A Dictionary of GENETICS

Seventh Edition

ROBERT C. KING
Emeritus Professor, Northwestern University

WILLIAM D. STANSFIELD
Emeritus Professor, California Polytechnic State University

PAMELA K. MULLIGAN

OXFORD
UNIVERSITY PRESS

2006

OXFORD
UNIVERSITY PRESS

Oxford University Press, Inc., publishes works that further
Oxford University's objective of excellence
in research, scholarship, and education.

Oxford New York
Auckland Cape Town Dar es Salaam Hong Kong Karachi
Kuala Lumpur Madrid Melbourne Mexico City Nairobi
New Delhi Shanghai Taipei Toronto

With offices in
Argentina Austria Brazil Chile Czech Republic France Greece
Guatemala Hungary Italy Japan Poland Portugal Singapore
South Korea Switzerland Thailand Turkey Ukraine Vietnam

Published by Oxford University Press, Inc.
198 Madison Avenue, New York, New York 10016

www.oup.com

Oxford is a registered trademark of Oxford University Press

Library of Congress Cataloging-in-Publication Data
King, Robert C., 1928–
A dictionary of genetics / by Robert C. King, William D. Stansfield, Pamela K. Mulligan.—
7th ed.
p. cm.
Includes bibliographical references (p.).
ISBN-13 978-0-19-530762-7; 978-0-19-530761-0 (pbk)
ISBN 0-19-530762-3; 0-19-530761-5 (pbk)
1. Genetics—Dictionaries. I. Stansfield, William D., 1930– . II. Mulligan, Pamela
Khipple, 1953– III. Title.
QH427.K55 2006
576.503—dc22 2005045610

9 8 7 6 5 4 3 2 1

Printed in the United States of America
on acid-free paper

Preface

The field of genetics continues to advance at an astounding pace, marked by numerous extraordinary achievements in recent years. In just the past ten years, the genomic sequence of a multitude of organisms, from archaebacteria to large eukaryotes, has been determined and in many cases, comparatively analyzed in remarkable detail. Expressed sequence tags are being used for the detection of new genes and for genome annotation. DNA microarray technology has taken the study of gene expression and genetic variation to a global, genome-wide scale. Hundreds of new genes and microbial species have been identified by reconstructing the DNA sequences of entire communities of microorganisms collected in environmental samples. A wide variety of new regulatory functions have been assigned to RNA, and RNA interference has become an effective tool for creating loss of function phenotypes.

Such momentous advances in genetics have been accompanied by a deluge of new experimental techniques, computational technologies, databases and internet sites, periodicals and books, and, of course, concepts and terms. Furthermore, as new terminology emerges, many old terms inevitably recede from use or require revision. All this is reflected in the changing content of *A Dictionary of Genetics*, from the publication of its first edition to this seventh edition, 37 years later. This new edition has undergone an extensive overhaul, involving one or more changes (additions, deletions, or modifications of entries) on 95% of the pages of the previous one. The seventh edition contains nearly 7,000 definitions, of which 20% are revised or new, and nearly 1,100 Chronology entries, of which 30% are revised or new. Three hundred of the definitions are accompanied by illustrations or tables, and 16 of these are new. In addition, dozens of recent research papers, books, periodicals, and internet sites of genetic importance have been added to the appropriate Appendices of the current edition.

The year 2006 marks the 100th anniversary of the introduction of the term *genetics* by the British biologist William Bateson. In this seventh edition of *A Dictionary of Genetics*, the term *genetics* itself has been updated, reflecting progress in understanding and technique over the years, and necessitated by the convergence of classical and molecular genetics. Genetics today is no longer simply the study of heredity in the old sense, i.e., the study of inheritance and of variation of biological traits, but also the study

of the basic units of heredity, i.e., genes. Geneticists of the post-genomics era identify genetic elements using forward or reverse genetics and decipher the molecular nature of genes, how they function, and how genetic variation, whether introduced in the lab or present in natural populations, affects the phenotype of the cell or the organism. The study of genes is increasingly at the core of genetic research, whether it is aimed at understanding the basis of Alzheimer disease in humans, flower development in *Arabidopsis*, shell pattern variation in *Cepaea* colonies, or speciation in *Drosophila*. Today's genetics thus also unifies the biological sciences, medical sciences, and evolutionary studies.

