**J Base** (β-D-glycosylhydroxymethyluracil): This occurs in the repetitive DNAs of protozoa and its presence is correlated with a J-binding protein and with the epigenetic silencing of telomeric surface glycoprotein genes (see Fig. J1). These surface glycoproteins mediate antigenic variations of Trypanosomes and other related parasites. ▶pyrimidine, ▶antigenic variation, ▶Trypanosomes; Sabatini R et al 2002 J Biol Chem 277:958; Yu Z et al 2007 Nucleic Acids Res 35:2107.

![Figure J1. J base](image)

**J Chain**: A 15-kDa polypeptide that is involved in the formation of antibody molecules. ▶immunoglobulins

**J Chromosome**: A J-shaped chromosome moving toward the pole (see Fig. J2).

![Figure J2. J chromosome](image)

**J Gene**: ▶immunoglobulins, ▶J chain

**J Protein**: ▶HSP

**JAB** (Jun activation-domain-binding protein): A co-activator of AP1 transcription factor functioning by transactivating c-Jun and JunD. It interacts with the β2 subunit of LFA-1. It may switch off cytokine signaling. ▶AP, ▶Jun, ▶transactivator, ▶SOCS-box, ▶CIS, ▶LFA; Harding TC et al 2001 J Biol Chem 276:4531.

\[
\tilde{\theta} = n\hat{\theta} - (n - 1) \frac{\sum_{i=1}^{n} \hat{\theta}_i}{n}
\]

**Jackknifing**: A statistical device for the estimation of bias and variance of genetic parameters without providing essential estimates on the distribution of the estimates. The jackknife estimator of a parameter is presented here where \(\hat{\theta}\) is the usual estimator using the complete set of \(n\) observations. In the jackknife procedure each sample member in turn is omitted from the data, thus generating \(n\) separate samples each of \(n - 1\) size. This method may be used for the estimation of the size of misclassification in conjunction with discriminant analysis. ▶discriminant function, ▶bootstrap; LaPointe FJ et al 1994 Mol Phylogenet Evol 3(3) 256.

**Jackpot Mutation**: This occurs early during the growth of a population and is represented by more copies than mutations, which occur late. Jackpot mutations may bias the calculations of the mutation frequency if not identified. ▶mutant frequency

**Jackpot Vessel**: In a series of dilutions or in a fluctuation test one vessel has more than the expected number of cells caused either by a clump of cells or a pre-existing mutation. ▶fluctuation test

**Jackson Laboratory Backcross DNA Panel Map Service**: This makes available DNA from the reciprocal mouse crosses (C57BL/6J x Mus spretus), characterized by SSLP markers, proviral loci and several other sequences. Information: Lucy Rowe or Mary Barter, Jackson Laboratory, 600 Main Str., Bar Harbor, ME 04608, USA. Phone: 207-288-3371 ext. 1687. Fax: 207-288-5079, lbr@aretha.jax.org (L. R.) or meb@aretha.jax.org (M. B.)

**Jackson-Lawler Syndrome**: is a keratosis of the skin and may involve teeth already at birth. It is caused by mutations at two chromosomal loci 17q12-q21 and 12q13. ▶keratosis, ▶ichthyosis

**Jackson-Weiss Syndrome**: ▶Crouzon syndrome, ▶Pfeiffer syndrome, ▶Apert syndrome

**Jacobsen Syndrome**: This dominant fragile site involves human chromosome 11q23.3 and is located at a distance of 100 kb from the CBL2 oncogene and CCG repeats. This trinucleotide repeat is also called FRA11B. The CpG repeats are liable to methylation. It involves growth and psychomotor retardation, anomalies of the face, finger and toe development. ▶fragile sites, ▶FMR1, ▶trinucleotide repeats, ▶Huntington’s chorea, ▶ataxia, ▶Machado-Joseph disease, ▶Kennedy disease, ▶dentatorubral-pallidolysian atrophy

**Jak Kinases** (Janus tyrosine kinases): Jak3 is required for the progression of the development of B lymphocytes. Jak kinases are involved in the transmission of interleukin signals. ▶signal transduction by interferon signaling, ▶interleukins,

**Jak-STAT Pathway:** Several Jak kinases, signal transducers and activators of transcription (STATs) regulate the signal transduction of interleukins and interleukin-mediated transcription. The pathway may be activated by interferons, phospholipase C (PLC), growth hormones, epidermal growth factor (EGF) and platelet-derived growth factor (PDGF). The SOCS/JAB/SSI and CIS proteins exert negative control. The Jak-STAT pathway regulates heterochromatin in the cell. Over-expression of Jak can lead to tumorigenesis in _Drosophila_ and disruption of the pathway can suppress tumorigenesis (Shi S et al 2006 Nature Genet 38:1071). This pathway is missing from _Caenorhabditis_.

