, generally spanning inversions are used that eliminate the cross-over gametes because of the duplications and deficiencies generated by recombination within the inverted region. The first balanced lethal of *Drosophila* contained the gene *Bd* 1 (*Beaded*, incised wing in heterozygotes, lethal in

homozygotes, at map location 3–91.9 [slightly different in some other alleles]) and *l(3)a*, a spontaneous lethal mutation (map position 3–81.6) within the inversion *In(3R)C* (see Fig. B7). In the multiple translocations of plants of different *Oenothera* species, there are gametic and zygotic recessive lethal genes that are prevented from becoming homozygous and thus help to maintain these lethal genes by balanced heterozygosity. Besides the biological advantage of balanced lethals, they may be useful for various types of research. The *Bd* alleles have been extensively studied at the molecular level and the developmental functions may be revealed by the availability of heterozygotes for the mutations. ►lethal factors, ►lethal equivalent, ►translocation ring; Muller HJ 1918 Genetics 3:422.

Balanced Polymorphism: When the fitness (reproductive success) of heterozygotes exceeds both homozygotes at a locus, a stable genetic equilibrium may be established and the heterozygotes may reproduce the homozygotes in equal frequencies. This type of heterozygote advantage may lead to balanced polymorphism, i.e., the population may maintain several genotypes in stable proportions even if some of the homozygotes have low adaptive value. ►selection coefficient, ►fitness, ►balanced lethals, ►autosomal recessive lethal assay, ►balance of alleles, ►Hardy-Weinberg theorem, ►fitness, ►selection, ►Muller' ratchet; Rucknagel DL, Neel JV 1961 Progr Med Genet 1:158.

Balanced Translocation: Refers to a reciprocal translocation where each of the interchanged chromosomes has a centromere. Unbalanced translocations have an acentric piece due to the interchange. ►translocation chromosomal

Balancer Chromosomes: These are structurally modified (by inversions, translocation) so the recombinants (because of duplications or deficiencies in the meiotic products) are not recovered in the progeny, and facilitate the maintenance of certain chromosomal constitutions without recombination. Balanced systems permit the maintenance of recessive lethal factors in a heterozygous condition. Balancer chromosomes have been developed in the past with the use of clastogenic agents. By inserting the *LoxP* gene in opposite orientations and bringing about recombination with the aid of the *Cre* recombinase, inversions can also be generated in e.g., mouse cells at particular intervals. ►*ClB* method, ►*Basc*, ►inversion, ►translocation chromosomal, ►Renner complex, ►balanced lethals, ►autosomal recessive lethal assay, ►*Oenothera, Cre/Lox*, ►targeting genes; Muller HJ 1918 Genetics 3:422; Yu Y, Bradley A 2001 Nature Rev Genet 2:780.

Balancing Selection: This includes heterozygote advantage (overdominance), or alleles differently selected by sex, season and niche in the habitat or in a frequency-dependent manner. ►selection, ►sexual selection, ►overdominance, frequency dependent selection; Verelli BC et al 2002 Am J Hum Genet 71:1112.

BALB/c Mice: An albino inbred laboratory strain used frequently in immunoglobulin (antibody) and cancer research (see Fig. B8). It is highly susceptible to *Salmonella*. ►mouse

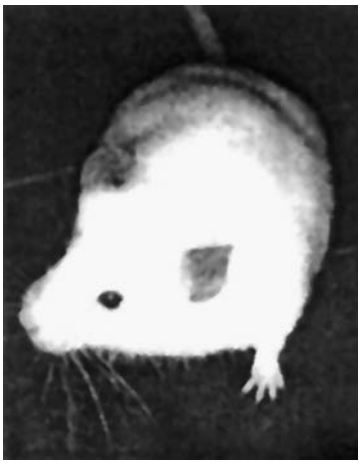

Figure B8. Albino Mouse (Courtesy of Dr. Paul Szauter, http://www.informatics.jax.org/mgihome/other/citation.shtml)

Balbiani Body: This is a large distinctive organelle aggregate found in developing oocytes of many species; it contains a mitochondrial "cloud" and Golgi bodies.

Balbiani Ring: Refers to a puff (bloated segment) of the polytenic chromosome indicating special activity (intense RNA transcription) at the site, it loosens up the multiple elements of the chromosome (see Fig. B9). ►polytenic chromosomes, ►puff, ►BR RNP

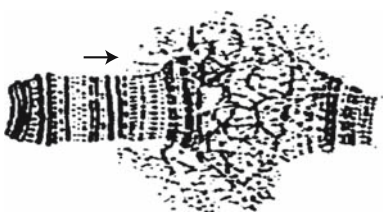

Figure B9. Balbiani ring

Baldness: This sex-influenced trait in humans is more common among males than females (particularly with later onset) and it probably depends to some extent on the level of androgen receptor.

Apparently a polyglycine-encoding GGN repeat in exon 1 of the gene in X chromosome causes

baldness (see Fig. B10). This also explains the fact that inheritance is primarily through the maternal line (Hillmer AM et al 2005 Am J Hum Genet 77:140). In this condition a developmentally manifested pattern is seen starting from the front hairline toward the top of the head. It has a strong hereditary component but it may be caused by certain diseases (alopecia), exposure to higher doses of ionizing radiation and to certain carcinostatic drugs. Early baldness may be determined by an autosomal dominant gene (10q24) encoding steroid 17α-hydroxylase with better penetrance in males than in females. Loss of hair on the human scalp may be controlled by a deletion in chromosome 3q27, the site of (LIPH) phospholipase (Kazantseva A et al 2006 Science 314:982). ►alopecia, ►hair, ►androgen, ►penetrance, ►monilethrix

Figure B10. X-chromosomal baldness

Baldwin Effect: The physiological homeostasis permits the survival of a species until mutation may genetically fix the adaptive trait in an originally inhospitable environment. ►homeostasis, ►canalization, ►genetic assimilation; West-Eberhard MJ 2003 Developmental Plasticity and Evolution, Oxford University Press, New York.

Ball-And-Stick Model (ball-and-spoke model): ►stick-and-ball model

Ball-In-Urn: This is used to characterize the distribution of repeats in the genome; urns correspond to all DNA words of a given size and balls refer to the observed words in a given DNA sequence. Rare words may be the binding sites of transcription factors. Also, they may be discriminated against because of structural incompatibilities. Frequent words may be repetitive, structural and regulatory sequences as well as transposable elements. (Karlin S 2005 Proc Natl Acad Sci USA 102:13355).

BamH1: Refers to restriction enzyme with recognition sequence G↓GATCC.

BAMBE (Bayesian Analysis in Molecular Biology and Evolution): This is a free software for the analysis of phylogeny based on nucleotide sequences. ►Bayes theorem, ►phylogeny; http://www.mathcs.duq.edu/larget/bambe.html.

β-Amyloid: This exists as extracellular deposits of the brain plaques in Alzheimer's disease (see Table B5). It is split off the amyloid precursor protein (APP) by secretase. In the neuronal tissue APP_{695} is prevalent. If Ile, Phe or Gly replaces Val642, the substitutions lead to fragmentation of nucleosomal DNA in the neurons and presumably contribute to neurotoxicity. ►Alzheimer's disease, ►scrapie, ►amyloidosis, ►amino acid symbols in protein sequences, ►secretase, ►prion

Table B5. Amyloid fibers in Alzheimer disease

The major amyloid fibers (Aβ1-42) in Alzheimer disease are truncated at the C terminus:
Aβ1-40: DAEFRHDSGYEVHHQLVFFAEDVGSNK-GAIIGLMVGGVV
Aβ1-42:DAEFRHDSGYEVHHQLVFFAEDVGSNK-GAIIGLMVGGVVIV
Aggregation of the fibers may lead to plaque formation seen in amyloidosis. The aggregation may be initiated by "seeding" like a crystallization process.

Banana (*Musa acuminata*, x = 11): This is a fruit plant. The diploid fruits are full of seeds and have minimal edible pulp. The majority of the edible fruits are harvested from seedless triploid plants. When the triploids are crossed with diploids the progeny is partly tetraploid (2n = 44) and heptaploid (2n = 77) because of the high frequency of unreduced 3x and 6x gametes and their fruits are also seedless. Some of the related species have chromosome numbers 2n = 14, 18 and 20. ►seedless fruits, ►triploidy, ►sugar beet; Simmonds NW 1966 Bananas, Longman, London.

Band: Refers to an element of the cross-striped chromosome. The banding (perpendicular to the length of the salivary gland chromosomes and continuous across the giant chromosome) may be due to condensation of the juxtapositioned chromomeres or to specific staining of the chromatin. ►bands of polytenic chromosomes, ►chromosome banding. The average DNA content in a single natural band of the *Drosophila melanogaster* salivary gland chromosome is 26.2 kb. The total number of salivary chromosome bands is 5,072. The number of distinguishable bands depends on the stage of condensation of the chromosome. The cytologically detectable chromomeres/bands are greater at pachytene than at the metaphase, and in the extended condition structural abnormalities are easier to identify microscopically.

Electrophoretic separation of restriction enzyme digested DNA, or pulsed field electrophoresis separated small chromosomes, as well as various proteins subjected to separation in the electric field, generate bands in the substrate (gel) when visualized either by staining or by special illumination. ►chromosome banding, ►coefficient of crossing over, ►electrophoresis, ►pulsed field electrophoresis, ►FISH

Band Cloning: Denotes amplifying DNA bands, extracted from electrophoretic gels, in genetic cloning vectors for molecular analyses. ►cloning vectors

Band-Morph Mutation: This is distinguished by electrophoretic analysis of the proteins. Mutations resulting in amino acid replacement of different charge appear as mobility difference in the electric field. Although studies of this type were very popular during the 1960s and 1970s, they have very poor resolution because they can detect only 1/4 or less of the mutations. The advantage of this type of research was that large populations could be screened for mutations that would not have necessarily other phenotypic effect. ►electrophoresis, ►band; Harada K et al 1993 Jpn J Genet 68:605.

Band III Protein: This is a transmembrane protein consisting of about 800 amino acids. ►spectrin; Low PS et al 2001 Blood Cells Mol Dis 27:81.

Band-Sharing Coefficient (S_{xy}): This denotes the proportion of shared DNA fragments separated by electrophoresis; $S_{xy} = (2n_{xy})/(n_x + n_y)$ where n_x and n_y are the number of bands in *x* and *y* samples and n_{xy} is the number of shared bands. This coefficient may be used to determine the genetic composition of populations on the basis of DNA. In multi-locus forensic tests the British legal system previously used the formula $(0.26)^k$ for calculating the match probabilities of alleles of at least 4 kb in length. 0.26 is an empirical constant and *k* is the average number of matching alleles. However, this latter formula lacks sufficient robustness. The single locus probes (SLP) based on the profiles of 6–8 short tandem repeat loci (STR) is more popular today. This procedure is useful with as low as 100 pg DNA samples when amplified by PCR. ►DNA fingerprinting; Zhu J et al 1996 Poultry Sci 75:25.

Band Shifting: ►gel retardation assay

Banding Pattern: Refers to the distribution of chromosome bands reflecting genetic differences or differences in the expression of genes displaying more or less loose puffs. ►polytenic chromosomes, ►lampbrush chromosomes, ►chromosome banding, ►puff

Bands of Polytenic Chromosomes: These are deeply stained prominent cross bands on the chromosomes where the chromomeres of the elementary strands are appositioned (see Fig. B11). The salivary chromosomes of *Drosophila* display about 5,000 bands and for a period of time it was assumed that each corresponds to a gene locus. It is now known that the number of genes is about 2.5 times the number of bands. In the region 2B of the X chromosome of *Drosophila* the bands may appear different and rather than being perpendicular to the axis, they may be roughly parallel to the axis. In situ hybridization with molecular probes suggests that this unusual structure is caused by inverted repeats in the DNA. ►polyteny, ►band, ►salivary gland chromosomes, ►coefficient of crossing over for the tip of the X chromosome

Figure B11. Bands, polytenic

BankIt: This is a GenBank submission form for protein coding sequences. It then generates a GenBank accession number. Its address is: http://www.ncbi.nlm.nih.gov/BankIt/. ►GenBank, ►Sequin

Bannayan-Riley-Ruvalcaba Syndrome: This is similar to Bannayan-Zonona Syndrome.

Bannayan-Zonona Syndrome: This condition is characterized by autosomal dominant macrocephaly with multiple lipomas and hemangiomas, as well as susceptibility to hamartomatous polyposis cancer. Haplo-insufficiency of PTEN may play a role in its manifestation. ►PTEN, ►lipomatosis, ►hemangioma, ►multiple hamartomas

BAP: Refers to 6-benzylaminopurine which is a plant hormone. ►plant hormones

BAP (B-cell receptor associated proteins): Denote three (32, 37, 41 kDa) proteins associated only with IgM membranes. The 32 and the similar 37-kDa molecules form heterodimers and seem to be inhibitors of cell division. BAP29 (240 amino acid) and BAP31 (245 amino acids) are 43% homologous and bind mainly to IgD and somewhat to IgM. ►B lymphocyte receptor

BAPG: ►bullous pemphigoid autoimmune disease

Bar: This is a regulator of the Fas- and Apaf-mediated pathways of apoptosis. ►apoptosis

***Bar* Mutation**: The mutation of *Drosophila* (*B*, map position 1–57.0) reduces the eye to a vertical bar with about 90 facets in the male and around 70 in the female compared with 740 in normal males and 780 in normal females; heterozygous females have 360. The *B* mutation is actually a tandem duplication of salivary band 16A, which arises because of unequal (oblique) crossing over.

Thus, the "normal allele" has 16A, *Bar* 16A-16A, *Ultrabar* 16A-16A-16A constitution (see Fig. B12).

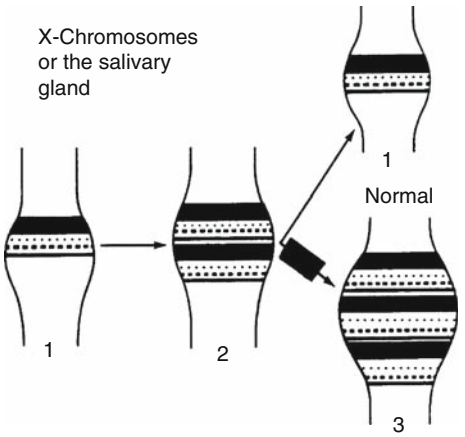

Figure B12. *Bar* mutation

The phenotype is actually a position effect and not the cause of a dosage effect as revealed by genetic analyses. The process of unequal crossing over may be repeated and as many as 9 copies of band 16A can accumulate in a single X chromosome. Also, the 16A band may be lost resulting in reversion by the loss of the *roo* transposable element. *B* mutations may also be induced by the P hybrid dysgenesis element whereas chemical mutagens have never produced this mutation. These facts indicate that the *breakage points in the duplications cause the Bar phenotype*. The *Bar* phenotype may be the result of a breakage in a regulatory element or within an intron and causes abnormal fusion of the exons. The *Bar* locus is very large, it spans at least 37 kb DNA. ►duplication, ►position effect, ►unequal crossing over, ►intron, ►exon, ►*ClB*; Bridges, Sturtevant AH 1925 Genetics 10:117; Bridges CB 1936 Science 83:210.

Bar-Code, Genetic (molecular): Bar-code generally represents vertical bars of varying widths (two or four) that correspond to digits 0 and 1, and which in turn specify numbers 0 to 9. An optical laser can read the bar-coded information and—through the computer—the scanner can identify various types of information, including properties of a gene, phenotypic expression, etc. Molecular bar-codes can be generated by two ~20-base (20-mer) oligonucleotides (UPTAG, DOWNTAG) introduced by transformation into special (deletion) cells. These molecular bar-codes can be identified in the genome by microarray hybridization or by sequencing. Also, the growth rate of various deletion strains can be monitored. DNA bar-coding can be used for the identification of species. In animals, mitochondrial cytochrome oxidase I is useful for the identification of species (Hebert PD et al 2004 PLoS Biol, 2:e312; Hajibabaei M et al 2006 Proc Natl Acad Sci USA 103:968). In plants, an approximately 450-bp intergenic spacer in chloroplasts appeared to be discriminatory (Kress WJ et al 2005 Proc Natl Acad Sci USA 102:8369). ►DNA chips, ►targeting genes, ►signature-tagged mutagenesis; Gad S et al 2001 Genes Chromosomes Cancer 31:75; Eason RG et al 2004 Proc Natl Acad Sci USA 101:11046; identification of various organisms and for taxonomy: http://www.barcodinglife.org; http://www.barcoding.si.edu.

Bar-Code DNA Isolation Method: ►bar-code genetic, ►nanoparticles

BARD1 (BRCA1-associated Ring domain protein): This inhibits polyadenylation of mRNA in cooperation with Cstf-50 (cleavage stimulation factor). BRCA1/BARD1 heterodimer modulates Ran-dependent assembly of the mitotic spindle (Joukov V et al 2006 Cell 127:539). ►cleavage stimulation factor, ►breast cancer, ►spindle, ►RAN; Joukov V et al 2001 Proc Natl Acad Sci USA 98:12078.

Bardet-Biedl Syndrome (BBS, MKKS): This heterogeneous recessive disease involves retinal dystrophy (retinitis pigmentosa), polydactyly, and other anomalies of the limbs, obesity, underdeveloped genitalia and kidney malfunction, diabetes. Mental retardation is also common. Six to seven chromosomal locations, including (BBS1) 11q13, (BBS5) 2q31, (BBS3) 3p12–p13, (BBS4) 15q23, (BBS2) 16q21, and (BBS6) 20p12 have been reported earlier. The BBS8 locus is at 14q32.11, the BBS10 locus is in chromosome 12. The latter involves a chaperonin (Stoetzel C et al 2006 Nature Genet 38:521). The BBS protein has been localized to ciliated structures and to the centrosome (see Fig. B13). The BBS11 locus was assigned to chromosome 9q33 by a high-density single nucleotide polymorphism marker and microarrays; it encodes an E3 ubiquitin ligase (Chiang A et al 2006 Proc Natl Acad Sci USA 103:6287). Actually, the other BBS genes seem to affect the same cellular structures (Ansley SJ et al 2003 Nature [Lond] 425:628). The major form of BBS shares a chromosomal position with McKusick-Kaufman (MKKS, 20p12) syndrome. One basic problem may

involve a chaperonin that folds improperly several proteins. The core complex of BBS in cooperation with the Rab8 GTPase promotes ciliary membrane biogenesis (Nachury MV et al 2007 Cell 129:1201). ►kidney diseases, ►eye diseases, ►triallelic inheritance, ►cilia, ►RAB, ►McKusick-Kaufman syndrome, ►chaperonin, ►Prader-Willi syndrome; Beales PL et al 2001 Amer J Hum Genet 68:606; Mykytyn K et al 2001 Nature Genet 28:188; Katsanis N et al 2001 Hum Mol Genet 10:2293; Badano JL et al 2003 Am J Hum Genet 72:650.

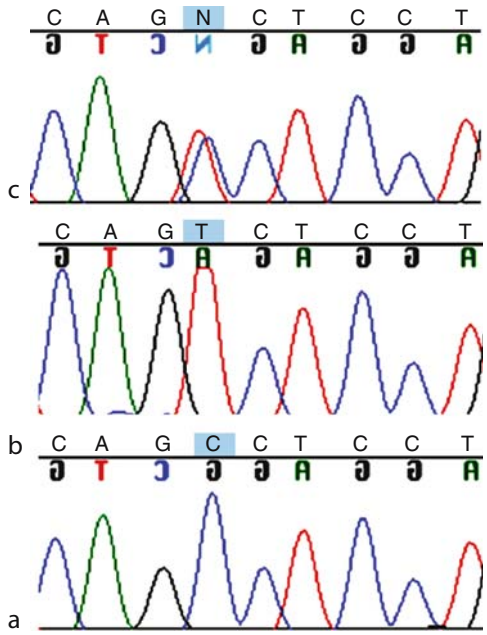

Figure B13. Bardet-Biedl. CCT (a) is normal proline, TCT (b) is homozygous for mutant serine, and (c) is heterozygous at position 130 of the TRIM32 sequence of the ubiquitin ligase gene of BBS11. Courtesy of Annie Chiang and Val Sheffield

Bare Lymphocyte Syndrome (BLS, 19p12 [RFXANK], 16p13 [MHC2TA], 1q21.1-q21.3 [RFX5], 13q14 [RFXAP]): This is a group of recessive severe immunodeficiency diseases caused by defects in the regulation of the major histocompatibility system by any of the four loci identified earlier. Some forms are due to defect(s) either in the HLA class I or class II genes involving lymphocyte differentiation. Some of the defects involve RFX proteins, which bind to the X box of the MHC2TA promoter. MHC II deficiency may also be due to mutation in MCC2TA transactivator. The MHCII molecules are heterodimeric transmembrane proteins. The current therapy is bone marrow transplantation. In future, gene therapy may be feasible. ►immunodeficiency, ►HLA, ►lymphocyte, ►MHC, ►gene therapy, ►RFX; Reith W, Mach B 2001 Annu Rev Immunol 19:331.

Barley (*Hordeum*): This cereal crop is used for feed, food and the brewery industry. The cultivated *H. sativum* is diploid 2n = 14. Some of the varieties of wild barley are polyploids. The cultivated varieties have either two-row (see diagram; right) or four-row spike (see Fig. B14) or six-row depending on the number of florets fertilized per spikelets and bearing seeds. The diagram of the kernel arrangement (two-row and six-row) on the rachis (the axis of the spike) is presented here. The *vrs1* (*six-rowed spike 1*) gene, responsible for the six-rowed spike in barley, has been isolated by means of positional cloning. The wild type *Vrs1* allele (for two-rowed barley) encodes a transcription factor that includes a homeodomain with a closely linked leucine zipper motif. The expression of *Vrs1* was strictly localized in the lateral-spikelet primordia of immature spikes, suggesting that the VRS1 protein suppresses the development of the lateral rows. The loss of function of *Vrs1* resulted in complete conversion of the rudimentary lateral spikelets in two-rowed barley into fully developed fertile spikelets in the six-rowed phenotype (Komatsuda T et al 2007 Proc Natl Acad Sci USA 104:1424). ►haploid [*H. bulbosum*], ►*Hordeum*, ►homeodomain, ►leucine zipper, ►positional cloning; http://www.barleycap.org/; http://www.shigen.nig.ac.jp/barley/Barley.html; http://barleygenomics.wsu.edu/; http://www.barleybase.org/.

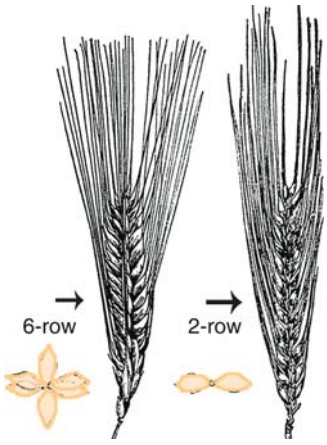

Figure B14. Barley

BARNASE (*Bacillus amyloliquefaciens* ribonuclease): This is a ribonuclease that may be associated with chaperones (see Fig. B15). ►chaperones, ►barstar, ►ribonucleases, ►RBF

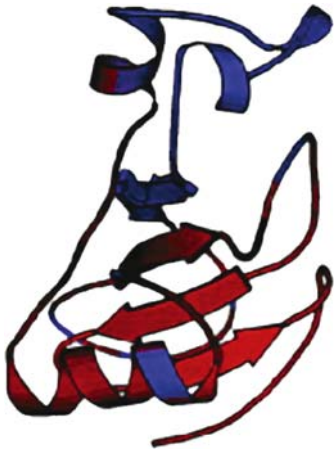

Figure B15. Barnase. Courtesy of Alm, E. & Baker, D. 1999 Proc. Natl. Acad. Sci. USA 96:11305

Barr Body: This dark-stained (heteropyknotic) structure is visible at the periphery of the interphase nuclei of cells that have more than one X chromosome. XY cells do not have a Barr body whereas normal XX female cells have one (see Fig. B16). The number of Barr bodies (named after M.L. Barr) is always one less than the number of X chromosomes, indicating that the non-active X chromosomes remain condensed (dosage compensation). Barr bodies are also present in XXY males. The Barr body is sometimes called sex chromatin. In the leukocytes the Barr body is enclosed in a special nuclear appendage called the "drum-stick" because of its shape. Methylation of CpG dinucleotides is the mechanism of inactivation. ►lyonization, ►methylation of DNA, ►dosage compensation; Heard E et al 1997 Annu Rev Genet 31:571; Hong B et al 2001 Proc Natl Acad Sci USA 98:8703.

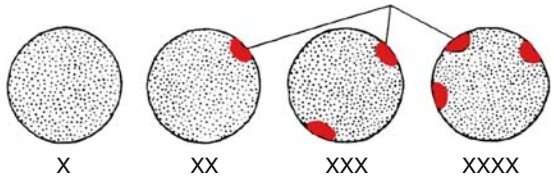

Figure B16. Barr body

Barrage: This is the sign of vegetative incompatibility in fungi. At the zone of contact between the two types of mycelia a distinguishable zone is formed as the result of antagonism between the two strains. (Rizet G 1952 Rev Cytol Biol Végét 13:51).

Barrel: Refers to protein β-sheets closing the interior and α chains on the exterior (see Fig. B17). ►protein structure

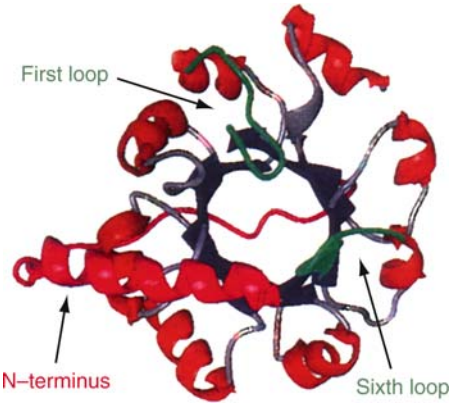

Figure B17. α/β barrel folds of phosphoribosylanthranylate isomerase. The β-sheets in the center are darker. (From Gerlt JA 2000 Nature Struct Biol 7:171)

Barren Stalk: Refers to (*ba*) maize genes (in chromosomes 3, 2 and 9) which affect the tassel or ear or both and cause partial/full sterility. The tassel is male inflorescence. ►tassel-seed; Gallavotti A et al 2004 Nature [Lond] 432:630.

Barrett Metaplasia (gastroesophageal reflux disease, 13q14): The efflux of the content of the stomach exposes the esophagus to acid and bile. This may result in ulceration and eventually adenocarcinomas of this organ. Demethylation of the CDX1 promoter is a key factor in the development of the disease (Wong NACS 2005 Proc Natl Acad Sci USA 102:7565). ►CDX1, ►acid reflux, ►esophagus

Barring Gene (*B*): ►autosexing

Barstar: This is an inhibitor of barnase. ►barnase, ►RBF; Hartley RW 1989 Trends Biochem Sci 14:450.

Barth Syndrome: ►endocardial fibroelastosis

Bartter Syndrome: Type 1 (15q15-q21.1) is a defect in the NaKCl transporter. Type 2 dominant, human chromosome 11q24 encoded disease is characterized by salt wasting and low blood pressure, accompanied by excessive amounts of calcium in the urine. The basic defect lies in an inward rectifier potassium ion channel. Type 3 (1p36) involves Chloride channel B. ►ROMK, ►Gitelman syndrome, ►Liddle syndrome, ►hypoaldosteronism, ►hypertension, ►ion channels, ►hypokalemia

β-ARK: β–adrenergic receptor kinase. ►adrenergic receptors

Basal: This means at or near the base.

Basal Body: Refers to a group of microtubules and proteins at the base of cilia and flagella of eukaryotes (see Fig. B18). ►microtubule, ►cilia, ►flagellum

Figure B18. Basal body

Basal Cell Carcinoma: ►nevoid basal cell carcinoma

Basal Lamina: This is the same as the basement membrane. ►basement membrane

Basal Level Elements (BLE): These perform enhancer-type functions in gene regulation and can occur at several positions. ►enhancer

Basal Promoter: This is normally situated in the region −100 bp upstream of the transcription initiation site and contains various regulatory elements of transcription. ►promoter, ►transcription factors, ►core promoter

Basal Transcription Factors: ►transcription factors

BASC: This is one of several similar *Drosophila* genetic stocks, containing the dominant *Bar* (*B*), the recessive eye color allele, *apricot* (w^a), and several *scute* inversions. The B and w markers identify the untreated chromosomes of untreated females and eliminate the cross overs with the treated X chromosomes of males. The mutagenic effectiveness is determined on the basis of the reduced proportion of males in F2 if a lethal or sublethal mutation was induced in the X chromosome by the treatment. This type of analysis is called Muller-5 technique after H.J. Muller who designed the first stocks. The advantage of these stocks is that both males and homozygous females are completely fertile whereas the XO males are poorly viable and no cross overs appear along the X chromosome. Variegation may occur in some unlinked genes.

Rarely some exceptional females are also detected due to an unequal sister chromatid exchange in the inversion heterozygote females. ►ClB, ►autosomal dominant mutation, ►autosomal recessive mutation; Inoue Y 1992 Genetica 87:169; Forbes C 1981 Mutation Res 90:255, see Fig. B19.

BASC (BRCA1-associated genome surveillance complex): This includes tumor suppressors, DNA repair proteins, DNA replication factor C, etc. ►breast cancer; Wang Y et al 2000 Genes Dev 14:927.

Base: Refers to the lowest part of a structure or a compound or an ion which can combine with protons to yield salt. *Nitrogenous bases* such as the

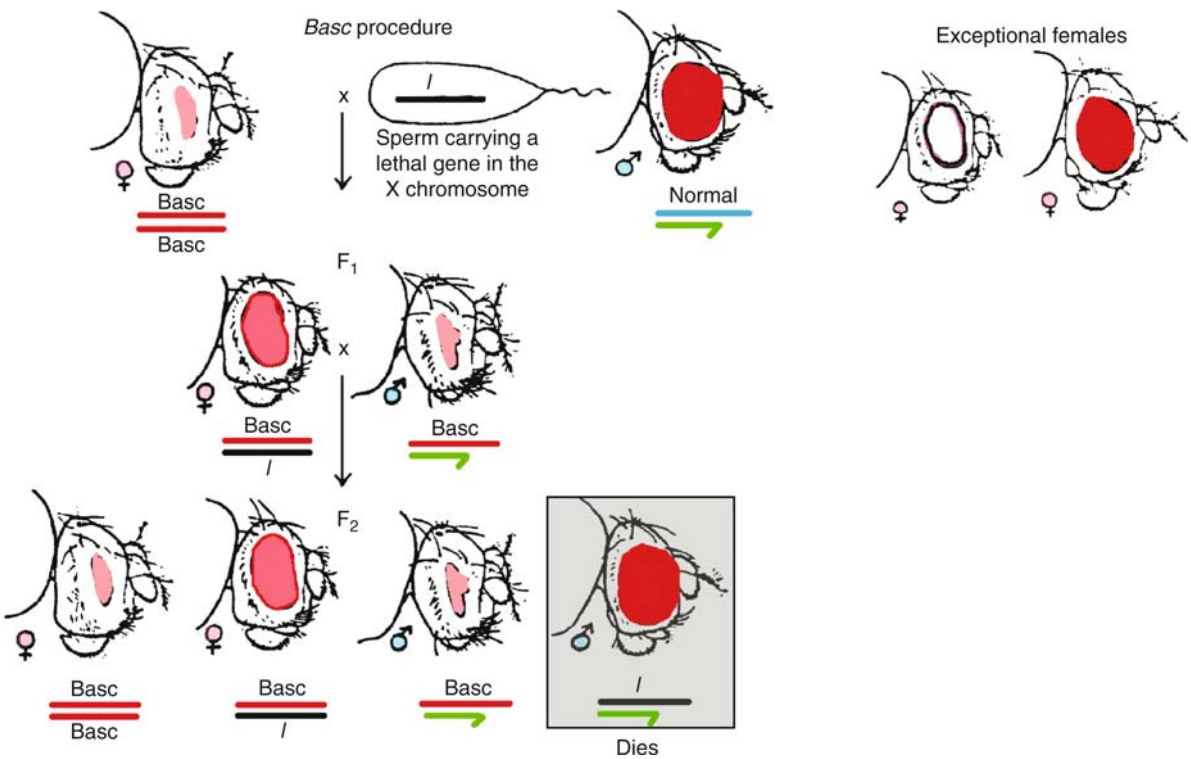

Figure B19. Basc

pyrimidines and purines of nucleic acids. ►nucleic acid bases

Base Analogs: These are nucleic acid bases or nucleosides similar to the normal compounds but cause mutation when incorporated into the DNA either by incorporating in the wrong place or by mispairing with the incorrect base. The most commonly used mutagenic base analogs are 5-bromouracil (thymine analog) and 2-aminopurine (adenine analog). Base analogs expanded by an intercalated benzene ring have been synthesized and the expanded xA and xT analogs form Watson–Crick pairs with the natural complementary base when incorporated into the DNA (see diagram). A single such pair decreases the stability of the double helix but longer tracts are stable. The dxA is a violet-blue fluorochrome (max. emission 389 nm). The dxT appears violet (max. emission 375 nm). When all expanded base analogs are in one strand the binding ability of the natural strand is enhanced making it potentially useful for microarrays, antisense and DNA–RNA analyses (Liu H et al 2003 Science 302:868). ►hydrogen pairing, ►base substitution, ►point mutation, ►universal base, ►microarray hybridization, ►antisense technologies, ►isoguanine, ►modified bases, Freeze E 1959 J Mol Biol 1:87.

Base Composition: Refers to the percentage of nucleotides in DNA or RNA.