As a broad-based reference work, *A Dictionary of Genetics* defines terms that fall under this expansive genetics umbrella and includes not only strictly genetic terms, but also genetics-related words encountered in the scientific literature. These include terms referring to biological and synthetic molecules (e.g., *DNA polymerase*, *Morpholinos*, and *streptavidin*); cellular structures (e.g., *solenoid structure*, *spectrosome*, and *sponge body*); medical conditions (e.g., *Leber hereditary optic neuropathy [LHON]*, *Marfan syndrome*, and *Tay-Sachs disease*); experimental techniques (e.g., *P element transformation*, *community genome sequencing*, and *yeast two-hybrid system*); drugs, reagents, and media (e.g., *ethyl methane sulfonate*, *Denhardt solution*, and *HAT medium*); rules, hypotheses, and laws (e.g., *Haldane rule*, *wobble hypothesis*, and *Hardy-Weinberg law*); and acronyms (e.g., *BACs*, *METRO*, and *STS*). Included also are pertinent terms from such fields as geology, physics, and statistics (e.g., *hot spot archipelago*, *roentgen*, and *chi-square test*).

As in previous editions, the definitions are cross-referenced and comparisons made whenever possible. For example, the *maternal effect gene* entry is cross-referenced to *bicoid*, *cytoplasmic determinants*, *cytoplasmic localization*, *grandchildless genes*, and *maternal polarity mutants*, and the reader is directed to compare it with *paternal effect gene* and *zygotic gene* entries.

In this edition of the *Dictionary* we have made every effort to identify the sources of the more than 120 eponyms appearing among the definitions, and following the example of Victor A. McKusick (distinguished editor of *Mendelian Inheritance in Man*), we have eliminated the possessive form, i.e., apostrophes, in most of the eponyms. Thus, the *Creutzfeld-Jakob disease* entry traces the names of the physicians who first described this syndrome in their patients and the time period when this occurred, and the *Balbiani body* definition identifies the biologist who first described these cellular structures and the time period during which he lived. This additional information under each eponym adds a personal, geographical, and historical perspective to the definitions and is one of the distinguishing features of this dictionary.

The Appendices

A Dictionary of Genetics is unique in that only 80% of the pages contain definitions. The final fifth of the *Dictionary* is devoted to six Appendices, which supply a wealth of useful resource material.

Appendix A, Classification, provides an evolutionary classification of the five kingdoms of living organisms. This list contains 400 words in parentheses, many of which are common names for easy identification (e.g., cellular slime molds, marine worms, and ginkgos). The italicized words in parentheses are genera which contain species notable for their economic importance (e.g., *Bos taurus*, *Gossypium hirsutum*, and *Oryza sativa*), for causing human diseases (e.g., *Plasmodium falciparum*, *Staphylococcus aureus*, and *Trypanosoma brucei*), or for being useful laboratory species (e.g., *Arabidopsis thaliana*, *Neurospora crassa*, and *Xenopus laevis*).

Appendix B, Domesticated Species, lists the common and scientific names of approximately 200 domesticated animal and plant species not found elsewhere in the *Dictionary*.

Appendix C, Chronology, is one of the most distinctive elements of the *Dictionary*, containing a list of notable discoveries, events, and publications, which have contributed to the advancement of genetics. The majority of entries in the Chronology report discoveries (e.g., 1865–66, Mendel's discovery of the existence of hereditary factors; 1970, the finding of RNA-dependent DNA polymerase; 1989, the identification of the cystic fibrosis gene). In addition, there are entries that present unifying concepts and theories (e.g., 1912, the concept of continental drift; 1961, the operon hypothesis; 1974, the proposition that chromatin is organized into nucleosomes). The Chronology also includes important technological advances and techniques that have revolutionized genetic research (e.g., 1923, the building of the first ultracentrifuge; 1975, the development of Southern blotting; 1985, the development of polymerase chain reaction; 1986, the production of the first automated DNA sequencer). There are also entries that contain announcements of new terms that have become part of every geneticist's vocabulary (e.g., 1909, gene; 1971, C value paradox; 1978, intron and exon).