**Jamaican Vomiting Sickness:** This is caused by the consumption of unripe ackee fruit, a common food of the people of the island. The obnoxious component of the fruit hypoglycin A may reduce blood glucose content to 10 mg/100 mL and may even cause death. The compound is a specific inhibitor of isovaleryl-CoA dehydrogenase, and isovaleric acid accumulates in the blood leading to depression of the central nervous system. The poisoning has a similar effect as human isovaleric acidemia. ▶sovaleric acidemia


**Jansky-Bielschowsky Disease:** ▶ceroid lipofuscinosis

**Janus Kinases:** These include Jak kinases and Tyk2; they are non-receptor tyrosine phosphorylating enzymes. ▶Jak

**Jarcho-Levin Syndrome:** ▶spondylocostal dysostosis

**Jarovization** (yarowization): ▶vernalization

**Jasmonic Acid** ([±]-1α,2β-[Z]-3-oxo-2-[2-pentenyl]cyclopentanc acid): is a fatty acid derivative protease inhibitor in plants and an activator of stress response genes in case of infection or wounding. Furthermore it controls a number of developmental processes (see Fig. J3). Jasmonate regulates catabolism of amino acids in the gut of herbivores and plays a role in protection against insects (Chen H et al 2005 Proc Natl Acad Sci USA 102:19237). Around 20 jasmonates and their conjugates perform complex and far-reaching regulatory roles. Several genes have been identified in the pathways. Jasmonate signaling appears to mediate long distance information transmission; systemic transcriptional response shares an extraordinary overlap with the local herbivy and wounding responses, indicating that jasmonates may be pivotal to an evolutionarily conserved signaling network that decodes multiple abiotic and biotic stress signals (Truman W et al 2007 Proc Nat Acad Sci USA 104:1075). JAZ proteins (jasmonate ZIM domain proteins) function as repressors of jasmonate signaling and are degraded through the SCFCOI1-dependent 26S proteasome pathway. Protein–protein interaction studies indicate that jasmonoyl-isoleucine (JA–Ile) specifically promotes COI1–JAZ1 interaction in the absence of other plant proteins (Thines B et al 2007 Nature [Lond] 448:661; Chini A et al 2007 Nature [Lond] 448:666). ▶plant defense, ▶wound response, ▶insect resistance in plants, ▶SCF; Seo HS et al 2001 Proc Natl Acad Sci USA 98:4788; Turner JG et al 2002 Plant Cell 14: S153; Gfeller A, Farmer EE 2004 Science 306:1515.

**Figure J3.** Jasmonic acid

**Jaspar:** This is the eukaryotic transcription factor binding profile database: http://jaspar.cgb.ki.se.

**Jaundice** (icterus): This may be caused by hyperbilirubinemia and is characteristic of several hereditary syndromes. ▶kerenicterus, ▶hyperbilirubinemia

**Java:** This is a commercially available computer language for various applications.

**Java Man:** This is a commercially available computer language for various applications.

**Java Man:** A representative of _Homo erectus_ with a small cranian (brain = 815–1067 cm²) and robust jaws who lived about 100,000 years ago. ▶hominids

**JE:** PDGF (platelet-derived growth factor) and serum-inducible cDNA. ▶PDGF

**Jefferson, Thomas:** President’s paternity. ▶Y chromosome

**Jellyfish:** http://www.ucis.uci.edu/biochem/steele/default.html.

**Jervell and Lange-Nielsen Syndrome:** This is 21q22.1-q22.2 and 11p15.5 recessive heart and auditory (deafness) syndrome. In the electrocardiograms the interval Q - T is prolonged. In this method the excitation of the heart atriun is denoted by the P wave, followed by the QRS complex of deflections.
and excitations (depolarization) of the ventricles, and the T waves indicate the repolarization of the ventricles. Fibrillations (uncoordinated arrhythmia) of the heart atrial muscles are also observed as a consequence of deficiency of potassium and/or sodium ion channels. Sudden death may occur.