Base Excision Repair (BER): ►DNA repair

Base Flipping: Some enzymes such as methyltransferases, glycosylases, T4 endonuclease V, *E. coli* phospholyase and endonuclease III must access the bases, which are inside the sugar-phosphate backbone of the B DNA double helix in order to be recognized by the active site of the protein. Some bases are swung out of the helix into an extra-helical position to meet the requirement. Base flipping is important for the hairpin processing reaction in transposition because it performs two opposite but closely related functions. It disrupts the double helix, providing the necessary strand separation and steric freedom, also the transposase appears to position the second DNA strand in the active site for cleavage using the flipped base as a handle (Bischerour J, Chalmers J 2007 Nucleic Acids Res 35:2584). ►methylation of DNA, ►glycosylases, ►endonuclease, ►DNA repair, ►ABC excinucleases, ►photolyase, ►cyclobutane ring; Cheng X, Roberts RJ 2001 Nucleic Acids Res 29:3784; Patel PH et al 2001 J Mol Biol 308:823; Huang N et al 2003 Proc Natl Acad Sci USA 100:68; Luo J, Bruice TC 2005 Proc Natl Acad Sci USA 102:194.

Base Modifying Agents: Nitrous acid causes oxidative deamination, hydroxylamine converts cytosine into hydroxylamino-cytosine (a thymine analog), and alkylating agents place alkyl groups at several possible positions to purines and pyrimidines. ►chemical mutagens, ►base substitution, ►point mutation, ►hydrogen bonding

Base Pair (bp): Refers to hydrogen bonded A = T and G≡C in DNA or A = U in double-stranded RNA. (►mismatch, ►mispairing, ►hydrogen pairing, ►universal bases, ►Watson-Crick model)

Base Pair Opening (base flipping): A nucleoside unit swivels out of the DNA helix and inserts into the recognition pocket of a protein. Such nucleoside extrusion and extra-helical recognition may take place by processing the DNA by various glycosylases and endonuclease action. ►base flipping

Base-Pair Stepping: ►nucleic acid chain growth

Base Promoter: ►core promoter

Base Sequencing: ►DNA sequencing

Base Stacking: The nucleotides in parts of a polynucleotide chain may lie in such a way that the faces of the rings are appositioned (see Fig. B20). The stacking is most likely to occur by non-covalent forces near the chain termini where the bases move somewhat. It imparts some rigidity to the strand(s). The stacking is detectable by physical methods such as circular dichroism and optical rotatory dispersion. Reagents, which weaken hydrophobic reactions, eliminate the stacking, and heating reduces the stacking resulting in hyperchromicity. Destruction of hydrogen bonding also reduces the stacking in double-stranded DNA. Base stacking may occur in double-stranded molecules where the pairing is weakened by deletions or mismatches. ►circular dichroism, ►optical rotatory dispersion, ►hyperchromicity, ►excimer; Kool ET 2001 Annu Rev Biophys Biomol Struct 30:1.

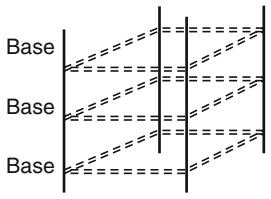

Figure B20. Base stacking

Base Substitutions: When a pyrimidine in the DNA is replaced by another pyrimidine or a purine is replaced by another purine the change is a *transition*. When a purine is replaced by a pyrimidine, or vice versa, a *transversion* takes place. These changes cause mutation in the DNA and they may also lead to amino

acid replacement in the protein if the base substitution involves a non-synonymous codon. Many mutagens cause base substitutions, e.g., hydroxylamine (NH_2OH) targets cytosine (C) and hydroxyaminocytosine is formed which is a thymine analog. As a result a C≡G base pair is replaced by a T = A bp. Similarly, 5-bromouracil may cause, through a tautomeric shift, T = A to be replaced by C≡G; and 2-aminopurine, an adenine analog, causes a tautomeric shift that may lead to the replacement of an A = T pair by a G≡C pair. Some other chemicals, e.g., nitrous acid, by deamination, changes cytosine to uracil and converts adenine into guanine. These base substitutions may also cause reversions. A hydroxylamine-induced mutation may be reverted by bromouracil, etc. It is generally assumed that base substitutions occur independently and coincidental double substitutions should be rare. Some genetic repair mechanisms may, however, bring about more than single replacements. A large-scale evolutionary study revealed that double substitutions may occur in the serine codons of primates at the high frequency of 0.1/site/billion years. (►DNA, ►hydrogen bonding, ►point mutation, ►base analogs, ►evolution and base substitutions, ►incorporation error, ►replication error, ►chemical mutagens, ►substitution mutations, ►mutation and DNA replication, ►mutation bias, Freese E 1959 Brookhaven Symp Biol 12:63, base substitution model selection tool: http://darwin.uvigo.es/software/modeltest_server.html).

Base-Call: This process evaluates the nucleotide sequence information using the PHRED program. ►base-calling, ►PHRED, ►PHRAP

Base-Calling: This means identifying the correct nucleotide in a sequence in the DNA. Also, identifying the correct base sequence on the basis of hybridization in microarrays compared to the actual direct sequencing information. Miscalls are false identifications The multifluorescence discrimination (pulsed multiline excitation, PME) correlates a sequence of excitation pulses from four monochromatic wavelength laser sources with detector response from emission intensities of fluorescently labeled DNA fragments. For this purpose, a new set of dyes has been developed that spans with absorption maxima the entire visible spectrum. This procedure appears superior to the ones previously used (Lewis EK et al 2005 Proc Natl Acad Sci USA 102:5346). ►microarray hybridization, ►PHRAP, ►PHRED; Walther D et al 2001 Genome Res 11:875.

Basedow Disease: ►goiter

Basement Membrane (basal lamina): This is less than 500-nanometer thick laminated condensation of the extracellular matrix (including laminin, collagen IV and other proteins) on the basal surfaces of epithelia and condensed mesenchyma. The basement membrane is an attachment platform and a barrier to cell mixing. Several human diseases involve anomalies of basement membranes and/or associated proteins. ►extracellular matrix, ►proteoglycan, ►laminin, ►collagen, ►Alport's disease, ►Goodpasture syndrome, ►fibromatosis; Quondamatteo F 2002 Histochem J 34:369; Masunaga T 2006 Connect Tissue Res 47:55.

Base-Pairing: ►hydrogen pairing

Base-Specific Reagents for DNA Single Strands: 1. Dimethylsulfate (DMS) + hydrazine the methylated cytosine is cleaved at the 3′ position. 2. DMS alone methylates guanine. 3. Osmiumtetroxide or potassium permanganate oxidizes the C5-C6 double bonds in thymidine. 4. Diethylpyrocarbonate ($O[CO_2C_2O_5]_2$) preferentially modifies adenine at N-7 although it affects other purines as well. (►DNA sequencing [Maxam & Gilbert method])

Base-Stacking: ►base stacking

Bash (B cell-restricted adaptor protein, BLNK/SLP-64): After ligation Sly tyrosine kinase phosphorylates it and it binds various B cell signaling proteins that control B cell development. Its role is similar to that of SLP-76 for T cells. ►SLP-76, ►B lymphocyte, ►ITIM; Tsuji S et al 2001 J Exp Med 194:529.

Basic Chromosome Number: This is found in the gametes of diploid organisms and it is represented by x; in polyploids the haploid (n) number may be 2x, 3x and so on, depending on the number of genomes contained. The basic number is frequently called a genome. ►chromosome numbers, ►genome, ►polyploid

Basic Copy Gene: It is the silent copy of a *Trypanosoma* gene that is activated by transposition to an activation site in the telomeric region of the chromosome. ►*Trypanosomas*, ►telomere

Basic Dye: The dye stains negatively charged molecules. ►stains

Basidiomycetes: A taxonomic group of fungi bearing the meiotic products in basidia. ►basidium, ►mushroom

Basidium: This is a fungal reproductive structure generally in the shape of a club where meiosis takes place and then the haploid basidiospores are released infecting the host plants (see Fig. B21). ►stem rust

Figure B21. Basidium with four meiotic spores

B

Basonuclin: This is a cell type-specific Zinc finger protein with a nuclear localization sequence and a serine stripe (serine-rich region). It is in abundance in the human keratinocyte nuclei but in the absence of phosphorylation it is in the cytosol. Basonuclin is also found in the epidermal cells and the germ cells of the testis and the ovary. It binds to the rRNA promoter and apparently regulates rRNA transcription. ►Zinc finger, ►nuclear localization signal; Tian Q et al 2001 Development 128:407.

Basophil: Refers to a type of white blood cell which is well stainable with basic cytological dyes. These cells contain conspicuous secretory granules and release histamine and serotonin in some immune reactions. Any other (acidic) structure or molecule with an affinity for positive charges. ►granulocytes, ►blood, ►immune system

Basta: This is a glufosinate ammonium herbicide, pesticide and a selective agent in plant transformation. ►herbicides, ►transformation genetic [plants]; Rathore KS et al 1993 Plant Mol Biol 21:871.

Bastard: This means a hybrid [in German]; and an illegitimate or undesirable offspring [in English].

Bat: *Carollia perpicillata* 2n = 21 in males, 20 in females; *Glossophaga soricina* 2n = 32; *Desmodus rotundus murinus* 2n = 28; *Atropzous pallidus* 2n = 46; *Eptesicus fuscus* 2n = 50; *Myotis velifer incautus* 2n = 44; *Nysticeius humeralis* 2n = 46. They are useful predators of insects but constitute a reservoir of different pathogenic viruses such as SARS, Ebola, Marburg and other strains. (Dobson AP 2005 Science 310:628).

Bateman's Principle: It states that the reproductive success of males shows greater variation than that of females because of greater competition among males and larger number of male gametes.

Batesian Mimicry: This is an adaptive evolutionary device. Certain species develop phenotypic characteristics of sympatric species (models) in order to increase their chances of survival. The models are repugnant (distasteful) to certain ►predators, which avoid them, and so the mimickers when mistaken for the models also escape destruction. Batesian mimicry is more common among females than males (butterflies) because females are more often subject to predation than males. (►adaptation, ►natural selection, ►Müllerian mimicry, see Fig. B22).

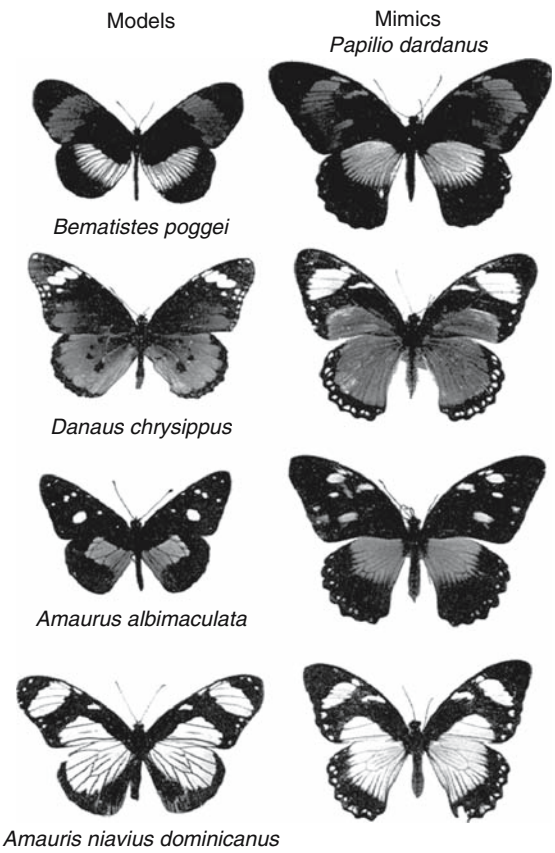

Figure B22. Batesian mimicry. Batesian mimicry in butterflies. (From Sheppard, P.M. 1959 Cold Spring Harbor Symp. Quant. Biol. 24:131)

Batten Disease: This is a recessive (human chromosome 16p12.1–p11.2) juvenile-onset familial amaurotic idiocy caused by lipid accumulation in the nerve tissues and vacuolization of the lymphocytes. Its incidence is $\sim 5 \times 10^{-5}$. The disease seems to be associated with low vacuolar pH. In the yeast *btn1* defects chloroquine reverses the phenotype. ►amaurotic familial idiocy, ►ceroid lipofuscinosis, ►chloroquine, ►Vogt-Spielmeyer disease, ►PPT; Luiro K et al 2001 Hum Mol Genet 10:2123, http://www.ucl.ac.uk/ncl/.

Batten-Turner Syndrome: ►myopathy congenital, ►human chromosome 16

Bauplan (body plan): Refers to the pattern of body organization. ►tagmosis

BAX (BCL2-associated X protein): ►BCL, ►porin, ►Puma, ►Bid; Chipuk JE et al 2004 Science 303:1010.

Bayes' Theorem: The theorem permits the estimation of various conditional probabilities and is used for

decision-making processes. In the simplest general form:

$$P(A|B) = \frac{P[B|A]P[A]}{P[B|A]P[A] + P[B|A']P[A']}$$

Its application in genetics can be illustrated by assuming that there are three individuals, two homozygotes for a semi-lethal dominant factor and one heterozygote for the same semi-lethal and incompletely dominant gene. There are problems with visual classification at an early stage of the development. Assuming that the two homozygotes [A] have 60%, and the heterozygote [A′] is expected to have 80% viability. Thus (P [B|A] = 0.6, and P [B | A′] = 0.8.

The chance of selecting an individual of either genotype is P [A] = 2/3 and P [A′] = 1/3. If the selected individual turns out to be very weak, and if one is uncertain about the choice, one may want to determine—in view of the information available—the probability of having selected the heterozygote:

$$P(A|B) = \frac{[0.8][0.33]}{[0.8][0.33] + [0.6][0.67]} \cong 0.4$$

Basically, the Bayesian method considers the classical population parameters as random variables with a specific *a priori* probability of distribution. Then the *conditional probability* is estimated on the basis of the a priori distribution. The conditional probability is thus a property of *a posteriori* distribution because the accepted or supposed *a priori* distribution is used for the estimation of an existing situation in a population. ►probability, ►*a priori*, ►prior distribution, ►conditional probability, ►risk, ►inference, ►Bernoulli process; Shoemaker JS et al 1999 Trends Genet 15:354; Bernardo JM, Smith AFM 1994 Bayesian theory, Oxford University Press, Oxford, UK; Beaumont MA, Rannala B 2004 Nature Rev Genet 5:251.

Bayesian Mapping: This can be applied to QTL. Inferences are made on the basis of the joint posterior distribution of all unknown variables given the prior distribution of all unknowns of the observations. It uses Monte Carlo approximation to the multiple integration required. It may be useful for analyzing complex animal pedigrees. ►QTL, ►genetic networks, ►BAMBE, ►Monte Carlo method, ►Bayes' theorem; Yi N, Xu S 2000 Genetics 155:1391.

Bayesian Network: This can represent systems with multiple interactions. It can detect direct molecular interactions and also indirect effects that have not been directly observed. ►networks, ►signal transduction, ►genetic networks; Sachs K et al 2005 Science 308:523.

BB-1: This is the same as B7 or CD80.

BBB (blood-brain barrier): The mechanism seriously limits transport to the central nervous system (CNS) because of the tight junction of the endothelial cells of the brain capillaries. Molecules larger than 180 MW and viruses may be excluded. Lymphocytes may enter the central nervous system but are not retained unless foreign antigens are present. Neurons and some other cells may poorly express MHC proteins and escape the effects of cytotoxic T cells. In such a situation different viruses (rubella, measles, polyoma JC, herpes simplex, rabies, mumps) may infect the brain. In the absence of such a barrier serum may leak into the brain causing edema. Angiotensin may be required to maintain BBB. Antagonists of the hypothalamic growth hormone-releasing hormone such as JV-1–42 (a peptide analog) enter the brain and can accumulate there. Since this hormone is present in several types of cancers there may be an opportunity for the treatment of glioblastomas (Jaeger LB et al 2005 Proc Natl Acad Sci USA 102:12495). Some lipids (but not by water-soluble or protein) molecules may however overcome the barrier if the cargo protein is associated with a carrier such as transferrin (Demeule M et al 2002 J Neurochem 83:924). High doses of the enzyme β-D-glucuroside glucuronosohydrolase injected into the bloodstream of mice, mutant in mucopolysaccharidosis VII, passed through the blood-brain barrier (Vogler C et al 2005 Proc Natl Acad Sci USA 102:14777). Receptors on the blood-brain barrier bind ligands to facilitate their transport to the central nervous system. The use of the lentivirus vector system can deliver the lysosomal enzyme glucocerebrosidase and a secreted form of GFP (green fluorescent protein) to the neurons and astrocytes in the CNS. Fusing the low-density lipoprotein receptor-binding domain of the apolipoprotein B to the targeted protein is useful in delivering to the CNS (Spencer BJ, Verma IM 2007 Proc Natl Acad Sci USA 104:7594). ►angiotensin, ►protein transduction, ►multiple sclerosis, ►MHC, ►glucose transporters, ►GH, ►gliomas, ►transferrin, ►enzyme replacement therapy, ►siRNA; Asahi M et al 2001 J Neurosci 21:7724.

β-Catenin: This is a component of the cadherin-catenin cell adhesion complex. ►adherens junction

B-Cell Differentiation Factor: ►interferon β-2 (IFNB2)

B-Cell Growth Factor: ►IL-4, ►interleukins

BCG: ►*Bacillus Calmette-Guerin*

BCGF (B cell growth factor): Refers to 12-kDa cytokine produced by activated T cells. ►B cell, ►T cell, ►cytokine, ►lymphocytes

BCIP: 5-bromo-4-chloro-3-indolyl phosphate is used in combination with nitroblue tetrazolium (NBT; it

reveals precipitated indoxyl groups) for the detection of antigen-antibody—antibody-AP (alkaline phosphatase) complexes. ►antigen, ►antibody

BCL1, BCL2, BCL3, BCL5, BCL6 (B cell lymphoma): These are leukemia oncogenes but they are also upregulated in various other types of cancers. Bc*l*-1 is cyclin D1 (see Fig. B23). Bc*l*-2 (18q21.3) suppresses apoptosis (by phosphatase action when bound to calcineurin) as a defense against malignant tumorigenesis and suppresses signaling by NF-AT. BCL2 apparently shuts off the voltage-dependent anion channel on the mitochondrial membrane and prevents the leakage of apoptotic cytochrome c into the cytosol to guard against apoptosis. It also promotes regeneration of severed cells in the central nervous system. The Bc*l*-2 protein functions as an ion channel and a docking protein. BCL-2 is located in the outer membrane of the mitochondria, nuclei and the endoplasmic reticulum. Bc*l*-6 regulates STAT and cytokine signaling. Bc*l*-6 suppresses p53, a pro-apoptotic and cancer suppressor protein in the germinal center B cells (Phanm RT, Dalla-Favera R 2004 Nature [Lond] 432:635). The Bc*l* protein is usually up-regulated in lymphomas, and gene therapy involving deoxyoligonucleotides (such as 5′-TCTCCCAGCGTGCGC-*CAT*-3′) targeted to the *AUG* initiator codon has been used. There are over a dozen members of the Bc*l* family. Bc*l*-2 is homologous to *Ced-9* of *Caenorhabditis*. Antimycin A, a complex of highly toxic antifungal substances, inhibitors of electron transport, bind Bc*l* and Bc*l*-X and favor apoptosis and may thus protect against cancerous growth. The Bc*l* proteins can be effectively modulated by engineered BH3 fragments and the so-activated BID protein promotes apoptosis and destroys leukemia cells (Walensky LD et al 2004 Science 305:1466). The anti-apoptotic BCL-2 favors the maintenance of blood cancer, leukemia (Letai A et al 2004 Cancer Cell 6:241). Bc*l*-2 interferes with RAD51 controlled recombination that may mediate error-free repair and may thus promote cancer-prone conditions independently from its anti-apoptosis effect. The normally anti-apoptotic Bc*l*-2 may interact with Nur77 orphan nuclear receptor and may promote apoptosis. The pro-apoptotic members of the BCL family, BAX and BAK, regulate the inositol triphosphate receptor (IP3R-1) and normally maintain a high calcium level in the endoplasmic reticulum (ER). Mutant BAX and BAK decrease the ER calcium level and the released Ca^{2+} increases mitochondrial permeability leading to apoptosis. BCL2 has the opposite effects (Oakes SA et al 2005 Proc Natl Acad Sci USA 102:105). ►leukemia, ►apoptosis, ►BAX, ►BAD, ►BAK, ►BID, ►IP3, ►Bim, ►malignant growth, ►cyclin, ►NF-AT, ►calcineurin, ►STAT, ►cytokine, ►G3139, ►G3854, ►lymphoma, ►nur77, ►IAP, ►germinal center; p53, Petros AM et al 2001 Proc Natl Acad Sci 98:3012; Saintigny Y et al 2001 EMBO J 20:2596; Vander Heiden MG et al 2001 J Biol Chem 276:19414; Deng X et al 2004 Proc Natl Acad Sci USA 101:153.

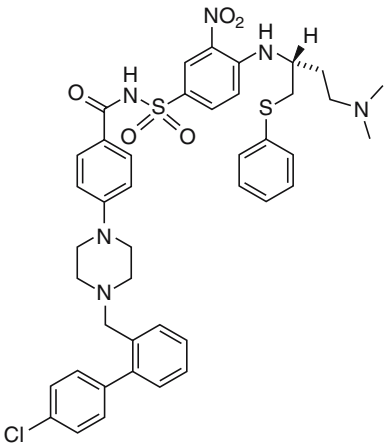

Figure B23. ABT-737 inhibits Bcl-2 family of proteins; regresses solid tumors. (Oltersdorf, T. *et al*. 2005 Nature 435:677)

BCMA: This is one of the B lymphocyte receptors. ►BCR, ►BAFF, ►Blys

β-Conformation: Refers to an extended conformation of a peptide chain; a type of secondary structure. ►protein structure

BCR: Refers to B-cell antigen receptor. ►B lymphocyte

BCR (bcr): The break point cluster region in the Philadelphia chromosome is an area where multiple chromosomal breakages have been observed leading to cancerous transformation (leukemia, ABL). The BCR polypeptide contains 1,271 amino acids and its normal function is a protein serine/threonine kinase. ►Philadelphia chromosome, ►leukemia, ►ABL

B-DNA: This denotes a conformation of the DNA most common in hydrated living cells. ►DNA types

BDNF (brain-derived neurotrophic factor): This is an autocrine growth substance of neurons but it is found in other organs as well. Genetic variant (Val codon 66→Met) predisposes to anxiety and depressive disorders (Chen ZY et al 2006 Science 314:140). ►autocrine, ►neuron, ►TRK, ►ovary, ►Parkinson's disease, ►Rett syndrome, ►anxiety, ►depression; Li Y et al 2005 Nature [Lond] 434:894.

Beacon, Molecular: These are hairpin-shaped single-stranded oligonucleotide genetic probes that become fluorescent after hybridization to the homologous target. One end is covalently bound to a fluorophore and the other end is attached to a non-fluorescent quencher. If the probe does not find homology there is no fluorescence because the quencher prevents it in the hairpin (see Fig. B24). If the probe locates a homologous sequence, unwinding removes the quencher from the vicinity of the fluorophore and the site lights up ❄. ►spectral genotyping, ►molecular beacon; Tyagi S, Kramer FR 1996 Nature Biotechnol 14:303; Heyduk T, Heyduk E 2002 Nature Biotechnol 20:171; Hopkins JF, Woodson SA 2005 Nucleic Acids Res 33:5763.

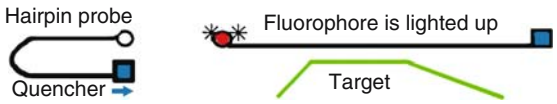

Figure B24. Molecular beacon

Beads-On-A-String: Originally this referred to the (light-microscopically visible) chromosome structure at the pachytene where the chromomeres could be seen as beads-on-a-string. In the 1920s chromomeres were equated with genes, units of function, mutation and recombination. Today, this is also used to describe the DNA strands wrapped around the eight histones (nucleosomes) as seen through an electron microscope. ►nucleosome, ►pachynema, ►chromomeres, ►complex locus

Beagle: ►copia

BEAMing (beads, emulsion, amplification and magnetics): This is a procedure for the quantification of variation in single DNA molecules among a large number of molecules. The purpose is the detection of SNIPS, rare mutations in genes and transcript of importance for function and disease or state of disease progression in specific tissues. Small numbers of DNA molecules that display variations are amplified by polo. Counting the attached beads on the basis of fluorescent labels using flow cytometry leads to the numbers of variants. ►flow cytometry, ►polo, ►SNIPS; Dressman D et al 2003 Proc Natl Acad Sci USA 100:8817.

Bean-Bag Genetics: This phrase was used for characterizing the work of Mendelian geneticists, studying/counting individual genes as they controlled the phenotypes and inheritance. It expressed the contempt of some evolutionists whose interest was in entire organisms rather than in dissecting the mechanisms of the Mendelian factors by statistical means. With advances in physiological, biochemical and molecular genetics new terms such as "factorial genetics/formal genetic" were proposed for the classical approaches. Today, genetics research relies on the wide-reaching methods and principles based on simple and macromolecules and single molecules, networks of molecules as well as biophysics and cybernetic tools and principles of bioinformatics. The boundaries of modern genetics are unrestricted and genetics has permeated the whole field of basic and applied biology. (Haldane JBS 1964 Persp Biol Med 7:343).

Beans (*Phaseolus* spp): These are pulse crops, all with 2n = 2x = 22 chromosomes, including the most common *P. vulgaris* (French or navy beans) and the *P. lunatus* ►Lima bean

Bear: Refers to *Ursus americanus* (black bear) 2n = 74; *Tremarctos ornatus* (spectacled bear) 2n = 52.

Beare-Stevenson Syndrome: An autosomal dominant disorder which is characterized by furrowed, corrugated skin (cutis gyrata), head bone fusions, facial anomalies, abnormal digits, umbilical, genital malformations and early death. The basic defect lies in the fibroblast growth factor receptor 2, encoded at 10q26. Some heterogeneity exists. ►FGF

Beaver: *Castor canadensis*, 2n = 40. A large (>80 cm long) brown rodent.

Becker Disease: ►myotonia

Becker Muscular Dystrophy (BMD): ►muscular dystrophy

Beckwith-Wiedemann Syndrome (EMG syndrome): This disorder is caused by a dominant gene in human chromosome 11p15.5. The symptoms include enlarged tongue (detectable at birth), umbilical anomalies (omphalocele = herniated intestines at the belly button area), hypoglycemia, enlargement of the internal organs (visceromegaly), frequent concomitant kidney and liver anomalies, tumorous striated muscles (rhabdomyosarcoma), etc. It may be associated with trisomy for chromosome 11 and it has been suggested that it is caused by paternal or maternal disomy when the normal (most commonly paternal) chromosome is lost from the trisomic cell lineage, or imprinting. A gene encoding a cyclin-dependent kinase inhibitor ($p57^{KIP2}$) is imprinted and preferentially expressed by the maternal allele may be

responsible for some of the cases. Insulin-like growth factor (IGF2) also has a regulatory effect. Deletion, duplication and balanced translocation have been suggested for some cases. It has been shown recently that KVLQT1 gene, encoding a putative potassium channel, and mapped to the 11p15.5 region controls imprinting not only the Beckwith-Wiedemann gene but also the Jervell and Lange-Nielsen syndrome and the LQT heart arrhythmia. ►disomic, ►disomic uniparental, ►Wilms tumor, ►cancer, ►Jervell and Lange-Nielsen syndrome, ►rhabdomyosarcoma, ►Simpson-Golabi-Behmel syndrome, ►imprinting, ►insulin-like growth factors, ►ion channels, ►cyclin, p57; LQT; epigenesis; Alders M et al 2000 Am J Hum Genet 66:1473; Bliek J et al 2001 Hum Mol Genet 10:467; DeBaun MR et al 2003 Am J Hum Genet 72:156.

Beclin 1 (17q21): This is a mammalian autophagy gene that is deleted in 40–75% of the sporadic ovarian and breast carcinoma cells. ►autophagy

Becquerel: This corresponds to 1 disintegration of radioactive material/second; 1 becquerel = 2.7027×10^{-11} Curie (≈ 27 picocuries). ►Curie

Bedwetting: ►nocturnal enuresis

Beech (*Fagus*): This is a hardwood tree, *F. sylvestris*, 2n = 2x = 24 (see Fig. B25).

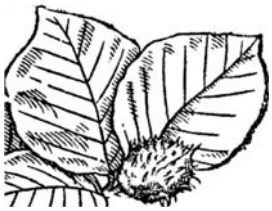

Figure B25. Beech, *F. sylvestris*

Beethoven, Ludwig van (1770–1827): Beethoven was one of the greatest musical geniuses of the eighteenth-nineteenth century. Both his grandfather Louis and his father Johann were outstanding singers in Louvain and Liège. Later his father worked as a musical director (Kapellmeister) in Bonn. His talent was obvious from early childhood (see Fig. B26). Only three of his seven siblings survived to adulthood but none of them had any special musical ability. At the age of 22 he moved from Bonn to Vienna and rose to become a highly esteemed member of the nobility and the imperial court.

Many famous contemporaries were his friends. He was financially independent because of the generosity of his admirers. Beethoven never married because he was plagued by various minor and major illnesses, including gradual loss of hearing leading to near total deafness by the age of 40. Ironically, during this phase, he composed some of his most famous works such as the Emperor concerto, a new version of the opera Fidelio, the seventh and the ninth symphonies. Although he had a platonic relationship with several women, he became isolated

Figure B26. This is the Brunszvik mansion of Martonvásár, Hungary where Beethoven was a guest two times. His bust is in the park at the lake behind the building. Above the spruce tree is the triple window of the music room where he composed the Moonlight Sonata. By the strange fate of life, many years ago author of this book lived in the same elegant room—with beautiful inlaid wood ceiling—for about a month

because he was troubled by his hearing loss (see Fig. B27). ►musical talent, ►genius, ►Bach, ►Mozart, ►Strauss; Mai FM 2007 Diagnosing genius: The life and death of Beethoven, McGill-Queen's University Press, Montreal, Canada.

Figure B27. Beethoven in the park. Beethoven bust in the park in Martonvásár. (Courtesy of Prof. J. Kiss)

Beethoven (*Bth*): This is a mouse mutation with dominant progressive hearing loss at the locus homologous to the human gene TMC1, causing DFNA36. Beethoven was apparently afflicted by the same mutation. ►deafness

Begonia (*Begonia semperflorens*): This is an ornamental plant, 2n = 34.

Behavior Genetics: This branch of genetics analyzes the genetic determination and regulation of how organisms behave. Most of the traits (courtship, bird and frog songs) are under multigenic control and they depend to a large extent on the influence of the environment. In a few cases large effects of single genes have also been observed (Hall JC 2002 J Neurogenet 16:135). In *Drosophila* the *fruitless* (*fru*) gene is involved in the determination of courtship. The Fru^M transcription factor protein is expressed in about 2% of the neurons of the central nervous system. When the yeast *GAL4* gene is inserted into the *fru* locus Fru^M is expressed in all the peripheral sensory systems involved in courtship. Gal4 is a positive regulatory protein of the yeast galactose gene. Inhibition of Fru^M in the olfactory system components reduces olfaction-dependent courtship. Transient inactivation of all Fru^M-expressing neurons terminates courtship behavior without affecting other behavioral traits. The expression of Fru^M in female flies results in the manifestation of male courtship ritual in females toward other females (Manoli DS et al 2005 Nature [Lond] 436:395). The mosquito *Toxorhynchites brevipalpis* "hears" the frequency of the wing beats and both males and females recognize the opposite sex on this basis (Gibson G, Russell I 2006 Current Biol 16:1311).

The Western scrub-jay (*Aphelocoma*), a corvid bird, hides food for future consumption and adopts a very elaborate pattern for protection of the cache (see Fig. B28). The bird can remember which individual saw the cache and alters its tactics to elude potential

Figure B28. Behavior genetics-*Aphelocoma*

thieves (Dally JM et al 2006 Science 312:1662). In the honeybee a single gene controls the habit of uncapping the honeycombs containing dead larvae but another gene is required for the removal of the dead brood. If both genes are present the colony becomes resistant to the bacterial disease foul brood because of improved hygienic behavior.

Alcoholism, criminality, etc. in humans may be determined by several genes and by the social environment. The Lesch-Nyhan syndrome is caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyl transferase, which renders the salvage pathway of nucleic acid inoperational. As a consequence purines accumulate and uric acid is overproduced leading to gout-like symptoms but more importantly the nervous system is also affected, leading to antisocial behavior and self-mutilation. This gene has been isolated and cloned and may be transferred to the afflicted human body for gene therapy. Since behavioral traits are determined by the nervous system, neurogenetics may provide the answer to many serious conditions such as Alzheimer's disease (an amyloid accumulating presenile dementia), neurofibromatosis (a soft tumor of the nervous system affecting the entire body involving a protein resembling a GTPase activator), etc. Behavioral alteration may result from brain damage without a genetic change. A lesion in the frontal lobe of the brain may result in sociopathy in humans and in macaques (Rudebeck PH et al 2006 Science 313:1310).

In recent years progress has been made in the molecular analysis of memory and learning ability through studies of simple organisms such as the slug *Aplysia* and *Drosophila*. Several genes involved in the development of the nervous system of *Drosophila* have been cloned. Recent research has shown that mice without the *fos* gene fail to nurse their pups presumably because of some brain lesions.

B

For genetic and developmental analysis of nerve functions, the nematode, *Caenorhabditis* is particularly well suited because its entire nervous system consists of only 302 cells. In the tobacco hornworm (*Manduca sexta*) the feeding preference depends on an acquired recognition template. The naïve larvae can feed on different plant species but once they have been exposed to a steroidal glycoside (indioside D) present in tobacco leaves, they become "addicted" to that host. Although all biological traits and attributes have some biological basis, the influence of the environment has a very substantial role in the development of behavioral traits. Frequently, for unknown reason, identical behavioral traits are expressed differently in isogenic strains maintained in different laboratories under apparently identical conditions. This fact indicates that genes cannot exonerate criminal behavior, neither should people be condemned on the basis of collective responsibility by supposed sharing of a common gene pool.

Microarray profiles of the brain are associated with behavior (Whitfield CW et al 2003 Science 302:296). Magnetic resonance imaging (MRI) of the brain distinguishes between extrovert and neuroticistic behavior on the basis of humor-driven blood oxygenation level-dependent signals in different regions of the brain (Mobbs D et al 2005 Proc Natl Acad Sci USA 102:16502). The molecular basis of a few mouse behavioral genes is now known. Deletion from the RORα gene causes the *staggerer* phenotype. The *vibrator* mouse carries a retroposon in an intron of the phosphatidylinositol transfer protein. The *weaver* ataxia is a serine→glycine replacement gene in a G-protein-gated inwardly rectifying K-ion channel. The different types of estrogen receptors control mating behavior. Introduction by a viral vector the single gene of the vasopressin V1a receptor into the ventral forebrain of the polygamous species of the Vole substantially increased monogamous behavior (Lim MM et al 2004 Nature [Lond] 429:754). Oxytocin knockout male mice are afflicted by an olfactory recognition problem, a deficit in the amygdala of the brain which can be corrected by injecting oxytocin. Trpc2 vomeronasal organ-specific neuron ion channel mutant mouse females display unique characteristics of male sexual and courtship behavior such as mounting, pelvic thrust, solicitation, anogenital olfactory investigation, and emission of complex ultrasonic vocalizations towards male and female conspecific mice. The same behavioral phenotype is observed after surgical removal of the vomeronasal organ of adult animals, and is not accompanied by disruption of the estrous cycle and sex hormone levels (Kimchi T et al 2007 Nature [Lond] 448:1009). Galanin, a neuropeptide with inhibitory action on neurotransmission and memory, causes Alzheimer's disease-like behavior in mice. Enkephalin (opioid peptide) knockout and/or the loss of its receptor lead to an increase in anxiety. Mutation in the dopamine receptor D2 results in Parkinsonism-like phenotype. The loss of the serotonin receptors increases anxiety. NO synthase knockout mice express greater aggressiveness. Single genes, encoding pheromone-binding protein(s) may regulate complex social behavior, such as recognition of conspecific individuals in social insects (Keller L, Parker JD 2002 Current Genet 12[5]:R180). The nursing of female rabbits and the sucking of pups seem to be regulated by the pheromone 2-methylbutenal (Schaal B et al 2003 Nature [Lond] 424:68). Mice deficient in the TRP2 gene lose their ability to distinguish between sexual types and consequently males mate with both males and females. In the carpenter ants (*Camponotus japonicus*) hydrocarbon blends produced by sensillae of the antennae are the chemical signals for the recognition of nestmates and for the aggressive behavior towards non-nestmates (Ozaki M et al 2005 Science 309:311). Behavior is subject to epigenetic modification. In rats well cared by their mothers during early life the hippocampal glucocorticoid receptor is methylated to a lesser degree than in the neglected pups. This condition persists during the later stages of development. Infusion of L-methionine, a precursor of S-adenosylmethionine and methyl donor for DNA methylation reverses the effect of the maternal behavior on DNA methylation (Weaver IC et al 2005 J Neurosci 25:11045). The higher level of maternal androgens—especially in the later phase of pregnancy—influences social rank and aggressiveness in spotted hyenas (Dloniak SM et al 2006 Nature [Lond] 440:1190). ►courtship in *Drosophila*, ►personality, ►alcoholism, ►cognitive abilities, ►autism, ►addiction, ►fate mapping, ►behavior in humans, ►altruistic behavior, ►aggression, ►ethics, ►instinct, ►morality, ►eugenics, ►avoidance learning, ►cross fostering, ►FOS, ►human intelligence, ►behavior, ►affective disorders, ►Huntington's chorea, ►Alzheimer's disease, ►mental retardation, ►attention deficit hyperactivity, ►dyslexia, ►homosexual, ►vomeronasal organ, ►oxytocin, ►microsatellite, ►antenna, ►sensillum, ►sex determination, ►Gal4; Pfaff D 2001 Proc Natl Acad Sci USA 98:5957; McGuffin P et al 2001 Science 291:1232; Toye AA, Cox R 2001 Curr Biol 2001 11:R473; Krieger MJB, Ross KG 2002 Science 295:328; Bucan M, Abel T 2002 Nature Rev Genet 2002 3:114; Stowers L et al 2002 Science 295:1493; Kucharski R, Maleszka R 2002 Genome Biol 3[2]:res.0007.1; Rankin CH 2002 Nature Rev Genet 3:622; Sokolowski MB 2002 Nature [Lond] 419:893; Sherman G, Vissscher PK 2002 Nature [Lond] 419:920; chemical communication; Proc Natl Acad Sci USA 100, Suppl 2 [2003]; application in law, education, employment and insurance: Rothstein MA 2005 Nature Rev Genet 6:793.

Behavior in Humans: For long human behavior has been suspected to be genetically determined but with a few exceptions (Lesch-Nyhan syndrome, Tay-Sachs disease, Huntington's chorea, etc.). However, the genetic control is not completely understood. The majority of the behavioral traits are under the control of several genes. In such instances the tools of quantitative inheritance are needed, such as heritability, comparison of monozygotic-dizygotic twins with the general population and QTL mapping. The approximate ratios of monozygotic:dizygotic concordance are for alcoholism, females (1.1) versus males (1.7), dyslexia (1.7), Alzheimer's disease (2.1), major affective disorders (2.4), schizophrenia (2.7), autism (6.7). Heritabilities determined by intraclass correlation were 0.22 for memory, 0.22 for mental processing speed, 0.38 for scholastic achievement in adolescence, 0.40 for spatial reasoning, 0.42 for adolescent vocational interest, 0.46 for neuroticism, 0.50 for verbal reasoning and 0.52 for general intelligence. [Data based on Plomin, Owen, McGuffin 1994 Science:264:1733]. Cognitive abilities are also studied as part of the developmental genetic pattern (longitudinal genetic analysis). Multivariate genetic analysis determines the covariance (►correlation) among multiple traits. Although some genetic effects are specific to certain abilities, the majority of the genetic components have overlapping effects. The studies must also consider in assessing behavioral, cognitive genetic traits that form a continuum and the anomaly in a proband or several individuals may be just the extreme form of a normally existing behavioral pattern. Behavioral traits in general have about 50% or more environmental components. These effects include family relationships and changes in such relationships (e.g., divorce, death, accidents), social environment (economic status, schools, drug use, neighborhood), etc. The quantitative genetic approaches assumes that behavioral traits are complex and are the end product of cooperative action of individual genes, expressed as a phenotypic class rather than one gene–one disorder (OGOD). Some behavioral anomalies may show co-segregation with DNA markers such as those used in QTL analysis. Some mental anomalies, such as phenylketonuria (single recessive defect in phenylalanine hydroxylation) may account for about 1% of the affliction in mental asylums (►the fragile-X syndrome). Over 100 single gene determined human diseases include mental retardation as part of the syndromes. Defects in the X-chromosomally encoded gene product mitochondrial enzyme, monoamine oxidase A (MAOA) was attributed to violent behavior and also to schizophrenia. MAOA degrades serotonin, dopamine and norepinephrine. ►behavior genetics, ►cognitive abilities, ►human intelligence, ►ethology, ►self-destructive behavior, ►personality, ►MAOA, ►homosexuality, ►aggression, ►autism, ►dyslexia, ►morality, ►instinct, ►cocaine, ►serotonin, ►dopamine, ►norepinephrine, ►heritability, ►QTL, ►determinism, ►differential psychology

Behcet Syndrome (TAP): This rare disorder is characterized by mouth and genital inflammation in humans. It is probably autosomal dominant.

Behr Syndrome: The disorder leads to recessive infantile optical nerve atrophy. ►optic atrophy

BEL: ►*copia*

Bell Curve: ►normal distribution

Bellevalia: This is a subspecies of lilies (2n = 8 or 16) with large and well stainable chromosomes.

Bellophage: Refers to a 1-kb RNA phage, encoding a nucleocapsid protein, a replicase component and an integrase. It has some retroviral-like properties. The small replicase binds to the host DNA polymerase and modifies it in such a way that the enzymes act as an RNA-directed DNA polymerase. Assisted by the integrase, the DNA formed is inserted into the host genome as a prophage. The nucleocapsid, because of its leucine zipper motif, can associate with its helper phage. The helper is originally the *Salmonella* phage Ω but it may recruit for this function adenovirus or influenza virus if mutation alters the leucine zipper. In chickens and some apes the provirus integrates into the mtDNA rather than into the nucleus. Thus it opens the possibilities of inserting foreign DNAs into the mitochondria and chloroplasts for genetic engineering. ►viral vectors, ►transformation of organelles, ►integrase, ►leucine zipper

BEM1: This protein with SH3 domain is involved in signal transduction. ►SH3, ►signal transduction

Bematistes pongei: This is an African butterfly mimicked by *Papilio dardanus*. ►Batesian mimicry

Bence-Jones Protein: Some immunoglobulin heavy chain diseases (HCD) such as the lymphoproliferative neoplasms may only contain the antibody heavy chains (IgM, IgG, IgA), and even those are truncated and are deficient in most parts of the variable region. The γ and α type HCD cells synthesize no light chains but the μ HCD cells secrete an almost normal light chain that is detectable in the urine and it is called Bence-Jones protein. Some of the bone marrow cancer (myeloma) patients also discharge Bence-Jones protein in their urine. These light chain immunoglobulins are generally homogeneous because they are the products of a clone of cancer cells and were historically useful to obtain information on the antibody structure. ►myeloma, ►monoclonal antibody, ►immunoglobulins, ►antibody; Beetham R 2000 Ann Clin Biochem 37(5):563.

B

Beneficial Mutation: The majority of new mutations are less well adapted than the prevailing wild type allele in a particular environment or at best they may be neutral. Beneficial mutations are rare because during the long history of evolution the possible mutations at a locus had been tried and the good ones preserved. Nevertheless, if a new mutation has 0.01 reproductive advantage, the odds against its survival in the first generation is $e^{-1.01} = 0.364$. Its chances to be eliminated by the 127th generation are reduced to 0.973 compared to a neutral mutation that would be eliminated by a chance of 0.985. Even mutations with an exceptionally high selective advantage may have a good chance to be lost ($e^{-2} \cong 0.1353$). Under normal conditions the selective advantage(s) is very small and the chance of ultimate survival is (y) = 2s and the chance of extinction is (l) = 1−2s. In order that the mutation would have more than 50% probability of survival, the requisites must be $(1-2s)n < 0.5$ or $(1-2s) > 2$. Hence,−n *ln*((1−2s) > *ln*2 or approximately −n(−2s) > *ln*2, and therefore n > (*ln*2)/2s or ≅ (0.6931)/2s. If (s) = 0.01 and (n) = number of mutations, (n) must be larger than 0.6931/(2 × 0.01) ≅ 34.66. In other words, at least 35 mutational events must take place with at least 1% selective advantage of the mutants over the wild type that one would ultimately survive. If the rate of mutation is 10^{-6}, nearly a population of 35 million may provide such a mathematical chance. Under evolutionary conditions neutral or even deleterious mutations may make it (succeed) by random drift or chance in small populations. ►mutation neutral, ►mutation rate, ►mutation spontaneous, ►mutation in human populations; Fisher RA 1958 The genetical theory of natural selection, Dover, UK; Dobzhansky T, Spassky B 1947 Evolution 1:191; Miura T, Sonigo P 2001 J Theor Biol 209:497.

Benign Hereditary Chorea: This disorder refers to dominant childhood chorea encoded at 14q. ►chorea

Benton-Davis Plaque Hybridization: This process involves the selection of recombinant bacteriophages on the basis of DNA hybridization with ^{32}P probes on an appropriate (nitrocellulose or nylon) membrane. For the screening of a mammalian or other large library, hundreds of thousands of recombinants need to be screened. In a 150 mm Petri dish 5 × 104 plaques may be used. ►DNA hybridization, ►Grunstein-Hogness screening, ►DNA library, ►recombination molecular mechanisms prokaryotes, ►plaque, ►plaque-forming unit; Benton WD, Davis RW 1977 Science 196:180; Lewis JA et al 1983 Mol Cell Biol 3:1815.

Benzimidazoles: These are tubulin-binding/depolymerizing chemicals used as herbicides (trifluralin, oryzalin) or fungicides (benomyl). The oral dose of LD50 for mouse is ∼2910 mg/kg. ►tubulin

Benzo(a)Pyrene: This is a highly carcinogenic polycyclic hydrocarbon generated by combustion at relatively lower temperatures by polymerization of organic material (see Fig. B29). It is present in automobile emissions, burning of coal, cigarette smoke, fried and grilled meat (in charbroiled T-bone steaks more than 50 µg/kg has been detected, etc.). It has been estimated that 13,000 ton is annually released into the world's atmosphere by these processes. A single 0.2 mg intragastric dose per mouse, resulted in 14 tumors in five of the 11 animals treated. Exposure of the skin and inhalation of the fumes substantiated high and rapid carcinogenicity. It is also a promutagen requiring metabolic activation in *E. coli*, yeast, Drosophila, various rodents and the plant Arabidopsis. Benzo(a) pyrene forms adduct not only with guanine by binding to the N2 position, but also with deoxyadenosine. It also leads to sister chromatid exchange and the formation of micronuclei. Exposure to benzo(a)pyrene results in the expression of cytochrome P450 (cyp1a1) in the skin and liver of mice if the aryl hydrocarbon receptor (AhR) is active. Cyp1a2 gene expression did not need AhR. For carcinogenesis by benzo(a)pyrene AhR is a requisite. ►environmental mutagens, ►carcinogens, ►Ames test, ►bioassays in genetic toxicology, ►sister chromatid exchange, ►micronucleus formation as a bioassay, ►adduct, ►cytochromes; arylhydrocarbon receptor; Chiapperino D et al 2002 J Biol Chem 277:11765.

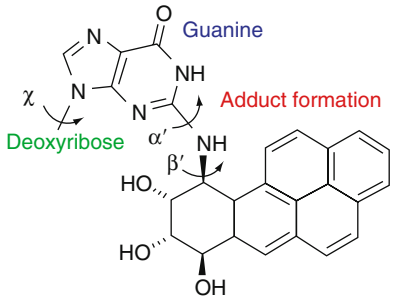

Figure B29. Benzo(a)Pyrene

Benzyladenine (6-benzylaminopurine): ►plant hormones

BER (base excision repair): ►DNA repair, ►excision repair

Berardinelli Disease: ►lipodystrophy familial

Berardinelli-Seip Congenital Lipodystrophy: This condition is characterized by recessive defects in seipin, an integral membrane protein of the endoplasmic reticulum, leading to neurodegeneration. The symptoms are somewhat similar to those of the Silver syndrome as the same region in human chromosome 11 is involved. ►spinal muscular atrophy, ►Silver syndrome, ►lipodystrophy congenital; Windpassinger C et al 2004 Nature Genet 36:271.

Bergamottin: This is one of the several structurally and functionally related derivatives of furanocoumarins affecting +/− drug transport. It is present in citrus fruits (grapefruit juice) and other natural products (see Fig. B30). ►grapefruit; Ohnishi A et al 2000 British J Pharmacol 130:1369.

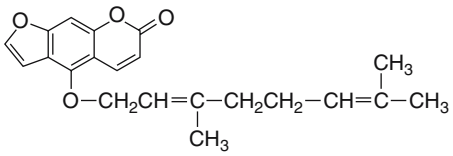

Figure B30. Bergamottin

Bermuda Standard: Refers to the 1997 agreement at the 2nd International Strategy Meeting of the human genome sequencing project that provides quality standards. The goals included accuracy at the 1×10^{-4} level of nucleotides and specifying the principles of various technical levels of operations, e.g., using the PHRED and PHRAP computer programs, and restriction enzymes. It also spells out some principles of etiquette. ►human genome projects, ►PHRAP, ►PHRED; see in AltaVista: http://www.gene.ucl.ac.uk/hugo/bermuda2.htm.

Bernard-Soulier Syndrome: This refers to 22q11.2 and 17pter-p12 recessive dysfunction of the platelets and thrombocytopenia. It is a potentially lethal bleeding disease. A platelet membrane receptor, glycoprotein Ib-IX-V is absent and the platelets do not agglutinate by interaction with the von Willebrand plasma factor. ►thrombocytopenia, ►platelet, ►von Willebrand disease; Ludlow LB et al 1996 J Biol Chem 271:22076.

Bernoulli Process: Independent experiments, which provide only two outcomes, yes or no, success or failure are called Bernoulli trials, and the two-event classes and their probabilities are called Bernoulli process where p = probability of success and $1 - p = q$ = probability of failure. If in a sequence of 10 trials there are 4 successes, the probability of that sequence is p^4q^6; if $p = \frac{2}{3}$ then the probability of the sequence becomes $\left(\frac{2}{3}\right)^4\left(\frac{1}{3}\right)^6$

The general formula becomes:

$$P(r \text{ success}|N,\ p) = \binom{N}{r} p^r\ q^{N-r} \qquad \{1\}$$

where p = probability of success, r = the exact number of successes and N = the number of independent trials. If it is assumed that one can observe a monogenic segregation where the penetrance of the mutant class is reduced from 25% to 20%, then the probability of finding a recessive mutant among 3 individuals will be according to {1}:

$$P(1 \text{ mutant among 3 individuals}) = \binom{3}{1}(0.20)^1(0.80)^2 = \left(\frac{3!}{2!(2-1)!}\right)0.2 \times 0.64 = 0.384$$

If the population is increased to say, 210, the chances of finding a mutant increase to 70. ►binomial probability, ►inference

BERT (background equivalent radiation time): If a diagnostic X-ray exam uses 360 mrem it corresponds to 1 BERT/year (approximate average in the USA). Sources contributing to the natural background (in mrem) are: radon (200), cosmic sources (100), medical treatment (39), consumer and industrial products (11), air travel (6) and nuclear industry (<1). The total may vary, however, from 100 to 600 mrem or even more at certain locations. The general public usually overestimates the risk of the nuclear industry and underestimates that from medical diagnosis and treatment. ►rem, ►radiation hazard assessment, ►cosmic radiation, ►risk

Berylliosis (CBD): This is a granulomatous (nodular inflammation) lung disease among people exposed to beryllium dust. Homozygosity of a rare major histocompatibility allele (MHC) predisposes certain individuals to this disease. ►MHC

Best Disease: ►macular dystrophy

BeT (best hit): This is a feature of orthologous sequences displaying homologies among individual genes (COGs) in different species. ►COG

Beta Barrel: ►barrel

Beta Blocker (beta adrenergic antagonist): This drug reduces blood pressure by slowing down the heart rate.

Beta Breaker Amino Acids: These disrupt beta sheets highly likely (Asp, Glu, Pro) or less frequently (Gly, Lys, Ser). ►protein structure, ►prions; Adessi C et al 2003 J Biol Chem 278:13905.

Beta Complex: Refers to one of the alternately distributed translocation complexes of the plant Oenothera. ►alpha complex alternative, ►multiple translocations, ►complex heterozygote

Beta Distribution: This distribution is very similar to the binomial probability function. This distribution is continuous whereas the binomial distribution is discrete. $f(x) = \frac{x^{\alpha-1}(1-x)^{\beta-1}}{B(\alpha,\beta)}$. ►binomial distribution

Beta Galactosidase: ►galactosidase; ►Lac operon

B

Beta-Lipoprotein (apolipoprotein, 2p24): This is a component of the low-density lipoprotein fraction (LDL) in the plasma. The ApoB-48 fraction is made in the gut, the ApoB-1000 in the liver by differential processing of the transcript of the same locus. ►hypobetalipoproteinemia, ►abetalipoproteinemia, ►hyperbetalipoproteinemia

Beta Particles: These electrons are emitted by radioactive isotopes; their mass is 1/1837 that of a proton. The negatively charged form of it is an electron whereas the positively charged is a proton. Beta particles have no independent existence; they are created at the instance of emission. In the biological laboratory the most commonly used isotopes emitting β radiation (with energy in MEV) are H3 (0.018), C14 (0.155), P32 (1.718), S35 (0.167), I131 (0.600 and 0.300 but emits also γ radiations of various energy levels). The mean length of the path of H3 is about 0.5 µm and that of P32 is about 2600 µm. ►linear energy transfer, ►isotopes

Beta Sheets: ►protein structure

Beta vulgaris: Refers to beets (*Chenopodiaceae*) having basic chromosome number 9. Included in this group are sugar beets, fodder beets, mangold and chards which are all-important food and feed crops. Sugar beets represent a glowing example of the success of selective plant breeding by increasing the sugar content (about 2% in the mid-eighteenth century) by over 10-fold in some modern varieties. The most productive current varieties display triploid heterosis and improved disease resistance and the monogerm "seeds" facilitate mechanization of cultivation, etc. ►heterosis, ►triploid, ►monogerm seed

Betel Nut (*Arecia catechu*): This seed palm tree is used as a stimulant; 2n = 4x = 32.

Bet-Hedging: Sexually mature individuals reduce their reproductive potentials due to environmental circumstances. (Menu F et al 2000 Am Nat 155:724).

Bethlem Myopathy: This is a dominant human disorder involving contractures of the joints, muscular weakness and wasting. It is associated with mutations of collagen type VII genes in human chromosomes 21q22.3 and 2q37. ►collagen, ►laminin

BEV: (Baculovirus expression vector): This is a potential tool to control insect populations by biological means. ►baculovirus, ►biological control, ►viral vectors

bFGF: Refers to the basic fibroblast growth factor.

BFP (blue fluorescent protein): This is similar to GFP (green fluorescent protein). Excitation at 368 nm causes light emission at 445 nm, which excites the Ser65Cis mutant of GFP and causes light emission at 509 nm. ►aequorin, ►EGFP

β-Galactosidase: This enzyme (lactase) splits the disaccharide lactose into galactose and glucose. It can also act on some lactose analogs, e.g., on ONPG (o-nitrophenyl-β-D-galactopyranoside). This substrate (10–3 M or less) when exposed to active enzymes (1010 molecules/mL) yields a yellow product (that has an absorption maximum at 420 nm) and can be used to measure the activity of the enzyme. In cells grown in A medium and with Z buffer the activity of galactosidase is determined by the formula:

$$> 1000 \times \frac{OD_{420} - (1.75xOD_{550})}{tx(0.1xOD_{660})}$$

where OD is optical density at the wavelength indicated, and *t* is time of the reaction run in minutes. On a Petri dish the activity of β-galactosidase is detected on EMB agar (containing eosin yellow, methylene blue and lactose) and in case the sugar is fermented, a dark red color develops. Bacterial galactosidase is an inducible enzyme and induction takes place by allolactose that is formed upon the action of the residual few galactosidase molecules in the non-induced cells. A gratuitous inducer (induces synthesis although the enzyme itself is not a substrate) is isopropyl-β-D-thiogalactoside (IPTG). Constitutive mutants of the *E. coli* z gene can be identified on Xgal media containing 5-bromo-4-chloro-3-indolyl-β-D-galactoside dissolved generally in dimethylformamide (20 mg/mL). This compound is not an inducer of the enzyme but it is cleaved by it and thus a blue indolyl derivative is released. ►Lac operon, ►galactosidase

BGH: ►bovine growth hormone

β-Glucuronidase: ►GUS

BH: ►BAK

BHK: This refers to the baby hamster kidney cell; cultured fibroblasts of the Syrian hamster. ►hamster

bHLH (basic helix-loop-helix protein): ►helix-loop-helix

b/HLH/Z Motif: A basic amino acid sequence at the N terminus which is probably required for DNA binding, helix-loop-helix structure, leucine zipper. This general structure is widely found in biologically active proteins involved in DNA binding. ►binding proteins, ►DNA binding protein domains, ►helix-loop-helix

Bialaphos: This inhibitor is a glutamine synthetase normally produced by *Streptomyces hygroscopicus*.

Upon splitting off two alanine residues it is activated into phosphinotricin. ►herbicides

Biallelic: For gene expression in diploids both alleles must be present. ►allele, Karolinska Institute human bi-allelic sequence: http://www.kisac.ki.se.

Bi-Armed Chromosome: This has two chromatids at the opposite sides of the centromere (see Fig. B31). ►telochromosome, ►chromosome morphology

Figure B31. Bi-armed chromosome

Bias: Denotes an average error of an estimate. It is a false difference of an observation from the correct value.

BIBAC (binary bacterial artificial chromosome): This is a plant genetic expression vector that can be propagated in *Agrobacterium tumefaciens* and *E. coli* and it can deliver to plant chromosomes large (160 kb) foreign DNA sequences. ►BAC, ►Agrobacterium, ►transformation, ►vectors

bicoid (*bcd*, 3–48): This is a maternal effect mutation in *Drosophila* (see Fig. B32). The larvae lack a head, thorax, some abdominal segments and duplicate telsons. In the wild type the *bicoid* mRNA is localized in the anterior part of the egg. The mammalian homologs are Pitx1 and Pitx2. Its protein product is in the cleavage embryos in a decreasing anterior-posterior gradient. The Bcd protein—through its binding region, BBR—interacts with d4EHP (an eIF-4E related protein) binds the cap region of *cad* (*caudal, Drosophila gene at* 2–55) mRNA and prevents its translation (Cho PF et al 2005 Cell 121:411). Caudal mutations are homozygous lethal because of an abnormal segmentation pattern.

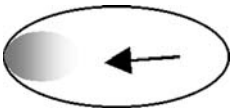

Figure B32. *bicoid*

VPS36 (a component of the ESCRT transport system) functions by binding directly and specifically to stem-loop V of the *bicoid* RNA 3′ UTR through its amino-terminal GLUE domain. VPS36 localizes to the anterior of the oocyte in a *bicoid*-mRNA-dependent manner, and is required for the subsequent recruitment of Staufen (a *Drosophila* RNA-binding protein) to the *bicoid* complex. This function of ESCRT-II as an RNA-binding complex is conserved (Irion U, Johnston D St 2007 Nature [Lond] 445:554). ►left-right asymmetry; Cha B-J et al 2001 Cell 106:36; Houchmandzadeh B et al 2002 Nature [Lond] 415:798.

BID: A pro-apoptotic protein which links proximal signals to the apoptotic pathways (FAS and TNFR). The inactive cytosolic form of BID (p22) is split into the 15-kDa fragment, tBID (truncated BID) that then moves to the mitochondria. Its BH3 domain oligomerizes with BAK and cause mitochondrial dysfunction and the release of cytochrome c. Post-proteolytic *N*-myristoylation triggers the BID-induced apoptosis. When ATM (ataxia telangiectasia mutated) and the related (ATR) kinases phosphorylate BID it can cause cell cycle arrest or by acting on BAX and BAK pro-apoptotic proteins induce apoptosis (Kastan MB 2005 Nature [Lond] 437:1103). One study concluded that Bid has no role in DNA damage or replicative stress-induced apoptosis or cell cycle arrest (Kaufmann T et al 2007 Cell 129:423). ►apoptosis, ►ATM, ►ATR, ►BAK, ►BAX, ►myristic acid, ►Bcl; Wei MC et al 2000 Genes Dev 14:2060; Zha J et al 2000 Science 290:1761.

BiDil (isosorbide dinitrate and hydralazine): This vasodilator drug is manufactured by Nitro Med, Inc (Bedford, MA), and it is particularly effective in heart failure in the case of some Afro-American individuals with ventricular ejection problems. The label of "ethnic/racial drug" is controversial. ►race, ►ethnicity; Wolf SM 2005 Nature Genet 37:789.

Bidirectional Replication: From the replicational origin the replication moves in opposite directions in the DNA (see Fig. B33). ►replication bidirectional, ►replication fork

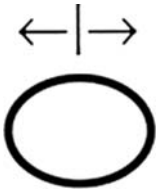

Figure B33. Bidirectional

Bidirectional Gene Organization: Genes in large genomes are not dispersed uniformly but form clusters. Among the 144 and 319 known genes of human chromosomes 21 and 22, 22% and 18%, respectively, are divergently arranged within ~1 kb from each other whereas the average spacing distance for all genes is ~85 kb. In most cases the spacing islands of the bidirectional genes carried a CpG island that

generally overlapped partially or entirely the first exon or rarely more than one. This organization has regulatory advantage. A study of 23,752 human genes revealed that more than 10% of the genes whose transcription start sites are separated by less than 1000 base pairs are transcribed bidirectionally (Trinklein ND et al 2004 Genome Res 14:62). ►operon, ►clustering; Adachi N, Lieber MR 2002 Cell 109:807.

BIDS: ►hair-brain syndrome

Biennial: Refers to plants with a life span of two years yet the total period is less than two years. These plants germinate in fall and mature and die before the end of the following year.

Bifunctional Antibody: This carries an antibody variable region fragment that secures its ability to recognize certain molecules (antigen). In addition, a fused heterologous component conveys either enzyme activity or carries a toxin or a pro-drug, etc. Such a complex can home in on the cognate molecules and destroy or alter them according to the specificity of the heterologous portion. ►bispecific antibody

Bifunctional Enzymes: These have apparently evolved with the potential to adapt the amino and carboxy terminal tracts to fulfill the metabolic requirements of different tissues. ►one gene—one enzyme theorem; Kitzing K et al 2001 J Biol Chem 276:42658.

Bifunctional Mutagen: ►functionality of mutagens

Big Blue: A commercially available mouse strain that carries the bacterial *LacI* gene (in about 40 copies) as a stably integrated lambda vector, thus it is very useful for the laboratory detection of in vivo mutagenic/carcinogenic effects. The transgene is extracted from mice and with the aid of the λ shuttle vector introduced into *E. coli* bacteria. In the presence of Xgal substrate mutations in the *LacI* gene give rise to blue plaques because of the loss of inhibition of the galactosidase gene, *LacZ*. Thus, mutations in the prokaryotic gene in the animals, activated by the animal metabolism, are screened in bacteria because of the convenience of detection in the prokaryotic system. This system permits the detection of mutation in different animal tissues, e.g., liver, brain, at different developmental stages and under different conditions. ►β-galactosidase, ►Xgal, ►Lac operon, ►Muta Mouse, ►host-mediated assay; Gossen JA et al 1989 Proc Natl Acad Sci USA 86:7971; Nohmi T et al 2000 Mutation Res 455:191.

BigSeq: A computer program with a similar purpose as Mask, it contains information on millions of contigs. ►contig, ►physical map

Bikont: This eukaryote has two cilium-bearing centrioles (nucleating cone of microtubules) such as plant and protozoa. Unikonts have one cilium on the centriole such as animals, fungi and amoebazoa. ►cilia, ►centriole; Richards TA, Cavalier-Smith T 2005 Nature [Lond] 436:1113.

Bilateral Symmetry: ►symmetry, ►zygomorphic (see Fig. B33), ►retinoic acid

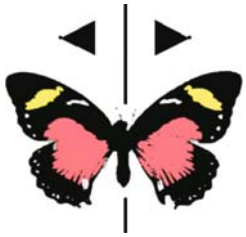

Figure B34. Bilateral symmetry

Bilayer: A bilayer of membranes consists of amphipathic lipids (and proteins) and the non-polar phase faces inward. The majority of cellular membranes are double membranes. ►amphipathic

Bile Salts: These are detergent-type steroid derivatives involved in digestion and absorption of lipids. ►lipids, ►cholesterol; Russel DW 2003 Annu Rev Biochem 72:137.

Bilineality: More than a single locus determines a particular trait and they may segregate independently making chromosomal localization, by genetic techniques, very difficult.

Bilirubin: This bile pigment is formed by the degradation of hemoglobin and other heme containing molecules such as cytochromes. It is circulated in the blood as a complex with albumin and when deposited in the liver it forms bilirubin diglucoronide. It may arise from biliverdin, a breakdown product of heme, through reduction. Bilirubin is a strong antioxidant and may cause brain damage in neonatal jaundice. ►hyperbilirubinemia, ►jaundice, ►cholestasis, ►Alagille syndrome, ►Byler disease, ►steroid dehydrogenase, ►steroid reductase, ►Gilbert syndrome, ►cerebral xanthomatosis, ►Crigler-Najjar syndrome, ►Dubin-Johnson syndrome; Tomaro ML, del C Battle AM 2002 Int J Biochem & Cell Biol 34:216.

Bim: This is a member of the BCL family of proteins. The proteins sharing the Bcl-2 homology domain BH3 promote apoptosis in contrast to other members of the family (e.g., Bax, Bak) that are anti-apoptotic. ►BCL, ►BAX, ►BAK; Chen D, Zhou Q 2004 Proc Natl Acad Sci USA 101:1235.

BimC: This family of motor proteins of the kinesin group is involved in the separation of the mitotic chromosomes by the spindle. ►motor proteins, ►spindle, ►monastrol, ►mitosis

BimD: A negative regulator of the cell cycle progression in *Aspergillus*, it mediates recombination and chromosome morphology.

BimE: This is an *Aspergillus* protein subunit of the APC complex, it is homologous to APC1. ►APC, ►cell cycle

Bimodal Distribution: In this case when the population is represented graphically, it displays two peaks (see Fig. B35). Or, in general, the data are clustered in two modes, in two classes.

Figure B35. Bimodal distribution

BIN: A group of markers (microsatellite DNA) mapped to the same location.