- heart disease
- deafness
- electrocardiography
- LQT
- HERG
- Ward-Romano syndrome
- Beckwith-Wiedemann syndrome

**Jesuit Model:** There are more potential replicational origins than actually selected in eukaryotes. replication bubble

**Jews and Genetic Diseases:** Common diseases among Ashkenazi Jews are Riley-Day syndrome, Tay-Sachs disease, Gaucher’s disease, Niemann-Pick syndrome, diabetes mellitus, pentosuria, dystonia and colorectal cancer. About 1% of women carries deletions at various positions in the BRCA1 and BRCA2 breast cancer genes, Cohen syndrome, Canavan disease, pentosuria and PTA deficiency disease. Diseases that are rare in this group include juvenile form of Gaucher’s disease, Glucose-6-phosphate dehydrogenase deficiency and, Bloom’s syndrome. A common disease among Sephardic Jews is Mediterranean fever, whereas Tay-Sachs disease is uncommon. Among Oriental Jews of Persian origin, hypochondroplasmosis and Dubin-Johnson syndromes are relatively common. In Libyan Jewish populations, Creutzfeldt-Jakob disease is disproportionately common. There is no valid explanation for these differences in the incidence of diseases (gene frequencies). It has been suggested that genetic drift in small isolated populations may be the cause. There are no selective advantages of the heterozygotes, specific for these particular populations has also been considered. See diseases at separate entries.

**JIL-1:** A chromosomal kinase which may upregulate gene expression in the single Y chromosome of Drosophila male.

**Jimpys Mice:** This is a special strain of mice with a lower rate of cerebrosidase synthesis resulting in neurological defects. cerebrosidase

**Jimson Weed:** Datura stramonium (see diagram of seed capsule) (see Fig. J4).

![Figure J4. Datura](image)

**JIP:** This is a JNK-interacting protein.

**JNK** (Jun amino terminal kinase): This kinase acts on the amino terminal of Jun oncogenes and other transcription factors. It is the same as SAPK. It belongs to the MAK family of protein kinases that are activated by stress (environmental stress, heat shock, tumor necrosis factor, etc.). SAPK appears to be inhibited by p21, a transforming protein. The activated JNK stimulates the transcriptional activity of AP1. The JNK interacting protein-1 (JIP-1) causes the retention of JNK in the cytoplasm and thus inhibits JNK-regulated gene expression. JNK signaling activates CD4 helper T cells (T CD4), which during clonal proliferation release interleukins and become T1 effector cells and mediate inflammatory responses. JNK is also involved in the mitochondrial release of cytochrome c and the apoptotic path. Obesity increases JNK activity and JNK1 deficiency results in reduced adiposity and improves sensitivity to insulin (Tuncman G et al 2006 Proc Natl Acad Sci USA 103:10741).

- aging

**Jockey:** non-viral retrotransposable elements

**Johanson-Blizzard Syndrome** (JBS; 15q14-q21.1): This is a recessive pancreatic (UBR1) insufficiency disease characterized by nasal wing defect (aplasia), facultative scalp defects, imperforated (closed) anus,
deafness, hypothyroidism, dental defects, genitourinary malformation and generally mental retardation. In the absence of the UBR1 function intruterine pancreatitis arises. The rate of prevalence is ~1/250,000. The UBR1 gene encodes at least four overlapping E3 ubiquitin ligases at the N-end rule pathway. ▶ ubiquitin, ▶ N-end rule, ▶ pancreatitis; Zenker M et al 2005 Nature Genet 37:1345.

**Joining of DNA:** ▶ ligase, ▶ blunt-end ligation, ▶ cohesive ends

**Joint Probability:** When two events are independent from each other, the probability of their joint occurrence can be obtained by multiplying the independent probabilities. The same rule also applies to more than two independent frequencies. Independence means that the occurrence of one has no bearing on the occurrence of the other(s). ▶ probability

**Josephine Domain:** This is a part of the ataxin-3 protein responsible for the neurodegenerative Machado-Joseph syndrome. Ataxin-3 functions as a polyubiquitin chain-editing enzyme as the Josephine domain is followed by an ubiquitin-interacting motif in spinocerebellar ataxia 3 (Mao Y et al 2005 Proc Natl Acad Sci USA 102:12700). ▶ Machado-Joseph syndrome; Nicastro G et al 2005 Proc Natl Acad Sci USA 1032:10493.

**Jost Factor:** ▶ Müllerian inhibitory substance

**Joubert Syndrome:** Heterogeneous, autosomal recessive developmental defect of the human brain (cerebelloparenchymal disorder, cerebellar vermis agenesis) has been traced to human chromosome 9q34.3. The syndrome with oculo-renal defects is located to chromosome 11p12-q13.3 (Keeler LC et al 2003 Am J Hum Genet 73:656). Joubert and Meckel syndromes are associated with cilius dysfunction (Delous M et al 2007 Nature Genet 39:875). ▶ Meckel syndrome, ▶ cilia

**Joule:** 1 joule = 10⁷ ergs, the energy expended per 1 second by an electric current of 1 ampere in a resistance of 1 ohm; approximately 0.24 calorie.

**Jüberg-Marsidi Syndrome:** Xq12-q21 mental retardation, growth and developmental anomaly are based on mutation in a helicase. The same protein appears to be involved in X-linked α-thalassemia and mental retardation. ▶ thalassemia, ▶ mental retardation, ▶ ATRX

**Judassohn-Lewandowsky Syndrome** (pachyonychia congenita, PC1, 17q12-q21, 12q13): This is a hereditary recessive keratosis of the nails (onchogryposis), palm, sole and mouth. It is due to mutation in keratin 16. ▶ keratosis, ▶ ichthyosis

**Judgment** (judicium): Denotes the power to arrive at a valid decision regarding facts that may not be fully understood. It is related to intuition, which means arriving at an understanding without conscious reasoning. Subjective expert judgment is the most important human quality in many areas of human activity. An important aspect of subjective judgment is its truth that can be assessed by Bayesian methods. ▶ Bayes’ theorem; Prelec D 2004 Science 306:462.

**Jukes-Cantor Estimate of Evolutionary Divergence:** This is based on the number of nucleotide substitutions since the separation of two DNA sequences during evolution. D = 2at where D = distance, a = the probability (p) that one nucleotide is replaced in time t. The separation in time t = D/2a. ▶ evolutionary distance; Chen FC et al 2001 J Hered 92:481.

**Jump Stations:** A collection of links for genetic and biological information regarding databases, journals, news groups, etc. ▶ databases [general directories].

**Jumping Frenchman of Maine:** This rare and obscure apparently autosomal recessive anomaly is characterized by very rapid emotional reactions.

**Jumping Genes:** These genes move in the genome because they are within transposons. ▶ transposable elements

**Jumping Library:** This is generated by circularizing large eukaryotic DNA fragments and cloning the junctions of the circle. The large fragments are obtained by using restriction enzymes that very rarely cut the DNA. ▶ chromosome jumping, ▶ linking library, ▶ slalom library, ▶ DNA library; Zabarovsky ER et al 1991 Genomics 11:1030.

**Jumping Translocations:** These involve one (donor) chromosome and multiple recipient chromosomes. Such unstable phenomena are common in cancers, mainly involving human chromosome 1. The break points are generally in regions of repetitions such as centromeric, telomeric and rRNA sequences. (See Levy B et al 2000 Cytogenet Cell Genet 88:25; Padilla-Nash HM et al 2001 Genes Chromosomes Cancer 30:349).

**JUN** (jun): The avian fibrosarcoma oncogene homolog JUN-A is in human chromosome 1p32-p31 and in mouse chromosome 4. Its homologs are present in other vertebrate species too and may be identical to a subunit of transcription factor AP-1. Along with the product of oncogene FOS, they activate several genes. The products of JUN and FOS are bound together with a leucine zipper and at their carboxyl
end they have a DNA-binding domain (5′-TGAGTC-CA-3′). They apparently form the C/EBP protein. JUN-B and JUN-D oncogenes are closely linked in mouse chromosome 8. JUN-B human homolog is in human chromosome 19p13.2. UV-irradiated mammalian cells may exit from the p-53 imposed block of the cell cycle by the induction of JUN. ►AP1, ►C/EBP, ►oncogenes, ►FOS oncogene, ►JNK, ►bZIP, ►signal transduction, ►de-etiolation, ►UV, ►psoriasis; Barr RK, Bogoyewitch MA 2001 Int J Biochem Cell Biol 33:1047.

Junction Complex: Refers to the assembly of various types of junctions (tight junctions, adhesion belt, desmosome) within cells. ►gap junctions, ►desmosome

Junction of Cellular Networks: These are integrators of molecular signals coming from different sources, and regulated by the interconnections. cAMP may represent such a junction because it is affected in a positive or negative manner by a variety of signals. Phosphokinase A as a node may then split the signals and directs them to multiple targets such as the cytoskeleton and cellular traffic, gene expression and cell growth, metabolism, ion channels, G protein-coupled receptors of signal transduction, and neuronal synapse. Another example of a node is Cdc42, which receives signals through receptor tyrosine kinases (RTK) and G protein-linked receptors (GPCR) and then sorts them into serum response factor (SRF) and p21 activated kinase (PAK), S6 kinase affecting transcription, translation and cellular traffic. ►signal transduction, ►Cdc42, ►RTK, ►G proteins, ►PAK, ►S6 kinase, ►coordinate regulation; Jordan JD et al 2000 Cell 103:193; McCarty DR, Chory J 2000 Cell 103:201; Vohradsky J 2001 FASEB J 15:864.