BIN1 (box-dependent Myc interacting protein-1): This is a tumor-suppressor protein (human chromosome 2q14) that interacts with the Myc oncoprotein. It is related to amphiphysin that serves a similar purpose in breast cancer and to the RVS167 cell cycle control gene of yeast. ►MYC, ►tumor suppressor; DuHadaway JB et al 2001 Cancer Res 61:3151.

Bin2 (Cct3, TriC): This is synonymous with the CCT γ chaperonin subunit. ►chaperonins

Bin3 (Cct2): This is synonymous with the CCT β subunit of chaperonins. ►chaperonins

Binary: Refers to any condition, choice or selection with two possibilities or a numeration system with a radix of 2. ►radix, ►founder cells

Binary Variables: These variables are either yes or no (0 or 1); their analysis can be performed by logistic regression or Bernoulli process. ►logistic regression, ►Bernoulli process

Binary Gene Expression: The expression is full or none.

Binary Fission: This means splitting into two parts (see Fig. B35). Bacteria, chloroplasts and mitochondria that do not have a mitotic mechanism reproduce in this way after DNA replication has been completed.

Figure B36. Binary fission

Binary Vector: Refers to *Agrobacterium* carrying two plasmids, one has the T-DNA borders and other sequences (special genes, selectable markers) that will integrate into the transformed cell's chromosomes, the other is a helper plasmid carrying the Ti plasmid virulence genes, required for transfer but no part of the latter plasmid is integrated into the host genome during transformation. ►T-DNA, ►virulence genes of *Agrobacterium*, ►transformation genetic, ►cointegrate vector, ►agrobacterial vectors; Bevan M 1984 Nucleic Acids Res 12:8711.

Binary Targeting: The specific recombinase gene (*Cre, Flp*) is carried by one of the mating pairs and the site-specific recombination site (*loxP, FRT*) is present in the other partner. ►targeting genes, ►*Cre/LoxP*, ►*Flp/FRT*

Binase: This is a 12-kDa dimeric (αβ) endonucleolytic ribonuclease binding to N-1 of 3′-guanine monophosphate. It has 82% amino acid identity with barnase. ►barnase; Wang L et al 2001 Proc Natl Acad Sci USA 98:7684.

BIND (Biomolecular Interaction Network Database): http://www.xml.com/pub/r/1290.

Bindin: An acrosomal protein that mediates the species-specific binding between gametes during fertilization. Apparently the egg surface has a species-specific bindin receptor. ►fertilization, ►acrosomal process, ►sperm, ►acrosome; Glaser RW et al 1999 Biochemistry 38:2560; Kamei N, Glabe CG 2003 Genes Dev 17:2502.

Binding Energy: This is derived from the non-covalent interaction between ligand and receptor, enzyme and substrate. ►ligand, ►receptor

Binding Proteins: These proteins are of a great variety and they control gene expressions at the level of transcription (transcription factors, hormones, heat-shock proteins, etc.). Most of them bind to upstream consensus sequences. The cap-binding proteins regulate the stability of the mRNA. Some of them are transcription termination factors, such as rho in bacteria or Sal I box binding proteins in the mouse. To study their position and function at the DNA level, footprinting or protein microarrays is undertaken (Ho S-W et al 2006 Proc Natl Acad Sci USA 103:9940). An in vitro method for specific and sensitive solution-phase analysis of interactions between proteins and nucleic acids in nuclear extracts is based on the proximity ligation assay. The reagent consumption is very low, and the excellent sensitivity of the assay enables analysis of as few as 1–10 cells. The method appears highly reproducible, quantitative, and in good agreement with both EMSA (electrophoretic mobility shift assay) and predictions obtained by using a motif finding software. This

B

assay can serve as a valuable tool for characterizing in depth the sequence specificity of DNA-binding proteins and for evaluating the effects of polymorphisms in known transcription factor binding sites Gustafsdottir SM 2007 Proc Natl Acad Sci USA 2104:3067.

Some proteins bind to the cellular membranes and control imports and exports, others mediate signal transduction. These proteins may have a combinatorial hierarchy and are thus capable of influencing a multitude of processes in the cell, far in excess of their individual numbers. Proteins may bind to the DNA in a non-specific manner and this property facilitates the binding to specific nucleotide sequences. Non-specific binding—by electrostatic forces—facilitates the recognition of the specific sites much faster than mere diffusion (Kalodimos CG et al 2004 Science 305:386). Gene expression is controlled primarily by proteins binding non-covalently to the DNA. In general, there is no particular specificity of recognition between amino acids and DNA bases yet some data indicate structural preferences (Mandel-Gutfreund Y et al 1995 J Mol Biol 253:370; Kono H, Sarai A 1999 Proteins 35:114). The interaction is the result of hydrogen bonding between polar atoms of the protein and nucleic acid bases. Therefore unnatural amino acid substitutions may alter the binding specificity (Maiti A, Roy S 2005 Nucleic Acids Res 33:5896). The interaction is not based on one to one relation rather a particular amino acid is more frequently distributed around a particular DNA base. Transcription factors recognize multiple genes, generally within families and this feature is conserved through evolution. Highly conserved bases seem to interact more specifically with particular protein residues. The binding may also be affected by changes in conformation due to mutation at a remote amino acid site. Through laboratory design binding proteins can be created for the regulation of gene function, which is of significance in biotechnology (Desjarlais JR, Berg JM 1994 Proc Natl Acad Sci USA 91:11099; King CA, Berg JM 1995 J Mol Biol 252:1). ►transcription factors, ►signal transduction, ►DNA-binding protein domains, ►single-strand binding protein, ►footprinting, ►EMSA, ►protein arrays, ►affinity-directed mass spectrometry, ►UAS, ►methyltransferase DNA, ►protein binding, ►threading, support vector machine based procedure for predictions: Bhardwaj N et al 2005 Nucleic Acids Res 33:6486; Ren B et al 2000 Science 290:2306; Verdine GL, Norman DPG 2003 Annu Rev Biochem 72:337; Sarai A, Kono H 2005 Annu Rev Biophys Biomol Struct 34:379; DNA and RNA binding server: http://bioinformatics.ksu.edu/bindn/, protein–protein binding site prediction: *http://bioportal.weizmann.ac.il/promate/*.

Binet Test: ►human intelligence

Binomial Coefficient: ►binomial probability

Binomial Distribution: A distribution that is useful in genetics for the direct estimation of segregation ratios in the case of dominance by expansion of $(3 + 1)^n$ where n = is the number of heterozygous loci (note: 3 + 1 must not be added). By expansion the binomial becomes:

$$1 \times 3^n + [(n!)/1!(n-1)!] \times 3^{n-1} + [n!/2!(n-2)!] \times 3^{n-2} + \ldots + [n!/(n-1)!] \times 3^{n-(n-1)} + 1 \times 3^{n-n}$$

The *exponent* of a base gives the number of loci with the dominant phenotype, the *power* identifies the frequency of that phenotype, and the *coefficients* show how many times—quadruple, triple, etc.,—dominant phenotypic classes will be expected theoretically. The solution for 4 heterozygous pairs of alleles:

$$\begin{aligned}
&(1 \times 3^4) + \left(\frac{4 \times 3 \times 2 \times 1}{1 \times 3 \times 2 \times 1} \times 3^{4-1}\right) \\
&\quad + \left(\frac{4 \times 3 \times 2 \times 1}{2 \times 1 \times 2 \times 1} \times 3^{4-2}\right) \\
&\quad + \left(\frac{4 \times 3 \times 2 \times 1}{3 \times 2 \times 1 \times 1} \times 3^{4-3}\right) + (1 \times 3^{4-4}) \\
&= (1 \times 3^4) + \left(\frac{24}{6} \times 3^3\right) + \left(\frac{24}{4} \times 3^2\right) \\
&\quad + \left(\frac{24}{6} \times 3^1\right) + (1 \times 3^0) \\
&= (1 \times 81) + (4 \times 27) + (6 \times 9) \\
&\quad + (4 \times 3) + (1 \times 1)
\end{aligned}$$

Translated into genetic language in the case of an Aa Bb Cc Dd heterozygote's F2 progeny the phenotypic classes will be:

81 ABCD, [**27** ABCd, **27** ABcD, **27** AbCD, **27**, aBCD], [**9** ABcd, **9** AbCd, **9** AbcD, **9** aBCd, aBcD, **9** abCD], [**3** Abcd, **3** aBcd, **3** abCd, **3** abcD], [**1** abcd]
or
81: 108 (4 × 27): 54 (6 × 9): 12 (4 × 3): 1

For the calculation of genotypic classes among the segregants see trinomials and multinomials. ►Mendelian segregation, ►Pascal triangle

Binomial Nomenclature: ►taxonomy

Binomial Probability: P is the complete binomial probability function whereas the $n!/(x!(n\text{-}x)!$ is the binomial coefficient, an integer that shows how many ways one can have x combinations of n, and $p = 0.75$ and $q = 0.25$ (because of the 3:1 segregation). In genetic experiments this shows—if we have

n = independently segregating gene loci, and the inheritance is dominant—how many ways can have x combinations of n; e.g., if we deal with $n = 5$ loci and we wish to know the chance that at 3 (=x) loci the dominant phenotype would appear is then:

$$p = \binom{n}{x} p^x q^{(n-x)} \text{ and } \binom{n}{x} = \frac{n!}{x!(n-x!)}$$

$$[5!/(3!2!)] = (0.75^3)(0.25^2) \cong 0.26367$$

The binomial distribution is obtained from the expansion of the binomial terms $(p + q)^n$; its standard deviation $\sigma = \sqrt{\frac{pq}{n}}$ and $p + q = 1$; n = is the exponent. ►Pascal triangle, ►transmission disequilibrium, ►binomial distribution, ►trinomial distribution, ►Bernoulli process

Binuclear Zinc Cluster: A domain of a DNA-binding transcriptional activator containing 2 Zinc ions about 3.5 Å apart and regulated by 6 cysteines. ►Zinc finger

Bioarrays: Methods/software systems for analysis of microarray hybridization and other high throughput platforms. (Troein C et al 2006 Methods Enzymol 411:99).

Bioassays (biological assays): Used for determining the biological effect(s) of chemicals, drugs or any other factor on live animals, plants, microorganisms and cells.

Bioassays in Genetic Toxicology: Bioassays have been designed to assess mutagenic (and indirectly carcinogenic) properties of factors that human, animal, plant and microbial populations may be exposed to. Their range varies from testing chromosome breakage and point mutations in a wide variety of organisms using different endpoints. All the different procedures cannot be discussed or even enumerated here but the major types of tests include: (i) excision repair, (ii) reversion studies in *Salmonella* and *E. coli*, (iii) sister chromatid exchange, (iv) mitotic recombination, (v) host-mediated assays, (vi) specific locus mutation assays, (vii) micronuclei formation, (viii) chromosome breakage, (ix) sex-linked lethal assays, (x) unscheduled DNA synthesis, (xi) sperm morphology studies, (xii) cell transformation assays, (xiii) dominant mutation,(xiv) somatic mutation detection, (xv) *Arabidopsis* mutagen assays, (xvi) human mutagenic assays, (xvii) mitotic recombination as a bioassay in genetic toxicology, (xviii) *Tradescantia* stamen hair somatic mutation assay is popular for monitoring environmental pollution, (xix) zebrafish assay for mutagens in aquatic media, (xx) mouse lymphoma test. (see the essential features of these tests under the separate entries, ►transgene mutation assay, ►mutation detection, ►hemiclonal, ►genotoxic chemicals, ►Big Blue ►MutaTM Mouse, ►toxicogenomics; Waters MD, Fostel JM 2004 Nature Rev Genet 5:936.

Bioavailability: The portion or fraction of drugs applied that can be accessed by living cells.

Biobank: A repository that is supposed to collect and maintain information on gene–environment relations in disease, based on large population studies.

Bio-Bar Code, Nano-Particle Based: ►nanoparticle-based bio-bar code

Biocatalysts: Enzymes mediating metabolic processes. They have numerous industrial applications and with the aid of molecular biotechnology means exist for improvement of their efficiency. (Burton SG et al 2002 Nature Biotechnol 20:37).

Biochemical Engineering: Synthesizes proteins and other new molecules in the laboratory without the direct involvement of the classical biosynthetic pathway. The products may have biological value in nutrition, therapeutics, etc. ►antibody engineering, ►nanotechnology, ►bioreactor, ►tissue engineering; unnatural amino acids.

Biochemical Genetics: Studies the genetic mechanisms involved in the determination and control of metabolic pathways. ►inborn errors of metabolism

Biochemical Mutant: The chemical basis of the mutant function is identified. ►auxotroph

Biochemical Pathway: The chemical steps involved in a biological function are represented in a sequence. Enzymes encoded by separate genes usually mediate the individual steps. ►One gene–one enzyme theorem, ►genetic networks, ►transcriptome, Rison SC, Thornton JM 2002 Current Opin Struct Biol 12:374.

Biochips: ►DNA chips, ►protein chips, ►electrical biochip

Biocoenosis (biocenosis): Different organisms living together within the same environment; some of them may be dependent on others for survival or interact in various ways.

Biocomputer: Use approaches different from the digital electronic computing. The input, output software and the hardware are biological molecules without electronic representation. This approach holds promise for the identification of disease conditions. ►DNA computer; Adar R et al Proc Natl Acad Sci USA 101:9960.

Bioconductor: Software for genome analysis (http://www.bioconductor.org).

Biocrystalization: DNA may be protected in prokaryotes by co-crystallization with the stress-induced protein Dps. Dps dodecamers protect against

oxidative damage (and nucleases) in a manner similar to ferritins. ►ferritin

BioCyc: A collection of a set of 160 pathway/genome databases (PGDB) based on the MetaCyc database. ►MetaCyc, ►Pathway Tools, Karp PD et al 2005 Nucleic Acids Res 33:6083; 142 databases: http://biocyc.org; http:/www.org/open-compounds.shtml, pathway tools for downloading: http://biocyc.org/download.shtml.

Biodegradation: Decomposition, destruction of substances by bacterial or other organisms. ►oil spills, ►Pseudomonas, ►bioremediation; http://umbbd.ahc.umn.edu/.

Biodiversity: See species extant: http://www.sp2000.org/; http://ip30.eti.uva.nl/BIS/index.php.

Bioengineering: The replacement of body parts by means of mechanical or biological manufactured devices. (Science 2002 vol 295:998–1031).

Bioethics: The application of ethical principles to biotechnology, medical, genetic and related fields. (►biotechnology, ►embryo research, ►nuclear transplantation, ►ethics, ►informed consent, ►human subjects; Merz JF et al 2002 Am J Hum Genet 70:965; http://www.bioethics.net; http://www.hhmi.org/bioethics); http://www.ornl.gov/sci/techresources/Human_Genome/elsi/elsi.shtml; http://www.ornl.gov/hgmis/elsi/elsi.html; http://www.nhgri.nih.gov/ELSI/.

Biofilm: A community of single-cell organisms established as a surface layer with chemical communication among the components. Bacterial aggregates of single or dozens of different species (including also fungi) surrounded by foamy substance and resistant to many types of disinfectants, antibiotics or to antibodies. Within the biofilm, the bacteria may show morphological differentiation depending on actual environment. Also in these cells, different mutations may arise (Kolter R, Greenberg EP 2006 Nature [Lond] 441:300). Aminoglycoside antibiotics can induce biofilm formation and the bacteria become resistant to antibiotics. It appears that an inner membrane phosphodiesterase with substrate for cyclic di-guanosine monophosphate regulates surface adhesiveness and guanosine triphosphate, an inhibitor of this enzyme, reduces biofilm formation (Hoffman LR et al 2005 Nature [Lond] 346:1171). Cells within the biofilm differentiate differently than free-living bacteria. Polymer production suffocates neighboring non-polymer producers and it suggests that polymer secretion provides a strong competitive advantage to cell lineages within mixed-genotype biofilms and global cooperation is not required (Xavier JB, Foster KR 2007 Proc Natl Acad Sci USA 104:876). Engineered bacteriophage T7 containing a biofilm- and *E. coli*-degrading enzyme eliminates the bacteria ~4.5 orders of magnitude more effectively than non-enzymatic procedures (Lu TK, Collins JJ 2007 Proc Natl Acad Sci USA 104:11197). The polysaccharide (alginate) film may corrode pipes, medical equipment and may be responsible for dental plaques, lung, kidney, prostate, etc. infections and inflammation. The development of the biofilm usually requires quorum-sensing signals. Some fungi (*Candida albicans*) can also form biofilm. ►algin, ►quorum-sensing, ►cystic fibrosis, ►antibiotics, ►antibiotic resistance, ►aminoglycosides; O'Toole G et al 2000 Annu Rev Microbiol 54:49; Whitely M et al 2001 Nature [Lond] 413:860.

Bio-Gel: A commercial ion exchange chromatography medium suitable for the separation of RNA from DNA, purification of oligonucleotides, linkers, etc.

Biogenesis: The cells arise only from cells rather than from non-living organic material. ►spontaneous generation

Biohazards: Working with pathogenic organisms or transgenic material containing potentially dangerous genes (coding for toxins). Containment (P1 to P4, the latter the most stringent) is necessary and the appropriate safety regulations must be complied with. Transgenic plants may transmit by cross-pollination the nuclear transgene (e.g., herbicide or antibiotic resistance) to other plants, including weeds. Some of this unwanted transmission problems can be prevented by using transgenic chloroplasts or mitochondria that are only maternally transmitted (Danielle H 2007 Proc Natl Acad Sci USA 104:6879). Information for particular cases can be obtained from the local biohazard committees or from National Institute of Health, Building 31, Room A452, Bethesda, MD 20205, USA. ►biological containment, ►recombinant DNA and biohazards, ►laboratory safety, ►GMO

Bioinformatics: The use of computers for developing algorithms, information gathering, storage and analysis of molecular biological data. ►GenBank, ►NCBI, ►GSDB, ►databases, ►image analyzer, Basset DE Jr et al Nature Genet Suppl Vol 21; Searls DB 2000 Annu Rev Genomics Hum Genet 1:251; Jenssen TK et al 2001 Nature Genet 28:21; Letovsky S (Ed) 1999 Bioinformatics. Databases and Systems 1999 Kluwer, Boston; Davidson D, Baldock R 2001 Nature Rev Genet 2:409; Yandell MD, Majoros WH 2002 Nature Rev Genet 3:601; Kanehisa M, Bork P 2003 Nature Genet 33 (Suppl.):305, http://www.ncbi.nlm.nih.gov/Tools/index.html, macromolecular structure, function, taxonomy, sequences: http://www.ebi.ac.uk/msd-srv/docs/sifts, bioinformatics assistance: http://www.ebi.ac.uk/2can/home.html, HSLS Online Bioinformatics Resources Collection (OBRC)

contains annotated information and guided links to 1542 open sources bioinformatics databases and software tools: http://www.hsls.pitt.edu/guides/genetics/obrc.

Biolistic Transformation: It (biological-ballistic) introduces genes into the nuclei of cells (of the germline) by shooting DNA coated particles into the target cells, propelled by high-power air or helium guns. It is a most useful procedure when other methods of transformation are not sufficiently successful. It can accomplish transformation also in terminally differentiated cells and chloroplasts and mitochondria. High efficiency mitochondrial transformation (100–250 transformants µg DNA) of *Chlamydomonas reinhardtii* with linearized plasmid DNA is feasible. Gene guns are also used for cancer gene therapy and genetic immunization. ►Transformation, ►chloroplast genetics, ►mitochondrial genetics, ►cancer gene therapy, ►immunization genetic, ►*Chlamydomonas*; Klein TM et al 1987 Nature [Lond] 327:70; Maenpaa P et al 1999 Mol Biotechnol 13:67.

Biological Clock: Frequently called circadian rhythm; it measures in various organisms daily periods and responses to alternation of daily light and dark cycles. The endogenous rhythms also influence gene activity and developmental patterns. ►circadian; http://www.cbt.virginia.edu.

Biological Containment: The preventive measures to avoid the spread of potentially hazardous organism outside the laboratory. Recombinant DNA-containing organisms with unknown biological impact in the environment may be prevented from spreading accidentally, by using transformation vectors that lack the *bom* and *nic* sites facilitating plasmid mobilization. The cloning bacteria may have an absolute requirement for diaminopimelic acid (lysine precursor), deficient in excision repair (*uvrB* deletion), auxotrophic for thymidine and *rec*$^-$ (recombination and repair deficient). Thus even after accidental escape, assuming a mutation rate of 10^{-6} for each of the 5 loci, they would require $(10^{-6})^5 = 10^{-30}$ chance to succeed in the environment. 10^{30} *E. coli* bacterial cell number has a mass of about 10^{11} metric tons. The mass of the Earth has been estimated to be 10^{20} tons. ►biohazards

Biological Control: Pathogens or parasites are contained by propagation of their natural enemies or by other pathogens or parasites (e.g., *Aphelinus mali*) or genetically engineered organisms. *Colletotrichum truncatum* fungus is used as weed killer bioherbicide, *Sesbania exaltata,* in various crops such as soybeans, rice and cotton. Attack of plants by armyworm (*Spodoptera exigua*) is concomitant with the oral secretion of *N*-(17-hydroxylinolenoyl)-L-glutamine [volicitin], which triggers the emission of chemical signals by the plants that attract parasitic wasps, predators of armyworm larvae (see Fig. B37). In some instances, the parasitic wasps are attacked by bacterial symbionts of the host and the presence of the latter may also adversely affect the host's fecundity and longevity. The parasitic wasp *Trichogramma brassicae* spies on mated *Pieris brassicae* butterflies and it is attracted to the pheromone passed from the male to the female to discourage conspecific males. The small wasp (0.5 mm long) then rides on the female and when she lays eggs, the wasp parasitizes them. It is interesting that the wasps show no interest in virgin female butterflies (Fatouros NE et al 2005 Nature [Lond] 433:704). When their roots are attacked by the weevil *Diabrotica virgifera*, wild ancestors and European varieties of maize emit the sesquiterpenoid (E)-β-caryophyllene (see Fig. B38), an attractant for the entomopathogenic nematode, *Heterorhabditis megidis* and it reduces the herbivorous insect population to about half (Rasmann S et al 2005 Nature [Lond] 434:732). Although biological control is frequently considered as the safest method of protection, some biologists worry about the general environmental impact. Some of the control organisms may adversely affect useful native species. ►*Bacillus thüringiensis*, ►genetic sterilization, ►Dengue virus, ►BEV, ►antisense RNA, ►conspecific, ►parasitoid, ►aphids; Howarth FG 1991 Annu Rev Entomol 36:485; Pemberton RW, Strong DR 2000 Science 290:1896; Louda SM et al 2003 Annu Rev Entomol 48:365, http://www.nhm.ac.uk/entomology/chalcidoids; http://ipmworld.umn.edu/.

Figure B37. There are many types of parasitic wasps. The female insect has a long egg depository tube used for placing their eggs into the eggs of another insect

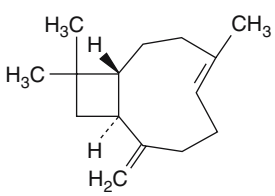

Figure B38. Caryophyllene

Biological Membranes: ►cell membranes

B

Biological Mutagens: A large number of natural products present in different organisms may be mutagenic for others. Spontaneous mutation may be also increased by endogenous factors such as defective DNA polymerase or defects in the genetic repair system. ►mutator genes, ►transposable elements, ►transposons, ►insertional mutation, ►epigenetics

Biological Systems: ►systems biology

Biological Weapons: It may contain highly pathogenic microorganisms such as *Bacillus anthracis* (anthrax), *Corynebacterium diphtheriae* (diphtheria), *Pasteurella pestis/Yersinia pestis* (bubonic plague), *Francisella tularensis* (tularemia), and various viruses. The *Clostridium botulinum* toxin or castor bean toxin, ricin may also be very dangerous. (The Botox toxin has been used recently for the very questionable cosmetic purpose to reduce facial wrinkles by immobilizing muscles through blocking of acetylcholine at the ends of the motor nerves.) In case the genetic signature of all organisms potentially usable for biological warfare would be available, the rapid identification and effective protection may be facilitated.

The agents are categorized as A if they constitute a national hazard by their easy transmission, have major health impact, possibly increase death rates, and they potentially impacts society to a serious extent. Category B agents constitute somewhat reduced hazards, yet require enhanced surveillance and diagnostic capacities. Category C agents include new pathogens that can be modified for higher disease effectiveness and are potentially hazardous. Research on these agents requires special containment facilities. The availability of genomic sequences of the pathogens facilitates the identification of the organisms and facilitates taking effective defensive measures. ►signature of molecules, ►anthrax, ►*Yersinia*, ►tularemia, ►diphtheria toxin, ►Pox virus, ►*Clostridium botulinum*, ►ricin, ►*Francisella*, ►sarin; Stone R 2001 Science 293:414; Kortepeter MG et al 2001 J Env Health 63(6):21; Hawley RJ, Eitzen EM Jr 2001 Annu Rev Microbiol 55:235; Fraser CM 2004 Nature Rev Genet 5:23, http://www.virology.net/garryfavwebbw.html.

Biologics: The material is of biological nature, the processing is biological, the quality of the product is determined by biological methods.

Bioluminescence: ►luciferase, ►aequorin

Biomarker: Any product of the body (e.g., a metabolite) that may respond to adverse environmental effects (carcinogens, mutagens, etc.) or that may be specific for a biological condition, e.g., cancer, other disease, developmental change or any other function. Biomarkers are important diagnostic and prognostic aids in health and disease. Identification of good biomarkers may facilitate the design of appropriate therapy at a particular stage of a disease. It may help to reveal effective drug targets. If good biomarkers are available, the most likely positive responses may be predicted and unresponsive individuals can be spared from the potential side effects of treatments. Biomarker availability is expected to meet the needs of personalized therapy. Unfortunately, only few effective biomarkers are available for clinical guidance. Gleevec, a tyrosine kinase inhibitor, is a very effective new class of drug against chronic myelogenous leukemia. Gefitinib, also a tyrosine kinase inhibitor capable of blocking epidermal growth factor receptor (EGFR) is used for treating non-small cell lung cancer. Japanese patients respond better to this drug because of differences in missense mutations in those populations compared to people of European descent. Trastuzumab/Herceptin is a monoclonal antibody targeting primarily a tyrosine kinase receptor encoded at 17q12 and involved in certain subtypes of breast cancer. Unfortunately, multiple genes affect the majority of cancers. Therefore only genomic analysis may provide satisfactory approach. Such studies can provide biological signatures for the disease and treatment and are in the frontline of research. The outcome of the research may be affected by genetic, developmental and environmental conditions, the methodology used (proteomics or RNA microarrays) and the statistical analysis employed (t-test or multivariate). (see Dalton WS, Friend SH 2006 Science 312:1165; ►Gleevec, ►Gefitinib, ►Iressa, ►Herceptin, ►Avastin, ►Trastuzumab, ►cancer, ►genetic medicine, ►receptor tyrosine kinases, ►Desatinib, ►Sutent, ►Lepatinib, ►biomarkers in development: http://www.imgenex.com/emarketing/083106_Tissuearray/elucidating_protein_ signatures.htm).

Biomaterial: Generally synthetic substances—other than drugs—useful for biological and/or medical devices such as tissue replacement, gene delivery vehicles, diagnostic and array technologies. (see Langer R, Tirrell DA 2004 Nature [Lond] 428:487).

Biometric: An electronic code of human physical features (fingerprints, eye iris scans), and can be used for digital personal identification.

Biometry: Mathematical statistical principles applicable to the study of genetic and non-genetic variation in biology. ►quantitative genetics, ►population genetics

Biomimetic Particles: The system is based on a peptide that recognizes clotted plasma proteins and selectively homes to tumors, where it binds to vessel walls

and tumor stroma. Iron oxide nanoparticles and liposomes coated with this tumor-homing peptide accumulate in tumor vessels, where they induce additional local clotting, and thereby producing new binding sites for more particles. The system mimics platelets, which also circulate freely but accumulate at a diseased site and amplify their own accumulation at that site. The self-amplifying homing is a novel function for nanoparticles. The system enhances tumor imaging, and the addition of a drug carrier function might also be exploited (Simberg D et al 2007 Proc Natl Acad Sci USA 104:932). ►nanoparticles

Biomining: Certain bacteria obtain energy by oxidizing inorganic materials. This process may release acid, which in turn can wash out metals from ores. ►*Thiobacillus ferrooxidans* can release copper and gold, ►*Pseudomonas cepacia* may assist phosphate mining. Eventually this biotechnology may become economical, especially for low-grade ores. Some plants, e.g., Brassica spp, Impatiens spp. may accumulate gold from the soil. ►Bioremediation; Mergeay M 1991 Trends Biotechnol 9:17; Guiliani N, Jerez CA 2000 Appl Environ Microbiol 66:2318.

Biomonitoring: Surveying potential mutagens, carcinogens or other health hazards using biological means such as organismal bio-assays for mutagens, blood cells, human buccal cells, nasal mucosal cells, scalp hair follicles, sputum, detached colon cells, cervical epithelia, exfoliated bladder cells, spermatozoa, bacteria, etc. The tests may involve cytological or molecular methods. ►quantum dots

Bionics: Construction of mechanical devices with the technology of engineering and biology.

Biopanning: The selection by repeated cycles for specific peptides (phage display), interactive ligands, etc. ►phage display, ►ligand; Giordano RJ et al 2001 Nature Med 7:1249; Shadidi M, Sioud M 2004 Methods Mol Biol 252:569.

Biopharming: Producing pharmacological agents by plants and animal transgenic for special genes. ►pharming, ►plant vaccines, ►single-chain Fv fragment; Ma JK-C et al 2003 Nature Rev Genet 4:794.

Biophilia: A hypothesis suggesting an innate human tendency to focus on life and life-like processes and interest in living beings as proposed by E.O. Wilson.

Biophore: A hypothesized hereditary unit of the pre-mendelian era. ►pangenesis

Biophore: A compound or structural element of it that may exert potential biological (carcinogen) activity. ►SAR, ►CASE, ►MULTICASE

Biophysics: The theory and practice of application of physical methods for the study of biological structures (e.g., nucleic acids, proteins) and mechanisms of function (energy conversions, thermodynamics). (http://www.biophysics.org/education/).

Biopiracy: The use of human genomic information for commercial purposes without the informed knowledge or consent of the individuals, or the sources of the DNA. ►informed consent

Biopoesis: The evolution of living cells from chemical substances rather than from other cells. ►evolution prebiotic, ►origin of life

Bioprospecting: Searching for natural products (genes) potentially useful for pharmaceutical or agricultural applications. ►biotechnology

Biopterin: A pterin derived co-factor of enzymes functioning in oxidation-reduction processes (see Fig. B39)

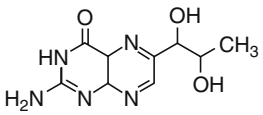

Figure B39. Biopterin

Bioreactor: A large-scale (industrial) culture of cells for the purpose of production and extraction of pharmaceuticals, enzymes, polypeptides, biodegradable plastics, etc. The use of transgenic organisms extended the range of utility of these procedures. Some constructions simulate conditions for growth in outer space. *Flow bioreactors* provide a continuous supply of fresh nutrients. In vitro operations may produce human organs, such a bones for therapeutic implantations. Skin and cartilage can already be proliferated outside the body for medical use. ►cell culture, ►chemostat, ►tissue culture, ►transgenic; Baoudreault R, Armstrong DW 1988 Trends Biotechnol 6:91.

Bioreactor: A collaborative software creation initiative for data acquisition, data management, data transformation, data modeling, combining different data sources, making use of evolving machine learning methods, and developing new modeling strategies suitable for computational biology and bioinformatics (Gentlemen RC et al 2004 Genome Biol 5:R80; http://genomebiology.com/2004/5/10/R80).

Bioremediation: A procedure of adding organisms to an environment for the purpose of promoting degradation of harmful or undesirable properties of that environment. Some observations indicate that 44 ±18% of polycyclic hydrocarbons of the atmosphere are captured by the vegetation and eventually

incorporated into the soil. Many polycyclic hydrocarbons are carcinogenic and mutagenic and pose serious health hazards to people and animals. Their removal from the atmosphere is desirable, however, it is not clear what is the consequence of eating plants that absorbed these semi-volatile compounds. Several organic compounds can be degraded by sequential exposure to anaerobic and aerobic bacteria. Bacterial mercuric ion reductase gene, in a re-engineered form, has been introduced into *Arabidopsis* plants by transformation and the transgenic plants became resistant to $HgCl_2$ and to Au^{3+}. The transgenic plants evolved substantial amounts of Hg^0 (vapors). *Arabidopsis* plants expressing the yeast gene YCF1 accumulate greater amounts of cadmium and lead and display greater resistance to these toxic metals (Song W-Y et al 2003 Nature Biotechnol 21:914). Plants can extract toxic substances from the soil (phytoextraction) and from water (rhizofiltration), thus facilitate the cleaning up of the environment. By a technique of genetic engineering cytochrome P450 monooxygenase genes can be combined with toluene dioxygenase genes in e.g., *Pseudomonas*. Such bacteria then can degrade polyhalogenated compounds such as 1,1,1,2-tetrachloroethane (a powerful narcotic and liver poison) to 1,1-dichloroethylene and eventually to formic and glyoxylic acids, which are still irritants but occur in natural products of ants and fruits, respectively, but do not pose serious threat at low concentrations. *Dehalococcoides ethenogenes* can degrade vinyl chloride to non-toxic ethane. ►environmental mutagens, ►biomining, ►biodegradation, ►soil remediation; Lovley DR 2001 Science 293:1444; Kramer U, Chardonnens AN 2001 Appl Microbiol Biotechnol 55:661; He J et al 2003 Nature [Lond] 424:62; phytoremediation: Pilon-Smits E 2005 Annu Rev Plant Biol 56:15; http://pdg.cnb.uam.es/MetaRouter; http://www.pdg.cnb.uam.es/biodeg_net/MetaRouter.

Bi-Orientation: The sister kinetochores are attached to spindle fibers that connect them to the opposite spindle poles. ►coorientation, ►spindle fibers, ►microtubule, ►kinetochore; Tanaka TU 2002 Current Opin Cell Biol 14:365.

Biosemiotics: The discipline of communication within and among biological systems.

Biosensors: They analyze macromolecular interactions in real time in intact cells. Among the different systems ligand-receptor binding and signal transduction pathways may be the most sensitive, especially when coupled to fluorescent stains. ►ligand, ►signal transduction, ►fluorochromes, ►aequorin, ►surface plasmon resonance; Aravanis AM et al 2001 Biosens Bioelectron 16:571; engineered protein biosensor: Kohn JE, Plaxco KW 2005 Proc Natl Acad Sci USA 102:10841.

Bioseq: It contains relevant information about a biological sequence beyond what is included in the ASN.1. ►ASN.1, ►gi, ►accession number

Biosphere: The range of habitat of organisms living in and on the soil, in bodies of water and the atmosphere.

Biostratigraphy: The relative dating of the succession of different evolutionary forms of organisms on the basis the paleontological relics.

Bisulfite Reaction: Sodium bisulfite is a mutagen inducing point mutations and chromosomal aberrations. The bisulfite reaction permits also the distinction between cytosine and methylcytosine. In bisulfite-treated single-stranded DNA cytosine is converted into uracil (*bisulfite conversion*) but methylated cytosine is essentially non-reactive. The chemically modified DNA tract can then be amplified by PCR and sequenced to determine the location of the methylated base. A procedure has been worked out to assess the extent of DNA methylation (involved in epigenetic modification), applicable genome-wide (Meissner A et al 2005 Nucleic Acids Res 33:5868). ►methylation-specific PCR, ►methylation of DNA, ►epigenesis; Sasaki M et al 2003 Biochem Biophys Res Commun 309:305.

Biosynthesis: Synthesis of molecules by living cells.

Biota: The community of all living organisms in an environment.

Biotechnology: The purposeful application of biological principles to industrial, medical and agricultural production such as molecular alteration of enzymes, cloned recombinant DNA and its translated products (e.g., human insulin produced by transgenic bacteria), replacement of defective genes by site-specific recombination, gene medicine (introducing transiently into cells genes capable of producing the medication required), transfer desirable genes into domestic animals and plants by genetic transformation to improve their economic value, clean up environmental pollutants by modified microorganism capable of digesting crude oil, etc. The economic importance of the biotechnology industry is indicated by their total value of $224 billion in 2002, and it employed more than 194,000 people (Rasnick D 2003 Nature Biotechnol 21:355). ►Genetic engineering, ►tissue engineering, ►bioethics, ►bioprospecting, ►genomics, ►GMO; applications to public health: Daar AS et al 2002 Nature Genet 32:229, approval by different social groups: Gaskell G et al 2005 Science 310:1908, agricultural biotech: http://www.cid.harvard.edu/cidbiotech/homepage.htm.

Bioterrorism: It produces fear and harm among selected individuals or in the general population or harm plants, animals or the environment with the use of agents like bacteria, viruses, fungi or toxins derived from biological agents. Mechanical detection of explosive devices carried by suicide bombers lack sufficient sensitivity for detection beyond distances of 10 meter and are too bulky and expensive for soft targets (Kaplan EH, Kress M 2005 Proc Natl Acad Sci USA 102:10399). ►Biological weapons, ►pathogen identification; Henderson DA 1999 Science 283:1279; Atlas RM 2002 Annu Rev Microbiol 56:167; Fauci AS 2003 Nature [Lond] 421:787; mathematical modeling of food supply poisoning: Wein LM, Liu Y 2005 Proc Natl Acad Sci USA 102:9984, http://www.health.gov/nhic/Scripts/Hitlist.cfm?Keyword=Bioterrorism, toxins, virulence factors, antibiotics for biodefense: http://mvirdb.llnl.gov/.

Biotic: Related to living organisms.

Biotidinase Deficiency (same as multiple carboxylase deficiency, 21q22.1, 3p25): An autosomal recessive disease yet the heterozygotes may be identified, however, by much less obvious symptoms. The biochemical basis is a deficiency of an enzyme (multiple carboxylase) that splits biocytin (biotin—ε-lysine) and thus generates free biotin from protein linkages. The symptoms that may have late onset or appear in neonates are hypotonia (reduced tension of muscles), ataxia (reduced coordination of the muscles), neurological deficiencies (hearing, vision), alopecia (baldness), skin rash, susceptibility to infections, etc. Generally, administration of biotin alleviates the symptoms and may restore normality. The prevalence varies within the 10^{-5} range. Simple procedure is available for the testing of blood by color on filter paper, without purification. ►genetic screening, ►biotin

Biotin: A vitamin, a mobile carrier of activated CO_2, its major biological role involves pyruvate carboxylase (see Fig. B40). It combines with avidin and thus used for non-radioactive labeling. ►non-radioactive labeling, ►fluorochromes, ►biotinylation; Mardach R et al 2002 J Clin Invest 109:1617.

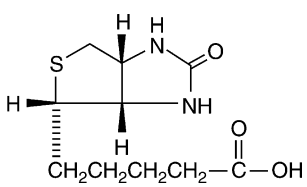

Figure B40. Biotin

Biotinylation: A very sensitive, non-radioactive labeling generated by incorporation into the DNA, with the aid of nick translation, biotinylated deoxyuridylic or deoxyadenylic acid. Biotin in the DNA has great affinity for streptavidin carrying a dye marker and the labeled DNA can thus be identified in light either cytologically or on membrane filters. ►biotin, ►fluorochromes, ►labeling, ►FISH; Demidov VV et al 2000 Curr Issues Mol Biol 2:31.

Biotope: A small, uniform ecological niche.

Biotrophpic: A parasite living on live host. ►saprophytic

Biotype: Physiologically distinct race within a species.

BiP: A soluble heat-shock protein 70, a chaperone. It is an immunoglobulin-binding protein. ►heat-shock proteins, ►Sps70, ►chaperone

Biparental Inheritance: The female and male parents transmit the nuclear genes (see Fig. B41), in contrast, cytoplasmic organelles (and their genetic material) are most commonly inherited only through the egg, and therefore, the inheritance is uniparental (through the female). ►allophenic, ►mtDNA, ►mitochondrial genetics, ►chloroplast genetics, ►meiotic drive

Figure B41. Bride & groom, ceramics By Margit Kovács

Bipedal: Animals walking on two feet as an evolutionarily developed characteristic.

Bipolar Mood Disorder: A complex human disorder involving manic depression fluctuating with euphoria. Putative genetic determinants have been found in human chromosomes 1p33-p36, 2p, 2q21-q33, 3p14, 3p21, 3q26-q27, 4p15.3-p16.1, 5p15, 6q21-q22, 8q24, 8p21, 10q25-q26, 7, 13q11, 13q31-q34, 14q12-q13, 15, 16, 17q and 18, 21q22. Apparently, a major factor associated with human chromosome

22q12 is XBP1 (X box-binding protein transcription factor controlling class II major histocompatibility complex antigens). It involves the control of a heat-shock protein (HSPA5) mediating protein folding in the endoplasmic reticulum (Kakiuchi C et al 2003 Nature Genet 35:171).

By all evidence, several genes contribute to bipolar disorders (Dick DM et al 2003 Am J Hum Genet 73:107; also corrections ibid. 73:979). Prevalence of the various manifestations during a lifetime is about 1%. Among artistically creative persons, Hemingway, Gogol, Strindberg, Byron, Goethe, van Gogh, Goya, Donizetti, Handel, Klemperer, Mahler, Schumann and others suffered from this illness to a variable degree (Janka Z 2004 Orvosi Hetilap [in Hungarian]: 145:1709). ► affective disorders, ►depression, ►manic depression, ►lithium, ►unipolar depression, ►psychoses, ►HLA, ►QTL, ►GSMA, ►reelin; Kelsoe JR et al 2001 Proc Natl Acad Sci USA 98:585; Cichon S et al 2001 Hum Mol Genet 10:2933; Mitchell PB, Malhi GS 2002 Annu Rev Med 53:173; Segurado R et al 2003 Am J Hum Genet 73:49, review: Belmaker RH 2004 New England J Med 351:476.

Bipolarity: Both strands of the DNA are transcribed in opposite $\overrightarrow{\ }_{\leftarrow}$ directions.

BIR (chromosome break-induced replication): A mechanism to repair a broken single strand of a chromosome by new DNA synthesis. In haploid budding yeast, Rad51-dependent BIR induced by HO endonuclease requires the lagging strand DNA Polα-primase complex as well as Polδ to initiate new DNA synthesis. Polε is not required for the initial primer extension step of BIR but is required to complete 30 kb of new DNA synthesis. Initiation of BIR also requires the nonessential DNA Polδ subunit Pol32 primarily through its interaction with another Polδ subunit, Pol31 (Leydeard JR et al 2007 Nature [Lond] 448:820). (See Kraus E et al 2001 Proc Natl Acad Sci USA 98:8255, BIR in yeast telomere elongation: McEachern MJ, Haber JE 2006 Annu Rev Biochem 75:115).

BIR (baculoviral IAP repeats): N-terminal motifs in the IAP proteins, in one or several copies. The Bir1p protein is an inhibitor of apoptosis and in cooperation with the kinetochore proteins Ndc10p, Cep3p, Ctf13p and Skp1p controls chromosome segregation. ►IAP, ►kinetochore

Birch (*Betula*): The silver birch, hardwood tree *B. pubescens* is 2n = 28 and the *B. verrucosa* is 2n = 56; x = 14 (see Fig. B42).

Figure B42. Birch

BIRN (Biomedical Informatics Research Network, http://www.nbirn.net/; http://birn.ncrr.nih.gov): Initially it was concerned with neurosciences but now it is extended to a wide range of biological projects. It provides an infrastructure for biology, computer science and statistics with the goal that in the mound of data, a pattern/patterns could be found that would suggest mechanisms of function and facilitate further experimental tests. ►Internet2, ►Abilene

Birnavirus: Icosahedral double-stranded RNA virus.

Birth Control: ►contraceptives, ►hormone receptors, ►sex hormones, ►menstruation

Birt-Hogg-Dubé Syndrome: A genodermatosis (genetic skin disease) involving tumorous hair follicles, renal neoplasia, lung cysts and pneumothorax (air accumulation in the serous membrane of the chest). It is linked to the pericentromeric region of human chromosome 17p. (See Schmidt LS et al 2001 Am J Hum Genet 69:867).

Birth Defect: A perinatal anomaly of either hereditary (~6% chromosomal, ~7–8% monogenic, ~20% polygenic) or of extraneous cause (maternal disease, infection or caused by environmental physical of chemical agents).

Birth Rates: ►age-specific birth and death rates

Birth Weight: It may be affected by a number of intrauterine factors such as maternal nutrition, disease, smoking and also genetic causes. Some of the initial relative differences may or may not be eliminated during subsequent development. ►glucokinase

Birth-and-Death Evolution: ►concerted evolution

Bisexuality: May be a case of hermaphroditism or just a behavioral anomaly. In the fruit fly some losses in the brain olfactory centers or receptors lead to a defect of the interpretation of pheromones causing anatomically male flies to court females as well as males. ►pheromones, ►hermaphrodite, ►homosexual, ►olfactory, ►olfactogenetics, ►sex determination

Bison: American buffalo (*Bison bison*), 2n = 60.

Bispecific Monoclonal Antibodies (diabody): One of the two arms of the antibody has the recognition site for the surface antigens of a tumor cell, the other for

the antigens of a killer lymphocyte. The bispecific antibody is thus expected to bring these two cells together and destroy the tumor cells. ►antibodies, ►monoclonal anti bodies, ►antibody engineering, ►diabody, ►triabody, ►quadroma

Bisphenol: An estrogen and an industrial chemical used in manufacturing polycarbonate food and beverage containers. It can leach out and may be hazardous to developing offspring (see Fig. B43). ►estradiol, ►sex hormones

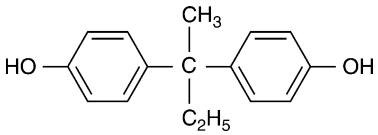

Figure B43. Bisphenol

Bistable Systems: They can toggle between two alternative steady-states but cannot rest in intermediate states. Such systems have importance for signal transduction, feedback and differentiation. ►signal transduction, ►feedback; Ferrell JE Jr 2002 Current Opin Cell Biol 14:140.

Biston betularia (peppered moth): A frequently used example for adaptive natural selection (see Fig. B44). The moth had predominantly overall grayish tones until the industrial revolution in the vicinity of Birmingham, England deposited black soot on the tree barks and favored the propagation of the dark colored (carbonaria) form of the moth that could hide better from predators. In unpolluted areas the light peppered form remained. These experiments have been subjected to criticism, mainly on ground of flawed methodology because during the decades after the publication of the Kettlewell experiments of the 1950s (Nature 175:943) the dark forms declined. The moth population is actually in a dynamic equilibrium and frequency-dependent selection affects the frequency of different types (Cook LM 2003 Quarterly Rev Biol 78:399). (Cook LM, Grant BS 2000 Heredity 85:580).

Figure B44. *Biston betularia*

Bisulfite (sodium bisulfite, $HNaO_3$): deaminates cytosine to uracil (Shortle D et al 1981 Annu Rev Genet 15:265).

Bit: A binary digit with a two-way choice such as a value of 1 or 0, on or off, etc. The smallest unit of information a computer recognizes. One byte = 8 bit and 1 kilobyte (K) is 1024 bytes (2^{10}); 1 megabyte (MB) = 2^{20} bytes.

Bithorax: (*bx*, 3–58.8) ►morphogenesis in *Drosophila* (see Fig. B45), ►Polycomb, Duncan I 1987 Annu Rev Genet 21:285.

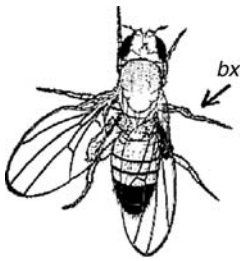

Figure B45. *Bithorax*. (From Bridges & Morgan 1923, *bx*)

Bitmap: Bits representing a graphic image in the memory of a computer.

Bitnote: A message communicated through the computer (e-mail) using the Bitnet system, an IBM mainframe connection to the Internet. ►Internet

BITOLA (Biomedical Discovery Support System): Directs potential relationships between biomedical concepts. The concepts are under Medical Subject Heading (MSH) that is indexed in Medline and HUGO. The system facilitates the discovery of disease–candidate gene relations. Background knowledge and chromosomal; locations are included. ►Medline, ►LocusLink, ►HUGO; http://www.mf.uni-lj.si/bitola/.

Bitransgenic Regulation: The *A* transgene to be expressed must have its regulator transgene *R* be present in the cell. For normal function, the appropriate ligand(s) must also be available within the cell. ►transgenic, ►ligand; Yao TP et al 1992 Cell 71:63.

Bitscore: The raw quantitated sequence alignment score that indicates the statistical property of the scoring system. It is the likelihood that a query sequence is a genuine homolog of the sequence in the database. The natural logarithm of this likelihood ratio is the bits score. ►Blast, ►likelihood

Bivalent: Two homologous chromosomes, consisting of altogether of 4 chromatids, paired in meiotic prophase. ►heteromorphic bivalent, ►interlocking bivalent, ►chromatid, ►synaptonemal complex

Bivalent Promoters: In embryonic stem cells, bivalent promoters carry both trimethylated lysines at 4 and 27 position of histone 3 (H3K4me3 and H3K27me3).

The former is conducive to transcription, the latter to silencing. This combination probably serves to poise key developmental genes for lineage-specific activation or repression, respectively.

Bivariate Distribution: The joint distribution of two random variables.

Bivariate Flow Cytometry: Sorting chromosomes tagged with two fluorochromes (Hoechst 33258), specific for A=T and chloromycin A3, specific for G≡C, and excited by laser. flow cytometry, laser; Nunez R 2001 Curr Issues Mol Biol 3:67.

Bivariate Plot: Two-dimensional arrangement of data of two variables.

Bivoltinism: Insects have two generations per year.

Bixin: A pigment extracted from *Bixin orellana*, a tropical American plant and it is used in food and cosmetic industry. It is produced by several steps from lycopene and can now be synthesized in *E. coli* transformed for three genes encoding the critical enzymes. ►lycopene; Bouvier F et al 2003 Science 300:2089.

BK Virus: It has 80% homology to Simian virus 40 with a somewhat different host range (human, monkey, hamster and other rodent cells). It may occur as an episomal element in two dozen to hundreds of copies. Its autonomous replicon may be useful for propagating DNA and genes in human cell cultures.

BKM Sequences: tetranucleotide GATA and GACA repeats in the W chromosomes (comparable to the Y chromosome) of birds and reptiles, occasionally in other eukaryotic chromosomes. ►satellite DNA, ►tetranucleotide repeats, ►Y chromosome

Black Box: A slang expression for equipment that is too complicated inside to be generally understood. Figuratively, a living cell was considered to be a black box because some of its functions were observed yet all the mechanisms that drove these functions were not fully understood. Geneticists knew segregation of genes and chromosomes but the molecular mechanisms underlying these processes were largely shut inside the "black box" until the discoveries of DNA replication, transcription, translation, gene regulation, cell cycle, etc.

Black Locust (*Robinia pseudoacacia*): A leguminous tree with fragrant flowers; 2n = 20.

Black Pepper (*Piper nigrum*): Southeast Asian spice. Basic chromosome number probably 12, 13, or 16, and 2n = 46, 52, 104 and 128 have been reported.

Bladder Exstrophy: An apparently recessive familial disease with poor penetrance. A defect of the hindgut (cloaca) results in open lower abdominal wall, pubis, lower urinary tract and the genitalia. The expressivity varies.

β-Lactamase: An enzyme (synonym: penicillinase) capable of cleaving the β-lactam ring of antibiotics of the penicillin family (see Fig. B46). Their activity is determined by the R-group attached to the lactam ring. The majority of the synthetic penicillins are not susceptible to penicillinase action. The coding gene was originally detected in Tn*3*. The ampicillin resistance gene in the pBR322 plasmid codes for 263 amino acid residue pre-protein containing a 23 amino acid leader sequence, which directs the secretion of the protein into the periplasmic space of the bacterium. The transcription of the gene starts counterclockwise at pBR322 coordinate 4146 and ends at 3297. Its mRNA in vitro contains a 5′-pppGpA terminus. The tetracycline resistance gene in pBR322 is transcribed from another promoter clockwise, starting at coordinate 244 or 245. The Tc^R gene encodes a polypeptide of 396 residues. Penicillinases occur naturally only in bacteria with peptidoglycan cell wall. The lack of the enzyme, in the absence of antibiotics, is inconsequential for the bacteria. The β-lactamase genes are used extensively in vector construction (to convey antibiotic resistance) and for the detection of insertional events that inactivate the enzymes. The lactamase enzyme can be used for real time monitoring gene transcription. A substrate (e.g., cephalosporin) complexed with a fluorochrome (7-hydroxy-coumarin) upon hydrolysis may generate a wavelength shift (from blue to green) in the emission of the substrate located in the plasma membrane. With aid of a cell sorter, transcription can be monitored in single cells. β-Lactamase is inhibited by clavulanic acid (an oxygen containing β-lactam). ►antibiotics, ►Tn*3*, ►periplasma, ►vectors, ►cell sorter, ►fluorochrome, ►coumarin, ►penicillin, ►PBP; Daiyasu H et al 2001 FEBS Lett 503:1; Jacoby GA et al 2005 New Engl J Med 352:380.

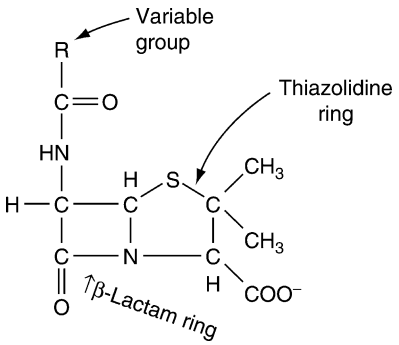

Figure B46. β-Lactamase

Blank Allele: It is not expressed.

BLAST (basic local alignment search tool): It is used for comparison of nucleotide sequences in apparently related (homologous) DNA (Altschul SF et al 1990 J Mol Biol 215:403) or for amino acid sequences in proteins (for DNA BLAST and for proteins PSI-BLAST: Altschul SF et al 1997 Nucleic Acids Res 25:3389). E-mail address blast@ncbi.nlm.nih.gov. ►DNA sequencing, ►MegaBLAST, ►homology, ►evolutionary tree, ►FASTA, ►BLOSUM, ►databases; Trends Supplement [Elsevier Science] 1998; Wolfsberg TG, Madden TL 1999 In: Ausubel FM et al (eds) Short protocols in molecular biology, Wiley, New York, p.18-1, http://www.ncbi.nlm.nih.gov/BLAST/, WU-BLAST: http://blast.wustl.edu ftp://ftp.ncbi.nih.gov/blast/executables/LATEST/.

Blast Cell: A cell, which may give rise to a progeny cell(s) different from itself.

BLASTP: BLAST for proteins. ►BLAST

Blastema: A group of cells resembling stem cells in function and instrumental in tissue or organ regeneration. Hsp60 is required for regeneration of animal organs from blastema. Mutation in Hsp60 leads into mitochondrial defects and apoptosis (Makino S et al 2005 Proc Natl Acad Sci USA 102:14599). ►chaperonin, ►apoptosis, ►regeneration in animals

Blastid: The site in the fertilized egg where cellular organization takes place.

Blastocoele: ►Blastula

Blastocyst: An early embryonal stage (of about 60 cells in mammals) when the blastocoele is enveloped by a trophoblast cell layer, a pre-implantation stage of the animal zygote when the zona pellucida (the envelop of the egg) is still visible and the blastula begins to develop its inner cell mass (see Fig. B47). Before implantation the blastocysts sheds the zona pellucida. The mode of implantation is somewhat different in different mammals. ►blastocoele, ►trophoblast, ►blastula, ►stem cells, ►uterus; Wang H, Dey SK 2006 Nature Rev Genet 7:185.

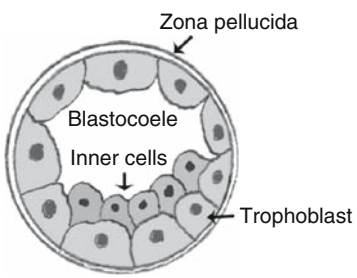

Figure B47. Blastocyst

Blastocyte: An undifferentiated cell of an early zygote.

Blastoderm: A single layer of cells at the embryonic stage of insects surrounding a fluid-containing cavity (blastocoele) at the blastula stage of cell divisions.

Blastoma: A cell in the early stage of differentiation or a neoplastic tissue containing embryonic cells.

Blastomeres: The large fertilized egg through cleavage divisions produces smaller cells, the blastomeres. These divisions are extremely fast and during the short process RNA synthesis ceases and protein synthesis depends on reserve mRNAs. Individual blastomeres of the mouse are already different at the two-cell stage as detectable by the localization of the Cdx2 transcription factor. This cell gives rise to the trophectoderm during the following divisions (Deb K et al 2006 Science 311:992). ►blastoderm, ►cleavage, ►founder cells, ►Cdx2, ►trophectoderm, ►oviduct, ►morula

Blastopore: Located near the site of the gray center of the animal pole where invagination of the blastula begins and eventually encompasses the vegetal pole. The *dorsal lip* of the blastopore organizes gastrulation. ►blastula, ►animal pole, ►vegetal pole, ►gastrulation, ►organizer

Blastula: A product of the cleavage of the early zygote when it becomes a spherical structure in which the blastoderm envelops the blastocoele cavity. ►blastocoele

BLASTX: A computer program for gene and protein searches. (Nature Genet. 3:266 [1993]).

BLAT: A computer program for aligning nucleotide or amino acid sequences. (Ogasawara J, Morishita S 2003 J Bioinform Comput Biol 1:363).

Blattner Number: Refers to the position of genes in the sequenced genome of *E. coli* bacterium. ►*E. coli*, Blattner FR et al 1997 Science 277:1453; Riley M, Serres MH 2000 Annu Rev Microbiol 54:341; Liang P et al 2002 Physiol Genomics 9:15; http://genprotec.mbl.edu/.

Blau Disease: A form of ulcerative colitis (intestinal inflammation) under the control of more than a single gene. Its symptoms overlap with those of Crohn disease. ►Crohn disease; Miceli-Richard; C et al 2001 Nature Genet 29:19.

Blebistatin: (-)-blebistatin is an inhibitor of myosin II of mammals and thus the contraction of the cytokinesis cleavage furrow without affecting the assembly of the contractile ring. ►cytokinesis, ►myosin; Straight AF et al 2003 Science 299:1743.

Bleeder Disease: ►hemophilia, ►antihemophilic factors

B

Blending Inheritance: Up to the time of Mendel it was erroneously considered to account for some of the variations observed in nature. A contemporary of Darwin, Fleeming Jenkins, an engineer, has pointed out that if this would be true, unique traits of single organisms should disappear in panmictic populations like a drop of ink in the sea. ►particulate inheritance

Bleomycins (BLM): Antitumor (malignant lymphomas, squamous cell carcinoma) antibiotics (see Fig. B48). Degrade DNA (and also RNA), especially when supported by redox-active metal ions (Fe, Cu) with specificity to 5′-GT-3′ and 5′-GC-3′ sequences in double or single strands. Fe(II)•BLM does not cleave, however all RNAs and shows preferences to some tRNAs although it may cleave a variety of RNAs (mRNA, 5S rRNA, DNA–RNA heteroduplexes, etc.). Mg^{2+} is inhibitory. BLM also affects lipid peroxidation. ►cancer therapy, ►ribonuclease, ►non-ribosomal peptide; Hoehn ST et al 2001 Nucleic Acids Res 29:3413.

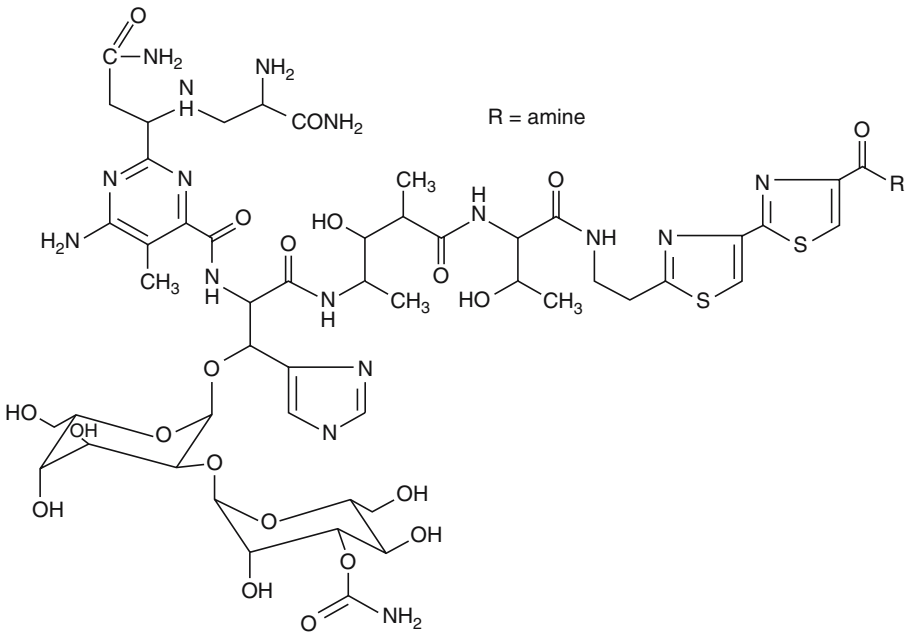

Figure B48. Bleomycin

Blepharophimosis: A defect of the eyelids and nose, associated also with ovarian atrophy and small uterus (see Fig. B49).

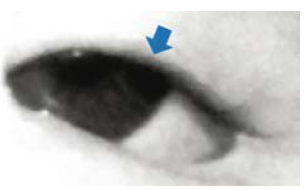

Figure B49. Note the drooping eyelids (ptosis) in blepharophimosis

The dominant disorder was assigned to human chromosome 3q21-q23 (FOXL2), another locus (TWIST) with ptosis is at the 7p21 (Dollfus H et al 2001 J Med Genet 58:470). The FOXC1 gene (6p15) defect may also be responsible. ►polled; De Baere E et al 2001 Hum Mol Genet 10:1591; De Baere E et al 2003 Am J Hum Genet 72:478.

Blepharoplast: basal body.

Blimp-1: A repressor of c-Myc oncoprotein expression that blocks plasmacytoma formation, and terminal differentiation of B lymphocytes, ►MYC, ►B lymphocyte

Blind Test: The treated individuals are unaware of the treatment or exposure.

Blindness: ►eye diseases

Blister: A vesicle-like skin abnormality. Autosomal dominant, recessive and X-linked types exist. The autosomal dominant class: bullous erythroderma involves a hyperkeratosis with anomalies of the keratin tonofibrils. In one form, the blisters are limited to hands and feet and appear only in warm weather after heavier exercises. Another form (epidermolysis bullosa dystrophica) may appear already at birth and the blisters may appear besides the extremities, on the ears and buttocks. The latter type accumulates and secretes sulfated glycosaminoglycans. In an early and transient form, the blisters disappear generally by the end of the first year and do not return. In the herpetiform epidermolysis

bullosa the larger vesicles appear in clusters on the palms, soles, neck and around the mouth apparently due to a mutation in the keratin 14 gene. Another epidermolysis bullosa appears to be due to deficiency of galactosylhydroxylysyl glucosyltransferase. In one form, a human chromosome 12-coded gelatin-specific metalloprotease deficiency may be involved. A mottled type (pigmented spots) displays recurrent blistering beginning at birth and premature aging. The epidermolysis bullosa with absence of skin and deformity of nails has a perfect penetrance. The autosomal recessive types: epidermolysis bullosa dystrophica (human chromosome 11q11-q13) is caused by excessive collagenase activity affecting primarily the hands, feet, elbows and knees at birth or infancy but may affect other organs too. The epidermolysis bullosa letalis may kill infants within about three weeks after birth but occasionally some survive to the first decade of life. In some forms the distal opening of the stomach (pylorus) may be constricted and atrophied and in other forms congenital deafness, muscular dystrophy may appear. X-linked epidermolysis with multiple complications (baldness, hyperpigmentation, dwarfism, microcephaly [small head], mental retardation, finger and nail malformation, and death before adult age is also known.) ►keratosis, ►ichthyosis, ►skin diseases

BLK: ►SRC oncogene family

BLM: ►Bloom syndrome

Bloch-Sulzberger Syndrome: ►incontinentia pigmenti

Block: A group of related sequences in proteins or DNA stretches between recombination sites. (Kimmel G, Shamir R 2005 Proc Natl Acad Sci USA 102:158; protein block database: http://motif.stanford.edu/eblocks/.

Block Design: Generally each treatment is present in every block (complete block) where the treatments may be randomized. In the incomplete block design, not all treatments are present in each block because of the large number of treatments causes technical difficulties. In the latter cases mathematics is used for compensation.

Block Mutation: It affects more than a single nucleotide in the cell, e.g., deletion; such mutations may not yield wild type recombinants if the defects overlap.

Blocked Reading Frame: Translation is interrupted by nonsense codons. ►nonsense codons

Blocking Buffer: 3% BSA (bovine serum albumin) in phosphate buffered saline containing also 0.02% sodium azide. BSA blocks the binding sites on nitrocellulose filter that are not occupied by proteins transferred from (SDS polyacrylamide) gels. ►gel electrophoresis

Blocks: An Internet tool for the search of functional DNA motifs: blocks@howard.fhcrc.org or http://blocks.fhcrc.org.

Blog: Informal communication through the Internet about various topics from science to politics. The site http://scienceblogs.com/ contains an index of blogs of possible interest to science. More information about blogging: Bonetta L 2007 Cell 129:445.

Blood: The fluid that carries nutrients and oxygen by circulating through the blood vessels in the animal body. It is composed of red (non-nucleated mature erythrocytes) and white (nucleated leukocytes) cells. The white cells include granulocytes, neutrophils, eosinophils and basophils, B and T lymphocytes and natural killer cells. The differentiation of these various types of cells from the multipotential hematopoietic stem cells is determined by a combination of growth factors, such as interleukins, stem cell factors, colony stimulating factors, etc. In the lymphoid developmental path the Pax5 gene and its product BSAP (B-cell-specific activator protein) plays a key role. The blood contains also platelets (thrombocytes) and the blood plasma, the non-corpusculate yellowish fraction. In *Drosophila* there are crystal cells (contain defense enzymes), plasmatocytes (the main phagocytotic defense cells) and lamellocytes (develop from plasmatocytes with similar functions). See these cell types, ►hemolytic disease, ►ABO blood group, ►blood groups, ►macrophages, ►dendritic cells, ►immune system, ►hematopoiesis, ►Pax, ►antihemophilic factors, ►B lymphocyte, ►T cell, ►erythrocyte, ►leukocyte, catalog of blood proteins relevant to disease: http://bpp.nci.nih.gov/; www.plasmaproteomedatabase.org, biology and disease: www.blood.interhealth.info/, blood cell development: http://hembase.niddk.nih.gov.

Blood Brain Barrier: ►BBB

Blood Clotting Pathways: These are the intrinsic pathway involving the successive participation of the Hagemann factor (XII), plasma thromboplastin antecedent (PTA XI), Christmas factor (IX), antihemophilic factor (VIII), Stuart factor (X), phospholipid and proaccelerin and the extrinsic clotting pathway requiring proconvertin (VII), Stuart factor (X), proaccelerin and calcium ions. With the aid of lentiviral vectors directed to the liver of mouse, the human Factor IX gene can be expressed and stably maintained for months. ►antihemophilic factors, ►tissue factor, ►vitamin K; Tsui LV et al 2002 Nature Biotechnol 20:53.