Junction Sequence: ►introns

Junctional Diversification: When immunoglobulin genes are recombined to generate specific antibodies a few nucleotides may be lost or added to the recombining ends. ►immunoglobulins, ►antibody, ►RAG, ►combinatorial diversification; Wang C et al 1997 J Immunol 159:757.


Jungles: These are chromosomal regions with a high frequency of recombination (genes). ►deserts, ►recombination by replication, ►gene space

Juniper (Juniperus communis): This is an evergreen woody species, 2n = 22.

Junk DNA: This term was coined in the 1970s to describe DNA that appeared without any obvious function such as some introns and spacers. It is now clear that several introns have maturate and other functions. Some of the non-coding DNA is interspecifically conserved, indicating some type of biological function. In animal chromosomes nearly 97% of the DNA is non-coding and this ‘junk’ DNA is predominantly intron material. Nowadays the term non-coding DNA is preferred. Contrary to earlier views, most of the bases in the DNA sequences are transcribed although all their functions are not yet known ►ENCODE, ►selfish DNA, ►non-coding DNA, ►trinucleotide repeats, ►SINE, ►LINE, ►C value paradox, ►TUF, ►non-coding RNA, ►antisense DNA, ►antisense RNA; Wong G-K-S et al 2000 Genome Res 10:1672.

Jurassic Period: Refers to a period nearly 190,000,000 to 137,000,000 years ago. During this period dinosaurs and reptiles were dominant although the ancestral forms of most vertebrates were also present and even primitive mammals had appeared.

Jurkat Cell Lines: These are derived from human T-cell leukemia and are used to study susceptibility to anti-cancer drugs and radiation.

Juvenile Hormone: ►juvenile hormone

Juvenile Hormone: This is secreted in the larval state and prevents precocious metamorphosis into the pupal stage of the insect (see Fig. J5). The hormone has ethyl-polyprenyl components. Similar terpenes and terpene-related substances, e.g., juvebione (in balsam fir) and gossypol (in cotton) occur in plants and also affect the feeding insects. Synthetic hormones have been produced with similar physiological effects. ►metamorphosis, ►molting, ►ecdysone, ►pupa, ►abscisic acid, ►allostatin; Davey K-G 2000 Insect Biochem Mol Biol 30(8–9):663; Gilbert L et al 2000 Insect Biochem Mol Biol 30(8–9):614.

Figure J5. Insect juvenile hormone

Juvenile Mortality: Often this is a function of the consanguinity of the parents. One study, for example, noted that stillbirth and neonatal death was 0.044 if the parents were unrelated and 0.111 if the parents were first cousins (consanguinity 1/16), similarly
infant and juvenile death rates were 0.089 and 0.156, respectively. ▶coancestry, ▶inbreeding, ▶mortality

**Juvenile Onset:** A hereditary condition appearing in childhood. ▶diabetes mellitus

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**Historical vignettes**

Peter Brian Medawar (cited by Colucci F et al 2002 Nature Immunol 3:807)

“The intensity of the conviction that a hypothesis is true has no bearing on whether it is true or not.”

Carl Wilhelm von Nägeli made an effort to convince Mendel about the insignificance of his experiments with peas because he felt it inconceivable that segregation in plants should obey statistical rules. Similarly, he was the founder and unbending adherent of the theory of pleomorphism of bacteria. According to this idea, bacteria did not have a stable heredity, but would change from one form to another by a change in the environment. Although Mendel's theory was not understood by his contemporaries, pleomorphism was subjected to serious criticism; yet von Nägeli's obvious influence definitely hampered bacteriology. Dr. W Migula, Professor at the College of Technology in Karlsruhe, gives a vivid account of the situation in his *System der Bakterien* (Fischer Vlg., Jena, 1897, p. 215):

“When Nägeli says, p. 20, that ‘Cohn [the founder of modern bacterial systematics in 1872] had established a system of genera and species, in which each function of the Schizomycetes [bacteria] is represented by a particular species; by this he expressed the rather widespread view exclusive to physicians. So far I have not come across any factual ground that could be supported by morphological variations or by pertinent definitive experiments.’ When Nägeli still says this in 1877, one must either assume that he was unaware of the work of the preceding 5 years, or that he chose to ignore it on purpose because it did not fit to his theory.”

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**Juxtacrine Signaling:** The membrane-anchored growth factors and cell adhesion molecules are signaled through juxtacrine mediators. ▶signal transduction

**JX₂ DNA:** ▶PX DNA