B

Blood Coagulation: ►blood clotting pathway, ►anti-hemophilic factors, ►aspirin

Blood Formation (hematopoiesis): During early embryonic development the yolk and the aorta-gonad-mesonephros (AGM) region are involved, later the function in the embryo is switched to the liver and after birth, the bone marrow is involved.

Blood Groups: An incomplete list of the types found in this volume: ABO, ABH, Ahonen, Colton, Diego, Dembrock, Duch, Duffy, En, Gerbich, I system, Kell-Cellano, Kidd, Lewis, Lutheran, LW, MN, Newfoundland, OK, P blood group, Radin, Rhesus, Scianna, Ss, Webb, Wright, Yt, and Xg. These are distinguished mainly by the epitopes on the erythrocytes. ►epitope, ►erythrocyte

Blood Pressure: The pressure of the blood on the blood vessels (arteries). ►hypertension

Blood Transposable Element: ►copia

Blood Typing: Identification the blood group a person belongs to. ►blood groups

Bloom Syndrome (BS, BLM): Semi-recessive human dwarfism; increases the frequency of chromosomal aberrations (particularly sister chromatid exchanges), and various forms of cancer (leukemia), sensitive to sunlight (red blotches over face) and usually shorter than normal life expectancy. It was attributed to a DNA ligase I deficiency but the cloning and sequencing of the gene indicates that this is not the primary defect rather a DNA helicase-like protein (RecQ family, homolog of budding yeast genes SGS1 and SRS2), encoded in human chromosome 15q26.1, is involved. The DRAFT protein complex is shared by Fanconi anemia and Bloom syndrome (Meetei AR et al 2003 Mol Cell Biol 23:3417). SGS1 mutations greatly enhance gross chromosomal aberrations and the rate of recombination. The wild type SGS1 represses chromosomal aberrations. The *Drosophila* homolog is Dmblm. In BS sister chromatid exchange and mitotic recombination are elevated but genetic repair seems to be normal. In Dmblm, an extra copy of Ku70 compensates for sterility. The BS helicase physically interacts with the Werner syndrome helicase. ►DNA repair, ►xeroderma pigmentosum, ►Fanconi anemia, ►Werner syndrome, ►Cockayne syndrome, ►Rothmund-Thomson syndrome, ►carcinogenesis, ►light-sensitivity diseases, ►co-suppression; Ku, Luo G et al 2000 Nature Genet 26:424; Myung K et al 2001 Nature Genet 27:113; von Kobbe C et al 2002 J Biol Chem 277:22035.

BLOSUMS: Amino acid substitution matrices used to determine evolutionary changes in proteins [Henikoff S, Henikoff JG 1992 Proc Natl Acad Sci USA 89:10915]. ►BLAST, ►FASTA, ►evolutionary clock, ►SSPA

Blotting: Macromolecules separated by electrophoresis in agarose or polyacrylamide are transferred to a cellulose or nylon membrane and immobilized there for further study. ►Southern blot, ►Northern blot, ►Western blot, ►colony hybridization, ►immunoprobe

BLOTTO (Bovine Lacto Transfer Technique Optimizer): A 5% solution of non-fat, evaporated milk in 0.02% sodium azide (NaN3). [It may contain RNase activity]. In 25-fold dilution, it may be used for blocking background annealing in Grunstein-Hogness hybridization, Benton-Davis hybridization, dot blots, and non-single copy Southern hybridization. ►Denhardt reagent, ►heparin, ►Grunstein-Hogness screening, ►Benton-Davis plaque hybridization, ►dot blot

Blue Grass (*Poa pratensis*): Lawn and pasture plant; 2n = 36–123 in the polyploid series.

Blue Diaper Syndrome: An intestinal failure to transport tryptophan and *Pseudomonas aeruginosa* bacteria convert the amino acid into indole, which upon oxidation stains bluish. ►amino acidurias

Blue Light Response: Photomorphogenetic reaction (of plants) to illumination in the range of 400–500 nm wavelength. ►photomorphogenesis, ►cryptochromes

Blueberry (*Vaccinium* spp): A fruit shrub with x = 12; *V. corymbosum* (high-bush blueberry) is tetraploid, the *V. angustifolium* (low-bush blueberry) is diploid.

Bluescript M13: A 2.96 kb genetic vector containing the bacteriophage M13 replication origin and a polycloning insertion site flanked by T7 and T3 phage promoters in opposite orientation and useful for generation of single-stranded DNA or RNA complementary to the double-stranded DNA insert. Several variations exist (e.g., (ZAP, bluescript SK). The name comes from the bacterial Lac fragment that upon expression of (β-galactosidase in Xgal medium forms an easily detectable blue color. ►Xgal; Short JM et al 1988 Nucleic Acids Res 16:7583; Snead M et al 1988 Methods Mol Biol 81:255.

Blunt End: The blunt-end of double-stranded DNA is generated by non-staggered cut and it terminates at the same base pair across both strands of the double helix (see Fig. B50). Bacterial DNA polymerase I (Klenow fragment) or phage T4 DNA polymerase can also generate 3′ blunt ends of DNA by 5′→ 3′ exonucleolytic activity. ►blunt end ligation

Figure B50. Blunt end

Blunt-End Ligation: T4 phage DNA ligase joins non-staggered DNA ends or adds chemically synthesized duplexes to double-stranded blunt ends. ►DNA ligases, ►linker

BLUP (best linear unbiased prediction): A statistical procedure based on covariance analysis of gametic genetic disequilibrium of QTL and other types of markers in multibreed populations. (Wang T et al 1998 Genetics 148:507, ►multibreed, ►QTL)

BLYM: Chicken bursal lymphoma oncogene, located to human chromosome 1p32. It is homologous to transferrin, a glycoprotein with important role in the synthesis of ribonucleotide reductase, and thereby in DNA replication and mitosis. ►oncogenes, ►lymphoma, ►transferrin

BLYS (B lymphocyte stimulator): A human chromosome 13q34-encoded protein of the tumor necrosis factor family is involved in B cell proliferation and immunoglobulin secretion. ►B lymphocyte, ►TNF

BMI: ►body mass index

BMK1 (big MAP kinase): ►ERK

BMP: ►bone morphogenetic protein

BMT: Transformed monkey cell line expressing the T antigen of SV40, driven by a mouse metallothionein promoter. ►SV40, ►metallothionein

BMYC: Oncogene isolated from rat has extensive homology to the MYC oncogene but it maps to a different location than the other members of the MYC family LMYC, NMYC, PMYC, RMYC. (►MYC and other members of the family, ►oncogenes)

BNA: ►locked nucleic acids

Bni: A member of the formin proteins involved in polar morphogenesis and cytokinesis of eukaryotic cells. Bni1 protein is associated with CDC42 protein, with actin, profilin and Bud6. ►actin, ►profilin, ►CDC42, ►Bud

Bob: ►OBF

BOB': ►*att* sites

BODIPY: ►fluorochromes

Body Map (human and mouse gene expression database): Abundance of mRNAs in different tissues of the body. It reveals also the functional relatedness of genes by the similarity of expression profiles. (See Kawamoto S et al 2000 Genome Res 10:1807; http://bodymap.ims.u-tokyo.ac.jp; http://bodymap.jp/).

Body Mass Index in Humans (BMI): BMI is determined as a measure of obesity by the formula: weight in kg/height in meter^2. In morbid obesity BMI > 40 kg/m2. Obesity (BMI>30) is detrimental to health although for centuries it was associated with fertility (see Fig. B51). The body mass index does not take into account unusually strong muscle development and in such cases does not provide good measure of obesity. Correlation of BMI between monozygotic twins is about 0.74 versus 0.32 for dizygotic twins indicating very high heritability. The correlation among parents and biological offspring were found to be 0.19 versus adopted children 0.06 indicating the major role of hereditary factors. The pro-opiomelanocortin locus in human chromosome 2p21 is a significant contributor to body mass according to some studies. Perola M et al 2001 Am J Hum Genet 69:117) failed to detect any linkage to BMI by QTL in Finnish populations. ►obesity, ►leptin, ►melanocortin, ►morbidity, Barsh GS et al 2000 Nature [Lond] 404:644.

Villendorl Venus
ca. 15000 B.C.

Figure B51. Fertility goddess from Willendorf, Austria ~15,000 B.C. (From Gowans CS 1974 Stadler Symp 6:113)

Body Plan (bauplan): The three-dimensional organization of the body of an organism, e.g., mammals are bilaterally, incomplete symmetrical quadripeds, star fish has five-fold symmetry.

Body Size: In insects a balance between ecdysone and insulin signals determines body size. Ecdysone is produced by the prothoracic gland and its size is controlled by activated RAS or PI3K although RAF also regulates body size by activation ecdysone-dependent genes (See Caldwell PE et al 2005 Current Biol 15:1785). ►ecdysone, ►RAS, ►PI3K, ►size, ►body mass, ►Kleiber's rule

bol: ►boule

Boltzmann Time Warping: ►dynamic time warping

B

bom: A bacterial gene (basis of mobilization), required for the transfer of plasmids. ►plasmid mobilization, ►mob, ►Hfr

Bombardia lunata: An ascomycete, n = 7.

Bombay Blood Type: A relatively rare blood type discovered in India and subsequently on the Reunion Island in the Indian Ocean. This blood type has two main forms determined by the recessive alleles h (for H type red cell antigen). In the h/h se/se individuals also the enzyme fucosyltransferase 1 is inactive; in the h/h Se/se individuals a weak expression of the H antigen may be observed; Se is apparently coding for fucosyltransferase 2. ►ABH antigen, ►Secretor, ►Lewis blood group, ►fucose

Bombesin (protein, $C_{71}H_{110}N_{24}O_{18}S$): Bombesin modulate smooth muscle contraction, hormone traffic, metabolism, hyperglycemia, hypertension and eating behavior. ►obesity, ►ovarian cancer

Bombyx mori: ►silkworm

BONCAT (bioorthogonal noncanonical amino acid tagging): A method for detection and separation of newly synthesized proteins in response to environmental cues. The metabolic machinery incorporates azides and ketones and subsequent ligation with reactive probes facilitates their detection. Azidohomoalanine can be introduced into mammalian cells and the tagged proteome can be separated by affinity chromatography and identified by tandem mass spectrometry (Dieterich DC et al 2006 Proc Natl Acad Sci USA 103:9482). ►azide, ►ketone, ►homoalanine, ►affinity chromatography, ►tandem mass spectrometry

Bond Energy: It is required to break a chemical bond. Such bonds in the piconewton range are important in biology and are represented by protein and nucleic acids interactions, receptor-ligand pairs and covalent bonds. Measurement of the forces involved may be important for, e.g., detection of DNA base mismatches and for proteomics. ►atomic force microscope, ►newton; Albrecht C et al 2003 Science 301:367.

Bone Development: A complex process that involves signaling, molecules, growth hormones, transcription factors, etc. ►osteoblast, ►osteoclast; Kronenberg HM 2003 Nature [Lond] 423:332; Boyle WJ et al 2003 Nature [Lond] 423:337; Harada S-i, Rodan GA 2003 Nature [Lond] 423:349.

Bone Diseases: ►collagen, ►campomelic dysplasia, ►achondroplasia, ►hypochondroplasia, ►pseudoachondroplasia, ►osteogenesis imperfecta, ►SED, ►PAPS, ►osteoporosis, ►osteosarcoma, ►osteolysis, ►Paget disease, ►osteopetrosis, ►diastrophic dysplasia, ►chondrodysplasia, ►dyssegmental dysplasia, ►dyschondrosteosis, ►dwarfism, ►adactyly, ►brachydactyly, ►polydactyly, ►syndactyly, ►exostosis, ►trichorhinophalangeal syndrome, ►ectrodactyly, ►Ellis-van Creveld syndrome, ►Holt-Oram syndrome, ►Hay-Wells syndrome, ►head/face/brain defects, ►pycnodysostosis, ►osteochondromatosis, ►Larsen exostosis Alagille syndrome, ►spondylocostal dysostosis, ►spondylometaphyseal dysplasia, ►spondyloepiphyseal dysplasia, ►Greig's cephalopolysyndactyly syndrome, ►craniometaphyseal dysplasia, ►Pallister–Hall syndrome, ►Townes–Brocks syndrome, ►Robinow syndrome, ►acromesomelic dysplasia, ►acrodysostosis, ►acheiropodia, ►nail-patella syndrome, ►symphalangism proximal, ►radioulnar synostosis, ►synostosis, ►cleidocranial dysplasia, ►Waardenburg syndrome, ►Stickler syndrome, ►hypophosphatemia, ►Kniest dysplasia, ►Marfan syndrome, ►osteolysis, ►osteopetrosis, ►osteoporosis, ►sclerosteosis, ►scoliosis, ►Camurati-Engelmann disease, Kornak U, Mundlos S 2003 Am J Hum Genet 73:447.

Bone Marrow: The red spongy tissue inside the bones gives rise to lymphocyte stem cells and erythrocytes; the yellow bone marrow is mainly made of fat cells (see Fig. B52). Hematopoietic stem cells egress from the bone marrow by signals from the sympathetic nervous system (Katayama Y et al 2006 Cell 124:407). From the stromal cells of the bone marrow, stem cells can be isolated that can generate muscle cells at very good efficiency (89%) and suitable also for the development of neural and other types of cells under appropriate conditions and can be exploited for repairing degenerated tissue. It has the advantage that these stem cells would not be rejected if taken from the same individual and would avoid some ethical objections to the use of embryonic stem cells (Dezawa M et al 2005 Science 309:314). It has been claimed that stem cells exiting from the bone can end up in the ovaries and produce oocytes. Newer experiments challenged this claim (Powell K 2006 Nature [Lond] 441:795). ►stem cells, ►thymus, ►sympathetic nervous system, ►hematopoiesis

Figure B52. Bone marrow is in the inner cavity (light brown)

Bone Morphogenetic Protein (BMP): A maternally expressed factor in *Xenopus* embryos; in addition to bone differentiation it is involved in dorso-ventral organization of the embryo. Osteoblast differentiation

and proliferation is controlled by BMP and Smad. BMP-1 is a procollagen protease (PCP) that assembles collagen within the extracellular matrix. The other BMPs belong to the transforming growth factor (TGF-β) family. BMP-4 regulates apoptosis in neural crest cells affecting skeletal bone and muscle formation. BMP-3 is a negative regulator of bone density. The growth/differentiation factors of mouse (GDF) belong to this family and their mutation shortens the limb bones (brachypodism) without affecting the axial skeleton. The CBFA-1 gene seems to be a major factor in ossification. BMP is the vertebrate homolog of decapentaplegic (dpp) in *Drosophila*. The Sog (short gastrulation) and the Chd (chordin) proteins in vertebrates and invertebrates negatively regulate the BMP/Dpp system, respectively. The metalloprotease Xld (Xolloid)/Tld (tolloid) release the Bpm/Dpp from inactive complexes. Thus, a balance between Sog/Chd and Xld/Tld determines a morphogenetic gradient. Noggin, gremlin, chordin and follistatin inhibit BMP and dorsalize the embryo. The process is, however, more complex since that other serine protease(s) may also be involved (astacin, furin). The Kuz metalloprotease (a reprolysin) regulates the Notch cell surface receptor by proteolytic cleavage. BMP in coordination with other signal proteins regulates the specification of teeth development: Msx-1^{+}(Barx-1^{-} activity state leads to the development of incisors whereas Msx-1^{-}(Barx-1^{+} state promotes molar formation in the oral mesenchyme in mouse. ►fibrodysplasia ossificans progressiva, ►pulmonary hypertension, ►decapentaplegic, ►furin, ►Notch, ►organizer, ►noggin, ►GLI, ►Smad, ►osteoclast, ►osteoblast, ►collagen, ►brachydactyly, ►tooth, ►hepcidin; Olsen BR et al 2000 Annu Rev Cell Dev Biol 16:191; Ray RP, Wharton KA 2001 Cell 104:801; Khokha MK et al 2003 Nature Genet 34:303.

Bonferroni Correction: Guards against type I error at some α values (false positives). In case of a small number of tests it is satisfactory and simple. It is frequently used when multiple hypotheses are tested: $p_{corrected} = 1 - (p_{uncorrected})^{n}$ where *p* is the values of the hypotheses and *n* is the number of hypotheses. The basic assumption is that all alternatives are equally likely and the results are tested against this hypothesis. ►significance level, ►error types, ►lest significant difference; Altman DG 1991 Practical statistics for medical research, Chapman & Hall, London; Hochberg Y 1988 Biometrika 75:800.

Bonobo (*Pan paniscus*): Pygmy chimpanzee is an ape closest to humans after chimpanzee. ►chimpanzee

Book Syndrome: An autosomal dominant defect of tooth development, high degree of sweating and premature loss of hair color. ►hair color

Bookmark: Indicates an address on the Internet or other items in the computer where you wish to return.

Bookmarking: The mechanism responsible for preventing compaction of a specific gene region during mitosis (Xing H et al 2005 Science 307:421).

Boolean Algebra: Developed by George Boole (1815–1864) for the use of formal logic. He supposed that in binary forms thinkable objects could be defined. Thus, if x = horned and y = sheep then by selecting x and y the class of horned sheep is defined. Also 1−x would define all things of the universe that are not horned, and (1−x)(1−y) would identify all things that are neither horned nor sheep. This approach defines sets and subsets in discrete forms without intermediates, yet capable of defining mutual relationships. Using simple symbols, syllogisms could be developed in mathematical forms. Learning of concepts by humans appears to be proportional to its Boolean complexity, i.e., to the length of the shortest logically equivalent proposition. The switch-gear of the telephone systems and the modern digital computers were developed on the basis of the Boolean binary logic. Boolean logic is employed in devising cellular automaton networks and some information retrieval system, e.g., PubMed is operated on the basis of Boolean principles. DNA-based digital logic circuits have also been designed for the detection of complex enzyme-free nucleic acid circuits for the monitoring of complex gene expression patterns (Seelig G et al 2006 Science 314:1585). ►automaton, ►networks, ►synthetic genetics, ►PubMed

Bootstrap: A statistical device that was introduced for computer operations (versus the classical type computations). The standard error by the classical method is computed as:

$$se(\bar{x}) = \left\{ \sum_{i=1}^{n} (x_i - \bar{x})^2 / [n(n-1)] \right\}^{1/2}$$

in comparison, with the bootstrap procedure:

$$se[t(x)] = \left\{ \sum_{b=1}^{B} [t(x^{*b}) - \bar{t}]^2 / (B-1) \right\}^{1/2}$$

where se[t(x)] is the standard error of the bootstrap statistic, t(x), B = bootstrap samples of size *n* from the data, $\bar{t}$ is the average of the B bootstrap replications ($\bar{t}(x^{*b})$). The bootstrap algorithm can be applied to the majority of statistical problems and it is widely used for estimating the confidence level in evolutionary trees. The data points x_i need not be single numbers, they can be vectors, matrices or more general quantities, such as maps, graphs. The statistic t(x) can be anything as long t(x*) can be computed for every bootstrap data set x*. Data set *x* does not have to be a random sample from a single distribution.

Regression models, time series, or stratified samples can be accommodated by appropriate changes. For details and specific references see Efron B, Tibshirani RJ 1993 An Introduction to the Bootsrap. Chapman & Hall, New York. ►jackknifing; Kerr MK, Churchill GA 2001 Proc Natl Acad Sci USA 98:8961; Davison AC, Hinkley DV 1997 Bootstrap methods and their application, Cambridge University Press, Cambridge, UK.

Bora Bora: ~220 kb centromeric sequences in *Drosophila*. ►centromere

Border Sequences: ►T-DNA

Borjeson Syndrome (Borjeson-Forssman-Lehman syndrome): Face, nervous system, endocrine defects, hypogonadism, assigned to human chromosome Xq26-q27. ►RBM, ►head/face/brain defects

Borna Virus: An enveloped negative-strand, non-segmented RNA virus with inverted terminal repeats (see Fig. B53). Its genome is replicated and transcribed in the nucleus of warm-blooded animals, including humans. Viral replication is controlled by genome trimming. From the 5′ end one or four bases can be eliminated in the hairpin structure limiting genome amplification but not protein synthesis. From three transcription units it transcribes mRNA for six proteins by overlapping open reading frames, read-through transcription signals and alternative splicing of the polycistronic transcripts. Infection of the nervous system of newborn rats results in inflammation and causing mood disorders reminiscent of schizophrenia and autism of humans. In adult animals, uncoordinated movement and serious weight loss were observed. (Diagram is modified after Schneider U et al 2005 Proc Natl Acad Sci USA 102:3441).

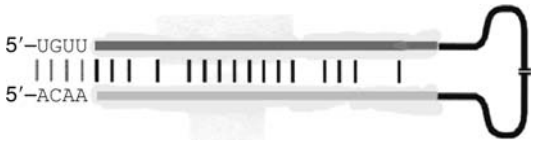

Figure B53. Borna virus

Borrelia: Spirochete bacteria; about 28 species (*B. burgdorferi*, *B. hermsii*, etc.) are responsible for relapsing fever or Lyme disease and other human ailments all over the world. The *Borrelia burgdorferi* genome B31 contains 910,725 bp linear DNA and 17 linear and circular plasmids. This bacterium, like *Mycoplasma genitalium*, has no genes for cellular biosynthetic functions but there are 853 genes for transcription, translation, transport and energy metabolism. The filamentous bacteria are 8 to 16 µm long flagellate cells infectious for birds and mammals. Their generation time is about 6 hours and in about 5 days within a single animal their population may exceed 10^6 cells and that coincides with the major symptoms (erythema migrans [enlarged red spots]) of the infection. Within weeks or months, the bacteria may invade all major organs of the body, primarily the joints and cause arthritic symptoms. If untreated, Lyme disease may be fatal. Intravenous injection of rocephin or other antibiotics (also orally administered doxycycline) may be the cures although some of the effects may persist for years. The vectors of the bacteria are the *Ixodes* arthropods (ticks) that live on grasses and low-growing bushes in wildlife (deer, mice, birds) frequented rural and suburban areas. The salivary protein Salp15 of *Ixodes scapularis* interacts with the surface protein of *B. burgdorferi* and protects the spirochete from the mammalian host antibodies (Ramamoorthi N et al 2005 Nature [Lond] 436:573). The bacterial σ^{54}, a RNA polymerase subunit deficient *Borrelia* cells can invade the tick but they are not infectious (Fisher MA et al 2005 Proc Natl Acad Sci USA 102:5162). The tick receptor (Trospa) that is required for spirochetal colonization (Pal H et al 2004 Cell 119:457). The *Bb* gene of *Borrelia* is required for the persistence of the bacteria in ticks and subsequent productive infection of the mammalian cell. The BptA a putative lipoprotein is a likely virulence factor for *Bb* (Revel AT et al 2005 Proc Natl Acad Sci USA 102:6972).

Identification of the disease is difficult because of the complexity of the symptoms. Serological detection encounters problems because the outer membrane of the bacteria displays variable serotypes. *Borrelias* harbor several copies of approximately 23–50-kb linear plasmids with genes for Vmps/Vsps (variable major proteins). Transposition within and recombination between the plasmids assures great antigenic variation in these organisms. New serotypes appear at an estimated frequency of 10^{-4} to 10^{-3} per cell per generation. This fact accounts for the difficulties in developing effective immunsera and no acceptable vaccine is available (Abbott A 2006 Nature [Lond] 439:525). An attenuated strain of *Mycobacterium bovis*, the bacillus Calmette-Guerin (BCG) may serve as a suitable vector for the *B. burgdorferi* surface protein antigen A and may secure more than a year long protection by mucosal delivery. About 10% of Lyme disease patients appear resistant to antibiotic treatment and display arthritis symptoms long after spirochetal DNA in fluids of the joints is no longer detectable. The arthritis is an immune response to the outer surface protein A (OspA) of the bacterium. Actually, OspA-reactive type 1 T helper lymphocytes are found in the joints many years after the infection is cured. OspA has

homology to human leukocyte-function associated antigen-1 (hlFA-1). Thus, it seems that the apparently antibiotic-resistant individuals have an autoimmune reaction to this major histocompatibility class peptide encoded by the dominant DRB*0401 allele. OspA is up-regulated when *Borrelia* is in its tick host and down-regulated when it is in a mammalian host. Before the bacterium enters the mammalian host, the host neuroendocrine stress hormones, epinephrine and norepinephrine, specifically bound by *B. burgdorferi,* and result in increased expression of OspA. This recognition is specific and blocked by competitive inhibitors of human adrenergic receptors. Propranolol significantly reduced uptake of *B. burgdorferi* by feeding ticks and decreased expression of OspA in the bacteria recovered from ticks that fed on propranolol (antihypertension drug)-treated mice (Scheckelhoff MR et al 2007 Proc Natl Acad Sci USA 104:7247). ►serotype, ►antigen, ►serum, ►mucosal immunity, ►*Ixodoidea*, ►σ, ►BCG, ►HLA, ►autoimmune disease, ►arthritis, ►leptospirosis; Ohnishi J et al 2001 Proc Natl Acad Sci USA 98:670, Kumaran D et al 2001 EMBO J 20:971; Revel AT et al 2002 Proc Natl Acad Sci USA 99:1562.

Borromean Rings: Interlocked rings. DNA may be arranged this or more complex ways (see Fig. B54). The simplest representation.

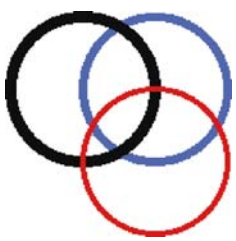

Figure B54. Borromean rings

Bos taurus (cattle): 2n = 60. ►cattle

Boss: A transmembrane protein product on the R8 photoreceptor in the eyes of *Drosophila,* encoded by gene boss (bride of sevenless, 3.90.5). It is the ligand and activator for the receptor tyrosine kinase, encoded by the sev (sevenless, 1–33.38) gene. ►sevenless, ►son-of-sevenless, ►rhodopsin, ►receptor tyrosine kinase, ►daughter of sevenless

Botany: A basic scientific field concerned with plants. (See herbaria and botanists: http://www.nybg.org/bsci/ih; all plants: http://plants.usda.gov/; Plant Name Index: http://www.ipni.org).

Bottleneck Effects: If the size of the population is periodically reduced substantially, genetic drift may alter gene frequencies. Usually bottlenecks reduce variation. Occasionally after bottlenecks, an increase of variation has been reported, and it was attributed to dominance and epistasis. Bottleneck effect is quite common in the transmission of mtDNA because only a small portion of it is passed through the germline and the heteroplasmy may be altered. ►genetic drift, ►mtDNA, ►heteroplasmy; Galtier N et al 2000 Genetics 155:981.

Bottom-Up Analysis: ►top-down analysis

Bottom-Up Map: It relies on STS-based information. These are useful for relatively short chromosomal distances. Two STSs are "singly linked" if they share at least one YAC and "doubly linked" in case they share at least two YACs. Single linkage is generally not useful because of the high degree of chimerism among the YACs. In the first step, STS are assembled into doubly linked contigs. Then, the doubly linked contigs are ordered either on the basis of radiation hybrids or traditional genetic recombination information. Finally, single linkage can also be used to join contigs to the same short genetic region. ►mapping genetic, ►STS, ►contig, ►radiation hybrid, ►bottom-down mapping, ►YAC; Carrano AV et al 1989 Genome 31:1059.

Botulin (botulinum): Highly toxic product of *Clostridium* bacteria (lethal dose 1 ng kg^{-1}) and frequent cause of potentially lethal food poisoning. It is approved for treatment of strabismus and blepharospasm (a nervous eyelid problem), and it is also a cosmetic treatment of "crow's feet" facial, signs of aging. It is also a potential biological weapon. The botulinum neurotoxin A enters neurons by binding to synaptic vesicle protein (SV2) isoforms (Dong M et al 2006 Science 312:592). Botulinum toxin also binds synaptotagmin II (Jin R et al 2006 Nature [Lond] 444:1092; Chai Q et al 2006 Nature [Lond] 444:1096). ►strabismus, ►synaptotagmin; Moore A 2002 EMBO Reports 3:714.

Boule (bol): An autosomal gene in *Drosophila* encoding a cell cycle protein regulating G2-M transition. It is homologous to the human Y-chromosomal gene DAZ responsible for azoospermia. Its suspected function is translation and localization of mRNA. ►infertility, ►fertility, ►azoospermia, ►cell cycle, ►twine, ►pelota, ►Dazla

Boundary Element (barriers): Limits the function of cis-regulatory elements or the spread of heterochromatinization. ►insulator, ►heterochromatin, ►RAP, ►CTCF

B

Bouquet (polarization): In leptotene the chromosomes are attached by their ends to a small area of the nuclear membrane (spindle pole body) while the rest of the chromosome length is looped across the nucleus (see Fig. B55). Meiotic proteins Bqt1 and Bqt2 tether the telomeres to the spindle pole body (fungal organ comparable to the centrosome). These proteins form a connecting bridge between telomere protein Rap1 and Sad1, a spindle pole protein (Chikashige Y et al 2006 Cell 125:59). ►meiosis, ►leptotene stage, ►spindle pole body, ►horsetail stage

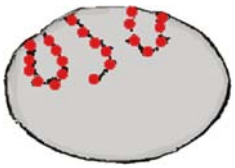

Figure B55. Bouquet

Bovine: Animals of *Bos taurus* (cattle) group. ►*Bos taurus*, ►cattle

Bovine Growth Hormone (BGH): A somatotropin; it has been commercially produced by genetic engineering and it boosts significantly milk production. ►somatotropin

Bovine Papilloma Viral Vectors: By genetic manipulations and with pBR322 bacterial plasmid sequences added, they can be converted into a shuttle vector carrying genes between mouse and *E. coli*. ►transformation genetic, ►vectors, ►viral vectors, ►shuttle vector

Bovine Papilloma Virus (BPV): A papova virus (about 7.9 kbp DNA), responsible for wart in animals. The BPV69T segment of their genome (5.5 kbp) has been used as a large capacity vector, which can multiply into 10 to 200 copies. It can stay as an episome or can be integrated into the chromosomes of mammals.

Bovine Spongiform Encephalopathy (BSE): ►encephalopathies, ►Creutzfreldt-Jakob disease, ►Alzheimer disease, ►prions, ►scrapie

Bowel Disease (chronic inflammatory bowel disease): ►CIBD, ►CARD15, ►Crohn disease

Bowman-Birk Inhibitors: These are prepared from plant tissues and can bind and inhibit simultaneously or independently, trypsin and chymotrypsin. They may decrease cancer development in animals exposed to alkylating agents or displaying sporadic tumorous growth. (Witchi H, Espiritu I 2002 Cancer Lett 183[2]:141).

Bow-Tie: A system with various inputs and outputs connected by a node, resembling a bow-tie.

Box: Generally used for a consensus sequence in the DNA, such as a homeobox; domains of the internal control regions (box A, box, B, box C) that are the sequences where transcription factors bind. Sometimes protein boxes are also distinguished.

Box Genes: Clustered mutations in exons or introns (in mosaic genes of mtDNA). ►mtDNA

β-Pleated Sheet: Extended polypeptide chains in parallel or antiparallel arrangement linked by hydrogen bonds between the amino and carboxyl groups. ►protein structure

BP (B.P.): Before present, for archeological age.

bp: Base pair.

BPV: ►bovine papilloma virus

BR RNP (Balbiani ring ribonucleoprotein): Transcripts of the Balbiani ring and associated with about 500 protein molecules of a total molecular size of 106 daltons. ►Balbiani ring

Brachmann-De Lange Syndrome: ►De Lange syndrome

Brachydactyly: Abnormally short fingers and toes controlled by autosomal dominant genes (see Fig. B56). In Type E the metacarpus and metatarsus (the bones between the wrist and the fingers of the hand and the corresponding bones in the foot) are shortened. In still other types nervous defects, hypertension, shortening the bones of the arm accompanies the hand and foot problems. The expression may vary. Most commonly the middle bones (phalanx/phalanges) are affected (Type A); in some cases not all the fingers express the gene. In type B (9q22, receptor tyrosine kinase ROR2), in addition to the middle phalanges, the terminal ones are also short or absent. In type C more than 3 phalanges may appear. Type D involves short and flat terminal phalanges of the big toe and the thumb. In Type E the metacarpus and metatarsus (the bones between the wrist and fingers of the hand and the corresponding bones in the foot) are shortened. In still other types nervous defects, hypertension, shortening the bones of the arm accompanies the hand and foot bone problems. In an autosomal recessive form the brachydactyly involves also small head (microcephaly). In another recessive form primarily the great toe is affected but the proximal (near the wrist) joints do not move. The dominant brachydactyly with severe hypertension gene has been assigned to human chromosome 12p. Brachydactyly type A-1 is due to mutation in the Indian hedgehog gene. Brachydactyly type A2 (4q21-q25) is due to mutation in bone morphogenetic protein receptor

1B (Lehmann K et al 2003 Proc Natl Acad Sci USA 100:12277). ►polydactyly, ►syndactyly, ►cartilage, ►bone morphogenetic protein, ►Robinow syndrome, ►hedgehog; Schwabe GC et al 2000 Am J Hum Genet 67:822; Gao B et al 2001 Nature Genet 28:386.

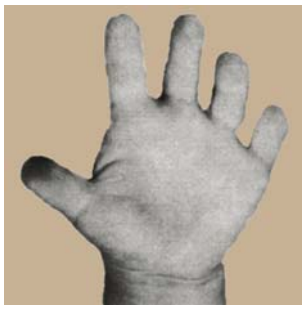

Figure B56. Brachydactyly

Brachymeiosis: When the second meiotic division is missing. ►meiosis

Brachyury: A homozygous (*TT*) dominant lethal (after 10 days of conception) gene in mice. The *Tt* heterozygotes are viable and have reduced tail (tailless), the homozygous *tt* also dies in 5 days. The different alleles of the complex locus have different effects of the development. The anomaly (chromosome 17) involves a genetic defect in the notochord development. The somites undergo differentiation but resorbed before birth. There are defects also in the posterior parts (limbs, allantois, umbilical vessels). The *t* alleles may display meiotic drive and from the male *t*/+ heterozygotes more than 90% of the progeny may receive the *t* allele. The distortion of the transmission (TRD) is controlled by at least six loci that do not normally recombine because of the presence of inversions. The *t* complex occupies about 1/3 of chromosome 17 and generally is inherited as a block because the region includes at least four inversions. The rare recombinants are called "partial *t* haplotypes." Females display normal transmission and fertility. In addition, there are at least 16 lethality loci within the *t* haplotype but these are not the primary causes of the distorted segregation. The Tcd1, Tcd2 and Tcd3 distorters act in response to the Tcr responders and can affect the transmission of any chromosome of mouse. The distorted segregation is due to the cis-acting so-called *T* complex responder (Tcr). The Tcr^t (acts only in cis) males avoid distortion and can fertilize the females but show the abnormal transmission. The distorted ratio is due to the Smok (sperm motility kinase) located at the C-terminus of the ribosomal Rsk3 kinase. Smok apparently phosphorylates the axonemal dynein of the microtubules. The T complex distorters (Tcd) are transacting factors, which increase transmission of the Tcr-bearing chromosome. Despite the preferential transmission of the *t* haplotype by the heterozygous males, the populations carry only 10–25% t haplotypes. The brachyury transcription factor is embedded by its carboxy-terminal into the minor groove of the DNA contacting a guanine residue but it is not bending the DNA. It is an important transcription factor for mesodermal specification. The *t* haplotype carries four Rho-GTPase-activating loci (*Tapgap-1*) whereas the wild type has only one. Brachyury-like anomalies occur also among cats, dogs, sheep, cattle and pigs (see Fig. B57). ►Manx in cat, ►meiotic drive, ►killer spore, ►somite, ►notochord, ►haplotype, ►axoneme, ►dynein, ►TCP-1, ►Holt-Oram syndrome; Schimenti J 2000 Trends Genet 16:240; Lyon MF 2003 Annu Rev Genet 37:393; Bauer H et al 2005 Nature Genet 37:969.

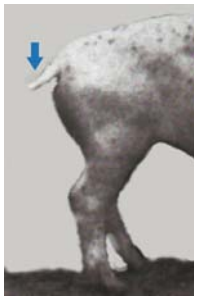

Figure B57. Short-tail pig

Bracken Fern (*Pteridium aquilinum*): Carcinogenic plant used as food in Japan.

Bract: A small, modified leaf from which flower may develop or a leaf on the floral axis subtending the flower (see Fig. B58).

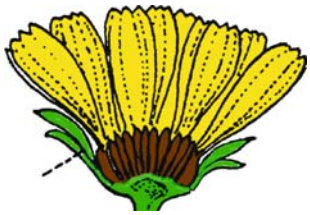

Figure B58. Bract

Bradford Method (Anal Biochem 72:248): The Bradford protein assay, for 1 to 100 µg protein. Prepare a standard solution (0.5 mg/mL) of bovine serum albumin (BSA) and make a dilution series 5 to 20 µL and dilute to 100 µL with 0.15 M NaCl. Prepare also 0.15 M NaCl blanks. Make a series of dilutions also from the unknown quantity of the protein to be

B

tested. Add 1 mL Coomassie brilliant blue to all and mix thoroughly. After 2 min determine absorption at 1-cm path-length at 595-nm wavelength in a spectrophotometer and extrapolate the concentration of the sample from the standard series. ►Lowry test, ►Kjeldahl method

Bradykinin: ►kininogen

Bradytelic Evolution: Evolution at very slow pace involving species whose adaptive environment extends over very long (geological) periods. In contrast, the tachytelic evolution is progressing at a fast pace whereas the horotelic evolution appears to show an average rate. ►evolution

BRAF: A cytoplasmic serine/threonine kinase that is mutant in 66% of melanomas but it occurs at lower frequencies in other cancers. Inhibition of MEK abrogates BRAF tumor growth (Solit DB et al 2006 Nature [Lond] 439:358). BRAF mutations occur in the majority of probands with the cranio-facio-cutaneous syndrome without an apparent increased frequency of cancer. ►RAS, ►MEK, ►cranio-facio-cutaneous syndrome, ►melanoma; Davies H et al 2002 Nature [Lond] 417:949.

Brahma: A catalytic component of the SWI/SNF chromatin-remodeling complex. ►nucleosome, ►chromatin remodeling, ►meCP2, ►Rett syndrome

Brain-Derived Neurotrophic Growth Factor: ►BDNF

Brain Diseases: ►Addison-Schilder syndrome, ►epiloia, ►mental retardation, ►affective disorders, ►craniofacial synostosis syndromes, ►prions

Brain, Human: A very complex structure and here only a few major landmarks are outlined as reference to several entries dealing with the central nervous system. The seven-layered hippocampus, consisting of "gray matter" is not shown although this is the most important area at the basal-temporal region involved in memory and learning. The functional areas of the brain can be identified by the increased blood flow upon stimulation by using positron emission tomography or functional magnetic resonance imaging. Microelectrodes applied to individual nerve cells reveal electrical activity at a single cell level. Since the mammalian brain contains thousands of distinct neuronal glial cell types, their synthesis requires separate transcription factors and the study of Gray PA et al (2004 Science 306:2255) found by in situ hybridization that that 349 genes in the brain displayed restricted expression pattern reflecting the anatomical organization of the mouse brain (see Fig. B59).

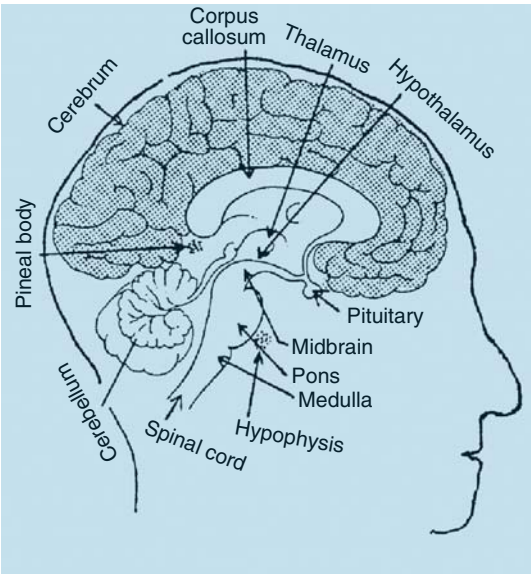

Figure B59. Major areas of the human brain

The new Allen Brain Atlas (ABA) allows one to view all the locations at which any of the more than 20000 genes in the mouse brain are activated, down to cellular-level resolution. The site allows one to search any anatomical region of the brain for any gene or combination of genes. The activated genes can be viewed with tools that can zoom from a whole-brain section down to a single cell while retrieving data at multiple levels of resolution (Markram H 2007 Nature [Lond] 445:160). The current technology visualizes the expression of single genes per cells but eventually larger the parts of the transcriptome might be visualized. Newly developed image-based informatics tools allow global genome-scale structural analysis and cross-correlation, as well as identification of regionally enriched genes. Unbiased fine-resolution analysis has identified highly specific cellular markers as well as extensive evidence of cellular heterogeneity not evident in classical neuroanatomical atlases. The new methods enable global analysis and mining for detailed expression patterns in the brain. The entire Allen Brain Atlas data set and associated informatics tools are available through an unrestricted web-based viewing application (Lein ES et al 2007 Nature [Lond] 445:168; http://www.brain-map.org).

The brain size of hominids (1,200–1,600 cm^2) approximately tripled in less than 3 million years of evolution. Brain volume and cognitive abilities seem to be positively correlated ($r = +0.4$) yet the correlation between brain size and cognitive performance is very low within families. The heritability of brain size of humans is very high and evolutionarily it has reached a plateau because the pelvic size of the mothers is restrictive in order to assure uncomplicated

child delivery. The positive correlation may be mainly non-genetic and influenced by socioeconomic status, cultural influences, etc. (See figure; ►memory, ►cerebellum, ►EQ, ►IQ, ►nerve function, ►tomography, ►nuclear magnetic resonance spectrography, ►human intelligence, ►brain scan, ►chimpanzee ►language, ►EPH, ►blood-brain barrier, ►microcephaly, ►encephalon; Nichols MJ, Newsome WT 1999 Nature Suppl 402:C35; brain evolution: Gilbert SL et al 2005 Nature Rev Genet 6:581; computer modeling of the brain: Herz AVM et al 2006 Science 314:81; neuronal networks: Destexhe A, Contreras D 2006 Science 314:85; computational models of cognition: O'Reilly RC 2006 Science 324:91; gene expression: http://www.loni.ucla.edu; http://www.loni.ucla.edu; mouse brain: http://www.mbl.org; expression pattern of ~21,000 mouse genes in the brain: http://www.brain-map.org/; http://www.brainatlas.org; http://www.trans.nih.gov/bmap; brain maps of different vertebrates: http://www.brainmaps.org/).

Brain Scan: Uses various sophisticated technologies such a functional magnetic resonance imaging (fMRI) and other technologies well as statistics to monitor sensorimotor and cognitive processes to shed light on human motivation, reasoning, emotions, possibility of deceptions and social attitudes. It reveals activation in different critical regions of the brain. It is becoming also a clinical tool of neurology. Some unresolved ethical issues still remain regarding the interpretation and validity of the data so obtained. ►brain human, ►tomography, ►nuclear magnetic resonance spectrography; Illes J et al 2003 Nature Neurosci 6:205; Check E 2005 Nature [Lond] 435:254.

Brain Stem: medulla + pons + midbrain. ►See diagram of brain

B-RNA: ►cowpea mosaic virus

Branch Migration: During the process of molecular recombination the exchange point between two fixed sites of the DNA single strands can move left or right when *the two single* strands are separated they can simultaneously reassociate in an exchanged manner in both double helices. This strand invasion brings about heteroduplexes. In *E. coli* the RuvA (a specificity factor) and RuvB (an ATPase) proteins (induced by ultraviolet radiation damage to the DNA) bind to the Holliday junctions and increase the length of the heteroduplex. RuvA and RuvB drive helical rotation of the DNA at the rate of about 8.3 bp/second (Han Y-W et al 2006 Proc Natl Acad Sci USA 103:11544). In eukaryotes, Rad54 and Rad51 operate homologous recombination at the Holliday junctures. Mismatches in the synaptic strands may interfere with branch migration. ►Holliday model, ►Holliday juncture, ►heteroduplex, ►recombination molecular mechanisms, ►mismatch repair; Ruv ABC; Walker box; Putnam CD et al 2001 J Mol Biol 311:297; Constatinou A et al 2001 Cell 104:259; Fabisiewicz A, Worth L Jr 2001 J Biol Chem 276:9413; Karymov M et al 2005 Proc Natl Acad Sci USA 102:8186.

Branch Length: of an evolutionary tree may be determined by the least square method or by using the maximum likelihood principle. ►evolutionary tree, ►least squares, ►maximum likelihood principle

Branch Point Sequence: Short RNA tract YNCURA Y, (Y stands for pyrimidine, R for purine and N can be either]) in the primary transcript near (18–38 base upstream) to the 3′ end of an intron (AG) of mammals. After exon1-intron boundary is severed and the intron end is released with a 5′-GU pair at the end, it forms then a loop as it folds back by **G** making a 2′→5′ bond with the A shown bold above. Subsequently, a cut is made at the 3′ end of the intron, the intron is released, and the exon1 is attached to exon 2. ►introns; Peled-Zehavi H et al 2001 Mol Cell Biol 21:5232.

Branched Chain Amino Acids: ►isoleucine-valine biosynthetic steps

Branched RNA: An intermediate of RNA splicing. ►introns

Branchio-Otorenal Syndrome (BOR): A human chromosome 8q13.3 dominant syndrome with incomplete penetrance and expressivity. It involves defects in the appearance of the ears, underdevelopment of the middle ear structures (malleus, incus, stapes) and the inner ear (cochlea) resulting in mild to severe hearing loss. This is accompanied by under-development of the kidney and the urinary tract. A variant form without the kidney symptoms maps to 1q31. The prevalence of BOR is $\sim 4 \times 10^{-4}$. The gene bears homology to the *Drosophila* gene *eyes absent* (*eya*). The 61.2 kDa Eya protein seems to be a transcriptional coactivator. (See Kumar S et al 2000 Am J Hum Genet 66:1715).

Brassica oleracea (cabbage, kale): Vegetable crops. Basic chromosome number is controversial 5 or 6 although cabbage is 2n = 18 (C genome) and there are some indications of being an amphidiploid. ►turnip, ►swedes, ►rapes, ►mustards, ►radish, ►watercress; Howell EC et al 2002 Genetics 161:1225; Lukens L et al 2003 Genetics 164:359; annotated genomes of the species: http://hornbill.cspp.latrobe.edu.au; http://brassica.bbsrc.ac.uk; *Brassica*, strawberry: http://bioinformatics.pbcbasc.latrobe.edu.au/index.htm.

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Brassica rapa: (*syn. B campestris*; mustard; genome AA; 2n = 20): Sequence-tagged linkage map is available (Kim JS et al 2006 Genetics 174:29).

Brassinolide: ►brassinosteroids (see Fig. B60).

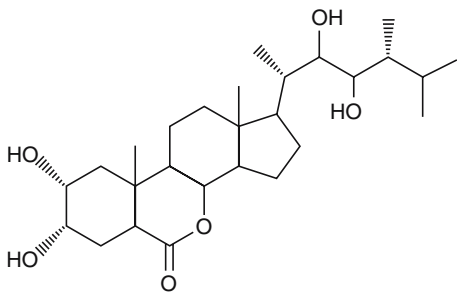

Figure B60. Brassinolide

Brassinosteroids (BR): Synthesized through the pathway campesterol→ campestanol→ cathasterone→ teasterone→ 3-dehydroteasterone→ typhasterol→ castasterone→ brassinolide. Uridine diphosphate glycosyltransferase mediates the glycosylation in the last two steps of this pathway in *Arabidopsis* (Poppenberger B et al 2005 Proc Natl Acad Sci USA 102:15253). The latter compound has been shown to remedy de-etiolation, derepression of light-induced genes, miniaturizing, male sterility and other symptoms of stress-regulated genes. These brassinosteroids bear close similarity to ecdysones, the animal molting hormones. The phytoecdysones were known for two decades in plants. All these plant hormones interact with each other in various ways and regulate signal transduction and gene activities. Unlike most animal steroid hormones, plant hormones are of small molecular size (except brassinosteroids) generally in the range of 28–350 Da. Brassinolide has a MW of about 480. A putative brassinosteroid receptor kinase shows similarities to the *ERECTA* and *CLAVATA1* gene products of *Arabidopsis* and share leucine-rich repeat with disease-resistance genes. Brassinosteroids bind to BRI1 leucine-rich receptor kinase in the outer plasma membrane (Kinoshita T et al 2005 Nature [Lond] 433:167). The *BES1* locus of *Arabidopsis* regulates the level of brassinosteroids and the response to light. BES1 interacts with the helix-loop-helix protein BIM1 and binds to an E box in some brassinosteroid gene promoters and regulates their transcription (Yin Y et al 2005 Cell 120:249). BESI is constitutively localizes to the nucleus and its activity there is modulated by BIN2 kinase (Vert G, Chory J 2006 Nature [Lond] 441:96). Another protein BZR1—when receives the brassinosteroid signal—turns some genes off although it controls homeostasis of BR (He J-X et al 2005 Science 307: 1634). BRI1 is a serine/threonine kinase cell surface receptor of brassinosteroids, and its coreceptor is BAK1. BKI1 (brassinosteroid receptor kinase inhibitor) is a negative regulator of signaling (Wang X, Chory J 2006 Science 313:1118). Removal of its C-terminus results in increased phosphorylation and increased activity. Ligand binding relieves inhibition of kinase activity (Wang X et al 2005 Developmental Cell 8:855). Ca^{2+}/calmodulin are important for brassinosteroid biosynthesis and growth (Du L, Pooviah BW 2005 Nature [Lond] 437:741). Brassinosteroids appear to be rate-limiting for auxin-responsive gene expression (Mouchel CF et al 2006 Nature [Lond] 443:458). ►plant hormones, ►de-etiolation, ►photomorphogenesis, ►hormones, ►steroid hormones. ►epidermis; Szekeres M et al 1996 Cell 85:171; Clouse SD, Sasse JM 1998 Annu Rev Plant Physiol Mol Biol 49:427; Neff MM et al 1999 Proc Natl Acad Sci USA 96:15316; Kang J-G et al 2001 Cell 105:625; Li J, Nam KH 2002 Science 295:1299; Yin Y et al 2002 Cell 109:181; Bishop GJ, Koncz C 2002 Plant Cell 14:S97; brassinosteroid signaling: Belkhadir Y, Chory J 2006 Science 314:1410.

BRCA1 (breast cancer antigen): An exclusively nuclearly located protein. ►breast cancer

BrdU: bromodeoxyuridine. ►bromouracil, ►hydrogen pairing, ►chemical mutagens

Breakage and Reunion: The broken chromatids or DNA single strands are broken at the position of chiasmata, and reunited in an exchanged manner during genetic recombination. This process is a physical event not requiring (normally) DNA replication as it was one time hypothesized with the copy choice idea. Recently, it was found that recombination takes place also by replication. ►recombination molecular models, ►recombination by replication, ►Holliday model; Creighton HB, McClintock B 1931 Proc Natl Acad Sci USA 17:492; Stern C 1931 Biol Zbl 51:547; Meselson M 1964 J Mol Biol 9:734.

Breakage–Fusion–Bridge Cycles: May cause variegation in the tissues because some of the genes may not be present in one of the daughter cells whereas the other cell receives two copies. If this dominant gene determines color, its presence is immediately recognized in the cell lineages. In telomerase-deficient and p53 mutant mice, epithelial cancer development is promoted by breakage-fusion-bridge cycles (see Fig. B61). In *Saccharomyces cerevisiae* dysfunctional telomeres may increase mutation rate ten to hundred fold. (See Fig. B62, ►*Ac-Ds*, ►cancer, ►telomerase, ►centromere silencing, ►p53; McClintock B 1941 Genetics 26:234; Hackett JA et al 2001 Cell 106:275).

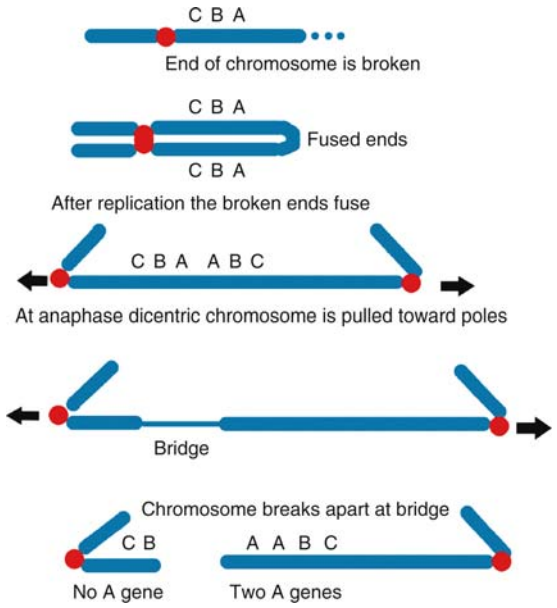

Figure B61. Breakage-fusion-bridge cycles may occur in the endosperm of maize plants if the end of the chromosomes is broken. The genetic and cytological consequences of such events are diagramed here. The relative size of the sectors (detectable when appropriate color markers are used) indicates the developmental time of the cycle. Early events involve large sectors, late events are indicated by small sectors. If the event occurs repeatedly, several sectors are observed. In case the two centromeres move toward the same pole no bridge is formed at anaphase and an intact dicentric chromatid is recovered. In case double bridge is formed and both chromatids break, two monocentric (most commonly defective chromosomes) go to the poles. The breakage-fusion-bridge cycle may not continue in the tissues of the growing plants because of apparent healing of the broken ends. The healing is attributed to the acquisition of new telomeres. (Photographs by courtesy of Barbara McClintock)

Breakpoint Mapping: ▶inversions

Breast Cancer (BRCA): Breast cancer is one of the most common diseases of women. The development of the disease proceeds through multiple steps. The premalignant stage is an atypical ductal hyperplasia. It progresses into the preinvasive stage of localized ductal carcinoma, which may change into invasive carcinoma, a potential lethal condition. Recently, for the better definition of these stages at the cell, rather than tissue level, microarray technology is combined with laser-capture microdissection. Although these different stages do not display significant differences in the pattern of gene expression, different grades of tumors are associated with distinct gene signature patterns and thus have predictive value regarding the condition of the disease (Ma X-J et al 2003 Proc Natl Acad Sci USA 100:5974). There is over 10% chance that the survivor to age 90 or over will develop breast cancer and ~5–10% of the cases are caused by mutation either in the BRCA1 or BRCA2 genes. Based on 22 studies involving 8,139 index cases, with unselected family with history of female (86%) and male (2%) breast cancers or epithelial ovarian cancers (12%) were evaluated. The average cumulative risks in BRCA1 carriers for breast cancer by age 70 were 65% and for ovarian cancer 39%. For BRCA2 the same risks were 45% and 11%, respectively (Antoniou A et al 2003 Am J Hum Genet 72:1117).

Deficiency of BRCA2 leads to impaired homologous recombination but maintains normal nonhomologous end joining (Xia F et al 2001 Proc Natl Acad Sci USA 98:8644). Fusion of Replication Protein A to BCR2 repeats reduced mutagenic recombination and promoted repair (Saeki H et al 2006 Proc Natl Acad Sci USA 103:8768). Even in non-hereditary (sporadic) cases of breast cancer, the BRCA1 gene is frequently lost or is inactive or rearranged. BRCA1, actually a tumor suppressor

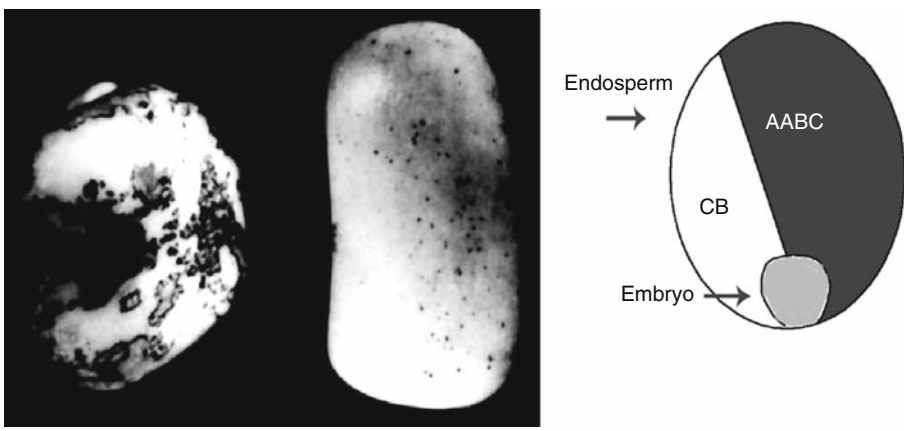

Figure B62. Left side is colorless. The right side is colored because of the AA gene

(in human chromosome 17q21), is responsible for 25% of the cases diagnosed before age 30. BRCA1 along with CREB is part of the pol II holoenzyme. It activates transcription when it is associated by its C-terminus with RNA helicase A and pol II. In BRCA1 of the 12 RNA polymerase subunits the activation of hRPB2 and hRPB10α are most critical. Predisposition to breast cancer is inherited as a dominant trait but the somatic expression (manifestation of cancer) requires that in these heterozygotes the normal allele would be lost or inactivated during the lifetime of the individual who inherited one BRCA susceptibility allele. The BRCA1 reading frame encodes 1,863 amino acids (22 exons) with a Zn finger domain at the NH_2 end. Amino acids 1,528 to 1,863, i.e., the C-terminal domain appear to be a transcriptional activator. The primary single transcript is 7.8 kb and is expressed primarily in the testis and the thymus but also in the breast and ovary. The transcript displays alternative splicing. Mice mutant in *Brca1* also develops increased risk for breast cancer because progesterone receptors are overexpressed in mammary epithelial cells. Treatment of *Brca1/p53*-deficient animals with the progesterone antagonist mifepristone can prevent tumorigenesis (Jovanovic PA et al 2006 Science 314:1467).

Gene Id4 is an important negative transcriptional regulator of BRC1 expression. BRCA1 is an important component of the 18-protein complex (SWI/SNF) involved in chromatin remodeling. BRCA1 strongly binds DNA and protects it from nucleolytic attack without sequence specificity and it is involved in double-strand DNA repair. BRCA1 binds to BRG1. As a transcription factor, it enhances the expression of several genes including p53. The BRCA1 sequences are well conserved in mammals but absent in chicken. The defects in the gene varies in the different kindreds from 11 bp deletion to frame shifts, nonsense, missense mutations or other alterations causing instability. The phenotype of the patients varies between the kindreds indicating that the specific mutations at the locus may affect its expression. It was noteworthy that there were female carriers of the mutation(s) who by age 80 failed to develop breast or ovarian cancer. Apparently, the expression is affected to some extent by extraneous genetic and environmental factors. Receptor-associated protein 80 (RAP80) is a BRCA1-interacting protein in humans. It contains a tandem ubiquitin-interacting motif domain, which is required for its binding with ubiquitin in vitro and its damage-induced foci formation in vivo. RAP80 specifically recruits BRCA1 to DNA damage sites and functions with BRCA1 in G2/M checkpoint control (Kim H et al 2007 Science 316:1202).

The BRCA1 protein may also be aberrantly localized in the cytoplasm and complicates the expression pattern. BRCA2 in human chromosome 13q12-q13 encodes 3,418 amino acids within a 6-cM region, is also a dominant early onset disease, responsible for about 45% of all *hereditary* breast cancers. Its highly conserved third exon is homologous to the c-Jun oncogene where the JNK protein binds. The 18–60 amino acid residues are potential activation sites. On both sides of exon-3 are inhibitory regions (IR1 & 2). It does not create a substantial risk for ovarian and other cancers but the chance for breast cancer in males may be slightly elevated, in contrast to BRCA1. Deletions 185delAG in BRCA1 and deletion 617delT in BRCA2 occur with carrier frequencies of 1.09% and 1.52%, respectively in Ashkenazy Jewish populations. The BRCA2 protein is cytoplasmically located. Its nuclear localization factor resides within the C-terminal 156 amino acids, and deletion in that region (e.g., 617delT) prevents the translocation of the protein to the nucleus and it consequently loses its ability to suppress tumorigenesis. The PALB2 protein normally binds BRCA2 and its mutation reduces its ability to bind and causes deficiency in homologous recombination and cross-link repair. Its mutation increases familial occurrence of breast cancer as well as prostate cancer (Erkko H et al 2007 Nature [Lond] 446:316). In 36% of the BRC families, chromosomal rearrangements (deficiencies, duplications) may be present (see Fig. B63).

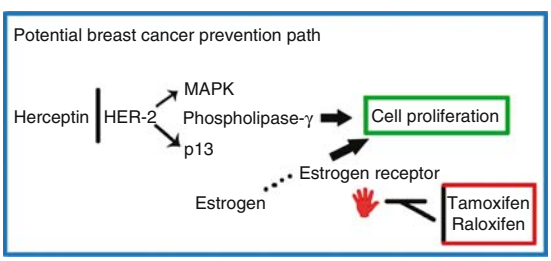

Figure B63. Breast cancer pathway. (Modified after Nass, S.J. *et al*. 1998 Nature Medicine 4:761)

The normal allele of BRCA1 transactivates the cyclin-dependent protein kinase inhibitor $p21^{WAF1/CiP1}$ without the cooperation of p53 and thus the entry into the S-phase of the cell cycle is prevented. This process, however, depends on the normal allele of p21. BRCA1 appears to be involved in transcription-coupled repair of oxidative DNA damage and the defective BRCA1 conveys hypersensitivity to ionizing radiation and hydrogen peroxide. Human Cds1 phosphorylates the BRCA1 protein at serine 988 after DNA damage and assists cell survival. The BRCA2 appears to be a cofactor of the radiation hypersensitivity genes Rad51 mediating double-strand DNA repair by homologous recombination or transcription-coupled repair upon phosphorylation by ATM or ATR (an ataxia telangiectasia mutated and RAD3-related protein). The BRCA2 tumor

suppressor is essential for error-free repair of double-strand breaks of the DNA. The repair is mediated by RAD51, which is attracted to the break-point by BRCA2. BRCA2 has eight ~30-amino acid repeats, which bind RAD51 and the ~700-amino acid domain binds to single-strand DNA. RAD51 elongates by polymerization the BRCA2-nucleated repairing DNA filament, which then ties the single strand to the double-strand DNA at the new junction (Yang H et al 2005 Nature [Lond] 433:653). BRCA2 has been localized to the midbody during cytokinesis and its defect results in chromosomal instability and aneuploidy (Daniels MJ et al 2004 Science 306:876). BRCA1 and BRCA2 proteins apparently interact in the cell. The Brc3 and Brc4 peptides of Brca2 protein and several low-penetrance genes are of potential importance. Homozygosity of mutant (truncated) BRCA2 (unexpectedly) also blocks cell proliferation and causes chromosomal breakage but mutations in spindle assembly checkpoint genes (p53, Bub1, Mad3L) relieve this growth arrest and restart (neoplastic) proliferation (Lee H et al 1999 Mol Cell 4:1). BRC2 (384 kDa) also involves double-stranded DNA repair (Yang H et al 2002 Science 297:1837). Normally BRC2 binds RAD51 protein, which mediates homologous pairing and recombinational repair. CDK-dependent phosphorylation of Ser 3291 site of RAD51 increases as cell proceeds toward mitosis. C-terminal damage of BRC2, however, may limit phosphorylation and interaction of BRC2 and RAD51 and leads to predisposition to radiation sensitivity and cancer because of the lack of repair (Ezashi F et al 2005 Nature [Lond] 434:598; Galkin VE et al 2005 Proc Natl Acad Sci USA 102:8537).

One of the remaining types of breast cancer is supposed to be due to a mutation in the *KRAS2* (Kirsten sarcoma) gene in human chromosome 6 when at codon 13 a G → A transition takes place resulting in Gly → Asp substitution. Ductal breast cancer was attributed to the loss of genes in human chromosome 1q21ter but also chromosomes 2, 14 and 20 were implicated. In families with high incidence of breast cancer a small secreted protein gene, expressed only in human breast cancer, was mapped to chromosome 21q22.3. BASE (**b**reast cancer **a**nd **s**alivary gland **e**xpression) a secreted protein (20q11,21), is associated with many breast cancers but not with normal tissues (Egland KA et al 2003 Proc Natl Acad Sci USA 100:1099). A dominant gene product serologically reacts with murine monoclonal antibody DF3. In the region of chromosome 17p13.3, the *TP53* regulator of tumor protein p53 was found but it could not be ruled out that a regulator exists 20 megabases telomeric to *TP53*. AIB1 steroid receptor (20q, member of the SRC-1 family oncogenes) is either amplified or overexpressed in the tumors. AIB1 seems to be a co-activator. Some observations suggest that loss of BRCA1 may lead to perturbation and destabilization of the inactive X chromosome and that explains why this disease occurs in females (Ganesan S et al 2002 Cell 111:393). Although BRCA1 and BRCA2 are the major suppressors of breast cancer, loss of heterozygosity at 1p, 1q, 3p, 6q, 7q, 8p, 11p, 13q, 16q, 17p, 17q, 18q, 19p, 21q and 22q may also affect the development of the cancer (Miller RJ et al 2003 Am J Hum Genet 73:748).

Some forms of breast cancer also affects males and the incidence of breast cancer in Klinefelter (XXY) men is almost as high as that in women. It appears the BRCA2 carrier males have increased risk for prostate and pancreatic cancer and possibly also bone and pharynx cancer (van Asperen CJ et al 2005 J Med Genet 42:711).

Some types of breast cancers are associated with cancers in other organs. FOXC2 transcription factor, which is involved in specifying mesenchymal cell fate during embryogenesis, is associated also with the metastatic capabilities of cancer cells. *FOXC2* expression is required for the ability of murine mammary carcinoma cells to metastasize to the lung, and overexpression of FOXC2 enhances the metastatic ability of mouse mammary carcinoma cells (Mani SA et al 2007 Proc Natl Acad Sci USA 104:10069). The risk of recurrence of breast cancer among first-degree relatives may be as high as 50%. In a transgenic mouse model, in the HER2-induced mammary tumors the increased level of the transcriptional repressor Snail, involved transition from epithelial-to-mesenchymal cells and higher relapse of cancer (Moody SE et al 2005 Cancer Cell 8:197). The risk of breast cancer is increasing with age, unmarried status, obesity, radiation exposure, etc. Several environmental chemicals (heterocyclic amines, o-toluidine, dibromoethane, glycidol, etc.) may cause breast cancer. In North America, the lifetime risk of breast cancer in females is about 10%. Of all breast cancer incidences about 5–10% is due to inherited causes. Genetic testing is now available for BRCA1 and BRCA2 for women who on the basis of family history have an increased risk for developing the disease. Unfortunately, individuals appearing negative in the standard test may be false negatives because rearrangements at these loci (~12%) remain undetected. Furthermore CHEK2, TP53 and PTEN mutations (~5%) also increase the risk, and members of the high-risk families should be tested for these mutations (Walsh T et al 2006 J Am Med Assoc 295:1379).

The incidence of breast cancer rises steeply between ages 25 to 50 and levels off after apparently because of the petering level of estrogens. Chromosomes 2q35 and 16q12 harbor estrogen receptor susceptibility (Hunter DJ et al 2007 Nature Genet 39:870). The development of breast cancer for BRCA1 and BRCA2 carrier females by age 70 is 28–87% and for ovarian cancer ~25–30%. The male

carriers also have an increased risk for breast, prostate, colon, pancreas, gall bladder, bile duct, stomach cancers and melanoma (Liede A et al 2000 Am J Hum Genet 67:1494). The contralateral occurrence of breast cancer is much higher than the prevalence of breast cancer in the general population. BRCA1 gene is responsible partly also for ovarian and prostatic cancers. DNA repair defect seems to be involved. BRCA1 forms a complex with proteins hRAD50-hMRE11-p95. BIC (breast cancer information core) address: http://www.breast-cancer-network.info/breast-cancer-information-core.html, environmental risk factors: http://envirocancer.cornell.edu/. (See the genes responsible for ►Peutz-Jeger syndrome, ►Cowden syndrome, ►androgen receptor, ►p53, ►ataxia telangiectasia, ►Muir-Torre syndrome, ►Li-Fraumeni syndrome and the ►Nijmegen breakage syndrome may also increase breast cancer risk, ►p53, ►p21, ►BRG1, ►BACH, ►hormone ►receptors, ►estrogen receptor, ►mifepristone, ►estradiol, ►AIB, ►abraxane, ►tamoxifen, ►raloxifen, ►herceptin; ►phospholipases, ►p13, ►HER-2, ►oncogenes, ►genetic screening, ►cancer, ►PARP, ►Li-Fraumeni syndrome, ►Nijmegen breakage syndrome, ►Fanconi anemia, ►multiple hamartoma, ►granin, ►Klinefelter syndrome, ►tumor suppressor, ►Stat, ►multiple hamartomas, ►Jun, ►JNK, ►RAD50, ►p95, ►RNA helicase, ►RNA polymerase, ►mouse mammary tumor virus, ►ZAG, ►ataxia telangiectasia, ►ATM, ►ATR, ►PTIP, ►Cdc1, ►contralateral, ►ovarian cancer, ►cyclin D, ►homologous recombination, ►Replication Protein A, ►laser-capture microdissection, ►microarray hybridization, ►midbody, ►NHEJ, ►plantibody, ►*Xist*, ►MammaPrint; Welcsh PL et al 2000 Trends Genet 16:69; Cui J et al 2001 Am J Hum Genet 68:420; Risch HA et al 2001 Am J Hum Genet 68:700; Nathanson K et al 2001 Nature Med 7:552; Welcsh PL, King M-C 2001 Hum Mol Genet 10:705; Nathanson KL, Weber BL 2001 Hum Mol Genet 10:715; Narod SA 2002 Narure Rev Cancer 2:113; Bhatia S, Sklar C 2002 Nature Rev Cancer 2:124; Chlebowski RT 2002 Annu Rev Med 53:519; susceptibility markers: http://cgems.cancer.gov/.

Breathing Of DNA: A reversible, short-range strand-separation below the melting temperature. ►melting temperature

Breeding System: Mating within a population may be random (each individual has an equal chance to mate with any member of the opposite sex) or it may be self-fertilization (in monoecious species) or inbreeding (in dioecious species). Also random mating and inbreeding both take place within the group. The term breeding system denotes these alternatives. The predominant breeding system of a few plant species is tabulated (A: apomictic, D: dioecious, I: self-incompatible, M: monoecious, O: outbreeding, S: selfing)

Table B6. Predominant breeding system in selected plant species

Alfalfa O-S	Cherry O-I	Mulberry D	Rubus O
Almond O-I	Chestnut O	Mustard O-I	Rye O-I
Alder M	Citrus O-I	Oak M	Rye grass O-I
Antirrhinum O-S	Clover O-I	Oat S	Sorghum O-S
Apple O-I	Coffee O-I	Oenothera O-S	Soybean S
Apricot O	Collinsia S	Onion O	Squash M
Arabidopsis S	Cotton O-S	Orchard grass O-I	Spinach D
Ash M-D	Cucumber-M	Osage orange D	Spruce M
Asparagus D	Date palm D	Parsley O	Stock S
Barley S	Datura S	Pea S	Strawberry D-S
Basswood O	Eggplant O	Peach O	Sugar beet O
Beach M	Elm M	Peanut S	Sugarcane O-I
Bean S	Fescue O-S	Pear O	Sunflower O-I
Belladonna O	Flax S	Petunia O-S	Sweetclover O-S
Beet O-I	Grape O	Pine M	Sweetpea S
Birch M-I	Hemp D-M	Pineapple O-I	Sycamore M
Blue grass O-A-S	Hop D	Plum O-I	Tea O
Broad bean S	Lentil S	Poplar D	Teosinte M
Brome grass O	Lespedeza O	Potato O-S-I	Timothy O
Buckwheat O-I	Lettuce S	Radish O-I	Tobacco S
Cabbage O-I	Lupine O-S	Ramie O	Tomato S
Carnation S	Maize M	Rape seed O-I	Tripsacum M
Carrot O	Maple O	Rice S	Triticale S
Castorbean M	Meadow foxtail O	Rose O	Walnut M
Celery O	Millet O-S	Rubber O	Wheat S

Breeding Value: The quantitative value a genotype, judged on the basis of the mean performance of the offspring. Actually, it is twice the mean deviation of the offspring from the mean of the parental population. The doubling is used here because each parent contributes a haploid gamete to the offspring and thus half of its genes. Breeders frequently call it the additive effect. The observed performance of individuals is called the *phenotypic value* (P) that is measured as the mean value of the population. The average value of two homozygotes is called the *midpoint*. It is equal to zero (only in case when the frequency of the two alleles is equal, 0.5) because the two parents deviate from it by a quantity of (+a) and (−a), by definition, and their sum cancel out each other. The *genotypic value* of the heterozygotes is designated by (d). In the absence of dominance d = 0, with complete dominance d = a, with overdominance d > a:

Table B7. Calculating breeding value

aa		0		*Aa*	*AA*
←	*−a*	→ ←		*+a*	→
		←	*d*	→	

The mean value (x) is usually calculated as the weighted mean, i.e., multiplied by the genotypic frequencies in the population. If the population is in equilibrium, the mean is:

$\overline{x} = p^2(a) + 2pq(d) + q^2(-a) + 2pq(d) = (a)(p + q)(p - q)(a) + 2pq(d)$ and because p + q = 1, $\overline{x} = (a)(p - q) + 2pq(d)$ and if several loci are involved, $\overline{x} = \sum [(a)(p - q) + 2pq(d)]$.
►gain; heritability, ►OMIA, ►Hardy-Weinberg equilibrium, ►merit; Falconer DS, Mackay TEC 1996 Introduction to quantitative genetics. Longman/Addison Wesley, White Plains, New York.

Brefeldin A (γ,4-dihydroxy-2-[6-hydroxy-1-heptenyl]-4-cyclopentanecrotonic acid λ lactone): An inhibitor of passing peptides from the endoplasmic reticulum and Golgi complex. In the absence of brefeldin, yeast cells suffer chromosome instabilities. ►ARF, ►translocase, ►ARNO; Wigge PA et al 1998 J Cell Biol 141:967; Lang BD et al 2001 Nucleic Acids Res 292567.

Bremsstrahlung (brake radiation, [from German words]): Electromagnetic radiation resulting from retardation or acceleration of a high-energy particle.

BRENDA: A database of at least 40000 enzymes and 6900 organisms. Nomenclature, reaction and specificity, structure, isolation and stability information is presented. ►Recon; ►http://www.brenda.uni-koeln.de.

BRF: The binding factor in RNA synthesis initiation.

BRG1 (BRAHMA related gene-product): A 1613 amino acid, DNA-dependent human ATPase, active in SWI/SNF, RSC mediating chromatin remodeling. ►chromatin remodeling, ►SWI/SNF, ►BRAHMA; Strobeck MW et al 2001 J Biol Chem 276:9273; Barker N et al 2001 EMBO J 20:4935; ►breast cancer

Bric: ►Byler disease

Bridge: Anaphase tie between separating centromeres in dicentric chromosomes (see Fig. B64). ►breakage-fusion-bridge, ►inversion

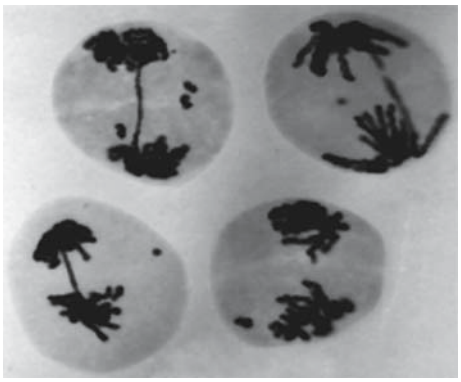

Figure B64. Bridge. Meiotic anaphase single and double bridges and 1 or 2 chromatid fragments resulting from 2 and 4 strand recombination in a paracentric inversion heterozygote (Courtesy of Dr. Arnold Sparrow)

BRIDGE: β-lactamase Reporter for Imaging Downstream gene expression. ►β-lactamase

Bridge Protein: Facilitates the interaction between viral particles and cell surface receptors.

Bridging Cross: If two genetically distant sexually incompatible species (A and B) are to be selected for gene transfer by sexual means. The problems can possibly be overcome by first mating one of the species with an intermediate compatible form (C) and then cross the hybrid (A × C) to the other (B). C serves as the bridge.

Bright Paramecia: ►symbionts hereditary

Bright-Field Microscopy: Ordinary light microscopy. ►dark-filed microscopy, ►fluorescent microscopy, ►phase contrast microscopy, ►Nomarski, ►stereomicroscopy

Brim: Break repair induced mutation. ►DNA repair, ►DNA repair

Bristle: ►chaetae

Britten & Davidson Model: This model was suggested as a working hypothesis in the 1960s for interpreting the processes involved in the regulation of eukaryotic

gene functions. The external stimuli were supposed to be directed to *sensor* genes that activated *integrator* genes that in turn transmitted the signals to *receptor* genes, which affected than the *structural* genes, coding for protein. These systems might have operated then in series of interacting batteries. (Britten RJ, Davidson EH 1969 Science 165:349; Britten RJ 1998 Proc Natl Acad Sci USA 95:9372).

BRM: Animal chromatin remodeling ATPases of ~1,600 amino acids. ►chromatin remodeling, ►BRAHMA, ►BRG1; Banine F et al 2005 Cancer Res 65:3542.

Brn: Eukaryotic transcription factors with POU domain controlling terminal differentiation of sensorineural cells. They are homologous to unc-86 in *Caenorhabditis*. ►POU, ►*unc-86*

Broad Bean (*Vicia faba*): 2n = 2x = 12 and large chromosomes has been used extensively for cytological research (see Fig. B65). It is an important crop in cool climates. ►favism, ►*Vicia faba* for karyotype

Figure B65. Broad bean

Broad-Betaliproteinemia: A hyperlipoproteinemia. ►apolipoproteins, ►hyperlipoproteinemia

Broad Sense Heritability: ►heritability

Brody Disease (ATP2A1): A rare recessive disease encoded in human chromosome 16p12.1-p12.2, and it involves impairment of muscle relaxation, stiffness and cramps in the muscles. The basic defect is associated with the muscle sarcoplasmic reticulum calcium ATP-ase (SERCA1). ►neuromuscular diseases, ►sarcoplasmic reticulum, ►Darier-White disease

Broken Tulips: Variegation (sectors) in the flowers is caused by viral infection (see Fig. B66). These sectorial tulips have commercial value in floriculture. In the seventeenth and eighteenth century the bulbs were so highly valued that they fetched gold of equal weight. ►symbionts hereditary, ►infectious heredity

Figure B66. Broken tulip

Bromodomain: A more or less cylindrical shape association of four helices of about 100 amino acids that form the docking sites in the chromatin for a large number of proteins (see Fig. B67). The lysine-acetylated H3 and H4 histones may be fitting into the bromodomains and are the conditions for gene transcription in eukaryotes. Bromodomains appear to anchor histone acetylase to the chromatin. ►chromatin remodeling, ►histones, ►histone methyltransferases, ►histone acetyltransferases, ►p300, ►SAGA, ►PCAF, ►SNF, ►TAF, ►$TAF_{II}250$; Ornaghi P et al 1999 J Mol Biol 287:1; Dhalluin C et al 1999 Nature [Lond] 399:491; Zeng L, Zhou M-M 2002 FEBS Lett 513:124.

Bromophenol Blue: ►tracking dyes

Bromouracil (BU): A pyrimidine base analog that is mutagenic in prokaryotes because it may lead to base substitution after tautomeric shift. When incorporated into eukaryotic chromosomes it may cause breakage upon exposure to light. On this basis, it has been successfully used as a selective agent in animal cell cultures. The non-growing mutant cells failed to incorporate it and survived while the growing (wild type) cells were killed upon illumination. Eosinophil peroxidase may produce 5-bromodeoxycytidine and the latter is incorporated into DNA as 5-bromodeoxyuridine, which by mispairing with guanine may become mutagenic. ►base substitution, ►tautomeric shift, ►hydrogen pairing, ►chemical mutagens, ►eosinophil; Benzer S, Freeze E 1958 Proc Natl Acad Sci USA 44:112.

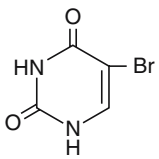

Figure B67. Bromouracil

Bronchitis: Inflammation of the air passages and lung.

Bronze Age: About 5000 years ago marked the development of crafts and urbanization.

Brood: The offspring of a single mating or the cluster of eggs (clutch) laid by a bird or reptile.

Brown Fat: Adipose tissue that mainly dissipates heat. Humans with phaeochromocytoma have large deposits of brown fat. ►adipocyte, ►pheochromocytoma

Brownian Ratchet: A hypothesis explaining transport of molecules into the mitochondria by Brownian movement (thermal agitation of molecules). Mitochondrial Hsp70-bound ATP complex associates with Tim proteins and nuclear encoded pre-proteins destined to mitochondria slide into import channels. The Mge1 protein initiates the ADP exchange and thus prevents backward movement and the protein is imported into the organelle. ►mitochondrial import, ►Mge1, ►ADP, ►ATP; Brokaw CJ 2001 Biophys J 81:1333.

Brownian-Zsigmondy Movement: Colloidal particles in solution may be in a continuously-agitated motion due to collision with the medium. It can microscopically be observed also in the cytosol of living cells.

Browsers: They provide comprehensive views by Internet access on genes, annotations, genomic regions, chromosomes and other important aspects of genomes. (UCSC Genome browser: http://genome.ucsc.edu; Hsu F et al 2005 Nucleic Acids Res 33:D54; National Center for Biotechnology: http://www.ncbi.nlm.nih.gov; Wheeler DL et al 2005 Nucleic Acids Res 33:D39; ENSEMBLE browser: http://www.ensembl.org; Hubbard T et al 2005 Nucleic Acids Res 33:D447; Revised and updated list appears annually in the first issue of Nucleic Acids Research).

Bruce: A 528 kD peripheral membrane protein of the trans-Golgi network and functions as an inhibitor of apoptosis. ►trans-Golgi network, ►apoptosis; Bartke T et al 2004 Mol Cell 14:801.

Bruce Effect: The termination of pregnancy in mice by olfactory influence on a pregnant female of a male that is genetically different from the inseminator. ►olfactogenetics, ►pheromones; Rajendren G, Dominic CJ 1987 Exp Clin Endocrinol 89:188.

Brucella: Bacteria responsible (for brucellosis) abortion and infertility of animals and serious febrile infection of male and female humans. *Brucella suis* 3.31 Mb and *B. melitensis* 3.29 Mb DNA have been completely sequenced. (See DelVecchio VG et al 2002 Proc Natl Acad Sci USA 99:443; Paulsen IT et al 2002 Proc Natl Acad Sci USA 99:13148; genome: http://bbrp.llnl.gov/bbrp/html/microbe.html).

Brugada Syndrome (SCN5A, 3p21): A dominant LQT-type heart disease (idiopathic ventricular fibrillation) due to defect in exon 28 of a sodium channel (SCN5A) gene. SUNDS (sudden unexplained nocturnal death syndrome) relatively common in Southeast Asia is controlled by an allelic gene. ►LQT, ►heart diseases, ►ion channels; Vatta M et al 2002 Hum Mol Genet 11:337.

Brush Border: A dense lawn of microvilli on the intestinal and kidney epithelium that facilitates absorption by increasing the surface. ►microvilli

Bruton's Tyrosine Kinase: ►BTK, ►agammaglobulinemia

Bryophytes: Mosses, liverworts and hornworts. They are green plants similar to algae but the organization of their body is more complex. Their gametangia is either unicellular or multicellular and show some cell differentiation. They usually have haploid and diploid life forms. The majority of them are terrestrial. ►alga

BS: ►Bloom syndrome

BSAP: ►pax

BSE (bovine spongiform encephalopathy): ►encephalopathies, ►Creutzfeldt-Jakob disease

BSL (biological safety level): BSL is specified by governmental regulations, BSL-1 is the minimal and BSL-4 is the most stringent, depending on the hazards involved.

bT: as a prefix for *Bos taurus* (bovine) DNA or protein.

BTB: Protein domains named for the *Drosophila* transcription factors Bric-a-brac (bab), Tramtrack (ttk) and Broad-Complex (BR-C) that associate with cullins, ubiquitin ligase and are required for the degradation of the meiotic spindle and the assembly of the mitotic spindle. BTB is an adaptor for the SCF complex. ►cullins, ►ubiquitin, ►SCF, ►spindle; Pintard L et al 2003 Nature [Lond] 425:311; Xu L et al 2003 Nature [Lond] 425:316.

BTF2: same as TFIIH. ►transcription factors

BTG: Antiproliferative protein encoded in human chromosome 1q32. Its synthesis is regulated by p53. ►p53

BTK (Bruton's tyrosine kinase): It belongs to a family of non-receptor tyrosine kinases. Its deficiency results in immunodeficiency by blocking differentiation of B lymphocytes. Syk and Lyn activate BTK with the mediation of BLNK. BTK phosphorylates transcription factor TFII-1 and loss of the TFII-1 gene results in Williams-Beuren mental defect. TFII-1 acts as a negative regulator outside the nucleus and interferes with calcium entry (Caraveo G et al 2006 Science 324:122). ►agammaglobulinemia, ►Sky, ►Lyn, ►B lymphocyte receptor, ►Williams syndrome; Liu W et al 2001 Nature Immunol 2:897.

βTrCP: A ubiquitin ligase, frequently associated with SCF and regulates by degradation of diverse

metabolic and developmental pathways. ►ubiquitin; Zhang J et al 2003 Proc Natl Acad Sci USA 100:14127.

BUB1, BUB2: These are checkpoint proteins arresting mitosis when the spindle attachment assembly is defective or there are problems with sister chromatid separation. ►cell cycle, ►meiosis I, ►MAD, ►p53; Krishnan R et al 2000 Genetics 156:489; Geymonat M et al 2002 J Biol Chem 277:29439; relation to p53: Gjoerup OJ et al 2007 Proc Natl Acad Sci USA 104:8334.

Bubble: A bubble is formed when at replication the two strands of the DNA separate or when transcription begins on DNA. Re-annealing the bubble at promoter clearance is called bubble collapse (Pal M et al 2005 Mol Cell 19:101). ►promoter clearance

Bubonic Plague: ►*Yersinia*

Buccal Smear: A sample of the epithelial cells from the inner surface of the cheek is spread onto microscope slides and used for rapid determination of the number of Barr bodies with 4–5% error rate. This procedure (mucosal swab) is non-invasive and the sampling is painless. The cells so sampled may also be used for DNA analysis by PCR after a rapid (ca. 1 hr) extraction. ►Barr body, ►PCR

Buckwheat (*Fagopyrum*): A feed and food plant (2n = 2x = 16). Eaten by animals or by humans, it may increase sensitivity to light and skin rash (see Fig. B68). ►favism

Figure B68. Buckwheat

Bud (budding protein of yeast): A cytoskeleton assembly-mediating protein family. Bud proteins associated with GTPase activating protein (GAP) are required for axial and bipolar budding patterns. ►Bni, ►cytoskeleton, ►profilin, ►polarity embryonic

Bud Scar: A chitin ring formed at the junction of the mother and daughter yeast cells that persists even after separation of the two cells. The number of bud scars may indicate the number of cell divisions (age) of the cell as well as polyploidy which is characterized by a different pattern of the bud scars.

Bud Sport: Genetically different sector (due to somatic mutation) in an individual plant (chimera).

Budding: Asexual reproduction by which the cell's cytoplasm does not divide into two equal halves, yet the bud after receiving a mitotically divided nucleus eventually reproduces all cytoplasmic elements and grows to normal size. Budding of enveloped viruses takes place by acquiring their membrane of lipid bilayer and proteins. They direct their surface glycoproteins into one or another type of cell membrane. They are pinching off either from the cell surface or into the cell lumen. The lipids of the bilayer come from the cell whereas the virus DNA specifies the proteins. ►*Saccharomyces cerevisiae*; retrovirus budding: Fisher RD et al 2007 Cell 128:841.

Budding Yeast: ►*Saccharomyces cerevisiae* (see Fig. B69).

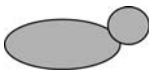

Figure B69. Budding yeast

BUdR (5-bromodeoxyuridine): Animal cells deficient in thymidine kinase enzyme are resistant to this analog. It is a base analog mutagen, substituting for thymidine and may cause mutation. Its mutagenic efficiency is low, especially in eukaryotes although if cells with BUdR substituted chromosomes are exposed to visible light, chromosome breakage is induced. When chromosomes with BUdR substitutions, at least in one of the strands, are exposed to low LET ionizing radiation, double-strand breaks occur in proportion of the amount of substitutions. The presence of reducing agents (e.g., e^{-}_{aq}•) favors breakage. ►bromouracil, ►base substitution mutation, ►sister chromatid exchange, ►hydrogen pairing

Buerger Disease: Autosomal recessive predisposition to thromboangiitis (inflammation of the blood vessels); its frequency is relatively high in some oriental ethnic groups.

Buffalo: Asiatic swamp buffalo (*Bubalus bubalis*) 2n = 48, the Murrah buffalo (*Bubalus bubalis*) 2n = 50, the African buffalos (*Syncerus caffer caffer*) 2n = 52, and the *Syncerus caffer nanus* is 2n = 54.

Buffer: A chemical solution capable of maintaining a level of pH within a particular range depending on the components of an acid-base system. Also a special storage area in the memory of a computer from where the information can be utilized at different rates by different programs; e.g., the printer can store information faster than it can print it.

Buffer Sequences: To avoid the loss of indispensable 5′ and 3′ tracts from linearized transforming DNA,

protective sequences may be added to the constructs. These buffers should not have cryptic splice or regulatory sites. Introns may be used.

Buffering, Genetic: A homeostatic mechanism that is supposed to maintain the function of genes at a certain level. ►Redundancy (duplications), ►feedback, ►epistasis, ►temperature-sensitivity, ►modifier genes, ►signal transduction, ►chaperones, ►apoptosis, etc. may mediate it. (Kitami T, Nadeau JH 2002 Nature Genet 32:191).

Bufo vulgaris (toad, 2n = 36): A primarily terrestrial small frog species and it lives in water environment during the mating season. (►*Rana*, ►*Xenopus*, ►frog, ►toad)

Build: The sequence of the human genome. The most complete sequence in 2004 is denoted as Build 35. (See Nature [Lond] 431:931).

Bulge: Unpaired stretches in the DNA. They are involved in binding of regulatory protein domains, in enzymatic repairs, slipped mispairing in the replication of microsatellite DNA, intermediates in frame shift mutations and essential elements for naturally occurring antisense RNA ►DNA repair, ►binding proteins, ►mispairing, ►frame shift mutation, ►microsatellite, ►antisense RNA

Bulimia: A psychological disorder involving excessive eating and self-induced vomiting based on serotonergic abnormality. ►anorexia, ►obesity, ►serotonin, ►addiction; Bulik CM et al 2003 Am J Hum Genet 72:200.

Bulked Segregant Analysis: It is used in mapping recombinant inbred lines. The individuals in the population are identical at a particular locus but unlinked regions in the chromosomes are represented at random. ►RAPD

Bulk-Flow Model: It postulates that targeting signals does not regulate export of molecules from the endoplasmic reticulum. Experimental evidence indicates, however, that even the constitutively excreted proteins carry some target specificities. ►endoplasmic reticulum

Bulking: A plant breeding procedure when selection of segregants after a cross is delayed to later generations when the majority of individuals become homozygous by continued inbreeding. The number of heterozygotes by Fn is expected to be 0.5(n−1) for a particular locus where *n* stands for the number of generations selfed. Note: the F_1 is produced by crossing therefore we use n − 1. ►inbreeding progress of, ►inbreeding rate

Buller Phenomenon: When from dikaryotic mycelia nuclei may move into monokaryotic ones. ►di-mon

Bullous Pemphigoid Autoimmune Disease: A human chromosome 6p12-p11 dominant autoimmune disease manifested as vesicles on the skin. It is based on a defect involving the ca. 230 kDa glycoprotein (antigen BPAG1e). This antigen affects also the nerve fibers. ►filament, ►autoimmune disease

Bundle: Protein α-helices running along the same axis. ►protein structure

Bundle Sheath: Cells wrapped around the phloem bundles. Their chloroplasts do not show grana and synthesize carbohydrates through the C3 pathway although the other chloroplasts in the same plant operate by the C4 system. ►chloroplasts, ►C3 plants, ►C4 plants

Bungarotoxin: ►toxins

Buoyant Density: A molecule (e.g., DNA) suspended in a salt density gradient (such as a CsCl solution, spun for 24 hr at 40,000 rpm in an ultracentrifuge tube) comes to rest in the salt gradient at the position where the medium (CsCl) density is identical to its own. The buoyant density of, e.g., *Chlamydomonas* nuclear and chloroplast DNA is 1.724 and 1.695, respectively. Higher buoyant density reflects higher G + C content in the DNA. The refractive index determined by a refractometer, capable of 5-digit resolution, can be used to determine the density (ρ) of a cesium chloride solution in any sample withdrawn from the centrifuge tube. The relevant relationships at 25°C are the following:

Table B8. Determining Buoyant Density

CsCl Weight%	Density g/mL	Refractive Index	Molarity
50	1.5825	1.3885	4.700
55	1.6778	1.3973	5.481
56	1.699	1.3992	5.651
57	1.7200	1.4012	5.823
58	1.7410	1.4032	5.998

The density of *E. coli* DNA is about 1.710 and that of *Mycobacterium phlei* is 1.732 and a deoxy A-T polymer has a (ρ) value of 1.679. Since none of these values are directly readable from the tabulation, interpolation is required that can be done by graphic representation. ►density gradient centrifugation

Buphthalmos: ►glaucoma

Burbank, Luther (1849–1926): American breeder credited with the production of over 800 new varieties and strains of plants, mainly in Santa Rosa, California. In addition, his success had a stimulating

effect on the development of plant breeding. Regrettably, his mystic, Lamarckian ideas also hindered scientific plant breeding. (See Crow J 2001 Genetics 158:1391).

Burden of Proof: The responsibility of proving the validity of an assertion, e.g., that a genetically modified organism or a drug is hazardous or harmful.

Burdo: A "graft hybrid" between tomato and nightshade (*Belladonna*), forming a periclinal chimera. The graft hybrids were assumed to be the result of fusion between the cells of different species combined at the site of the grafting. From this site then somatic hybrid cells regenerated into plants. ►periclinal, ►somatic cell hybrids; Winkler H 1907 Ber Dtsch Bot Ges 25:568.

Burkitt's Lymphoma: A human cancer caused frequently by the Epstein-Barr virus is most common in central Africa but occurs in other parts of the world, involved in nasopharyngeal (nose and throat) carcinoma and neoplasias of the jaws and the abdomen. It frequently involves a translocation between human chromosomes 8 (c-myc oncogene) to 14 (immunoglobulin promoter). The receptor of the Burkitt's lymphoma is activated by a chemokine that is targeted to B cells in the lymphoid follicles. The Epstein-Barr virus positive lymphomas display elevated amount of reactive oxygen species (ROS), a likely contributing factor to cancer (Cerimele F et al 2005 Proc Natl Acad Sci USA 102:175). ►Epstein-Barr virus, ►chemokine, ►B cell, ►lymphoma, ►methylation of DNA, ►immunoglobulins, ►myc

Bursa: Generally a sac like pouch; the bursae Fabricius are located in the intestinal tract of birds and produce the B lymphocytes. ►lymphocytes

Burst Size: The average number of phage particles released by the lysis of bacteria.

Bus: It is the circuit system in a computer that transmits information within the hardware or cables that link together various devices with the computer.

Bushmen: A designation for nomadic people who live in the wilderness (bush), such as the Australian aborigines or people in the Kalahari Desert. The anthropological characteristics are shared within the group but there is no known evolutionary relationship among the Bushmen inhabiting different geographical regions.

Busulfan ($C_6H_{14}O_6S_2$, most common trade name myleran): It is antineoplastic/carcinogenic chemical (see Fig. B70). It is a mitotic germ cell toxicant; eliminates primordial follicles. It is also a chemosterilant pesticide. ►Myleran

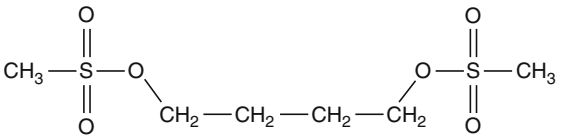

Figure B70. Busulfan

Buzzword Index (BWI): BWI indicates the varying trends of use of certain terms in the science literature.

$$\mathrm{BWI} = Ln(n)X\frac{(n+1)/(N+1)}{(n^*+1)/(N^*+1)}$$

Where n is the number of times the term was mentioned in the past year and n* is the number of times the word was mentioned during the past 10 years. N and N* are similarly calculated on the basis of the occurrence of the word "biology" and this normalizes the score with respect to the general growth of biology papers. (Adopted from Jensen LJ et al 2006 Nature Rev Genet 7:119).

BvgS: *Bordetella pertussis* (bacterial) kinase affecting virulence regulatory protein BvgA. ►pertussis toxin

Bypass Replication: It is capable of bypassing a DNA defect and continues the process beyond it. It does not lead to permanent repair. ►DNA repair

Byr 2: A serine-threonine kinase of *Schizosaccharomyces pombe*. An analog of RAF. ►raf, ►signal transduction, ►serine/threonine kinase

Byler Disease (PFIC1, 18q21): Progressive intrahepatic cholestasis. It is apparently allelic to the benign recurrent intrahepatic cholestasis (BRIC). Bile acid secretion is defective due to defect(s) in the ATP-binding cassette (ABC) transporter. ►cholestasis, ►Alagille syndrome, ►ABC transporter

Bystander Activation: A hypothesis for the origin of autoimmune reaction. Viruses may induce inflammation resulting in cytokine production. The cytokines may reactivate dormant T cells with low activation threshold and these T cells may then attack self-antigens that normally escape their attention. Non-malignant immune cells infiltrating follicular lymphoma have profound effect on the development of this type of cancer (Dave SS et al 2004 New England J Med 3561:2159). ►self antigen, ►immune response, ►autoimmune disease, ►immune tolerance, ►bystander effect; Fournie GJ et al 2001 J Autoimmun 16:319.

Bystander Effect: Genetic vectors targeted to a particular cell type do not spread to neighboring cells, yet their synthesized transgene product (e.g., a toxin) may diffuse and also kill the surrounding cells. The bystander effect is a complex result of intercellular communication through gap junctions, apoptotic cell

death, release of cytokines, blocking of angiogenesis, etc. ►gene therapy, ►cancer gene therapy, ►Mo-MulLV, ►transgene, ►gap junctions, ►cytokines, ►apoptosis, ►angiogenesis, ►vectors, ►radiation response, ►radiation hazard; Zheng X et al 2001 Mol Pharmacol 60:262.

Byte: A computer unit of information consisting of a number of adjacent bits. Most frequently 1 byte is 8 bits that represent a letter or other characters that the computer uses. ►bit

bZIP: basic leucine zipper. ►leucine zipper, ►DNA-binding protein domains

B-Zip Protein: A DNA binding, protein contains a basic amino acid zipper domain. ►binding proteins

Historical vignette

William Bateson, the most ardent Mendelian, among many of his original contributions, discovered that the interactions among gene products modify the Mendelian ratios without compromising the validity of the basic principles. In 1926 TH Morgan eulogized Bateson with these words: "His intellectual rectitude was beyond all praise and recognized by friend and foe alike" (*Science* **63**: 531).

Bateson enjoyed popularity in America and was personally familiar with most of his American colleagues. In 1922, at the University of Pennsylvania, he concluded a memorial lecture with the following warning:

"I think we shall do genetical science no disservice if we postpone acceptance of the chromosome theory in its many extensions and implications. Let us distinguish fact from hypothesis. It has been proved that, especially in animals, certain transferable characters have a direct association with particular chromosomes. Though made in a restricted field this is a very extraordinary and most encouraging advance. Nevertheless the hope that it may be safely extended into a comprehensive theory of heredity seems to me ill-founded, and I can scarcely suppose that on a wide survey of genetical facts, especially those so commonly witnessed among plants, such an expectation would be entertained. For phenomena to which the simple chromosome theory is inapplicable, save by the invocation of a train of subordinate hypotheses, have been there met with continually, as even our brief experience of some fifteen years has abundantly demonstrated" (*J. Genet*. **16**: 201 [1926]).