Developments in evolutionary genetics figure prominently in the Chronology. Included in this category are important evolutionary breakthroughs (e.g., 1868, Huxley's description of *Archaeopteryx*; 1977, the discovery of the Archaea by Woese and Fox; 2004, the proposal by Rice and colleagues that viruses evolved from a common ancestor prior to the formation of the three domains of life), and publication of books which have profoundly affected evolutionary thought (e.g., 1859, C. Darwin's *On the Origin of Species*; 1963, E. Mayr's *Animal Species and Evolution*; 1981, L. Margulis's *Symbiosis in Cell Evolution*).

Relatively recent additions to the Chronology are entries for sequencing and analysis of the genomes of species of interest (e.g., 1996, *Saccharomyces cerevisiae*; 1997, *Escherichia coli*; 2002, *Mus musculus*). Finally, the Chronology lists 59 Nobel Prizes awarded to scientists for discoveries that have had a bearing on the progress of genetics (e.g., 1965, to F. Jacob, J. Monod, and A. Lwoff for their contributions to microbial genetics; 1983, to B. McClintock for her discovery of mobile genetic elements in maize; 1993, to R. J. Roberts and P. A. Sharp for discovering split genes). We hope that these and other Chronology entries, spanning the years 1590–2005, provide students, researchers, educators, and historians alike with an understanding of the historical framework within which genetics has developed.

The Chronology in Appendix C is followed by an alphabetical List of the Scientists cited in it, together with the dates of these citations. This list includes Francis Crick, Edward Lewis, Maurice Wilkins, and Hampton Carson (who all died late in 2004), and Ernst Mayr (who died early in 2005), and it provides the dates of milestones in their scientific careers. Finally, Appendix C includes a Bibliography of 170 titles, and among the most recent books are four that give accounts of the lives of David Baltimore, George Beadle, Sidney Brenner, and Rosalind Franklin. Also listed is a video collection (*Conversations in Genetics*) of interviews with prominent geneticists.

Appendix D, Periodicals, lists the titles and addresses of 500 periodicals related to genetics, cell biology, and evolutionary studies, from *Acta Virologica* to *Zygote*.

Appendix E, Internet Sites, contains 132 prominent web site addresses to facilitate retrieval of the wealth of information in the public domain that can be accessed through the World Wide Web. These include addresses for "master" sites (e.g., National Center for Biotechnology Information [NCBI], National Library of Medicine, National Institutes of Health), for individual databases (e.g., GenBank, Single Nucleotide Polymorphisms [SNPs], and Protein Data Bank [PDB]), and for species web sites (e.g., *Agrobacterium tumefaciens*, *Chlamydomonas reinhardii*, and *Gossypium* species).

Appendix F, Genome Sizes and Gene Numbers, tabulates the genome sizes and gene numbers for 49 representative organisms, viruses, or cell organelles that appear in the *Dictionary*. These are listed in order of complexity. The smallest genome listed is that of the MS2 virus, with 3.6×10^3 base pairs encoding just 4 proteins, and the largest listed is that of man, consisting of 3.2×10^9 base pairs of DNA encoding 31,000 genes. Between these entries appear the genome sizes and gene numbers of other viruses, organelles, and a diverse range of organisms representing all five kingdoms.

This is but a small representation of the larger and increasingly complex collections of genomic data which are being generated at an exponential

rate and transforming the way we look at relationships between organisms that inhabit this planet. A quick glance at Appendix F raises some intriguing questions. For example, why does *Streptomyces*, a prokaryote, have more genes than *Saccharomyces*, a eukaryote, whose genome size is 28% larger? And why do the genomes of the puffer fish, *Takifugu rubripes*, and man encode roughly the same number of protein-coding genes, even though the puffer fish genome is nearly 88% smaller than the human? Such questions and others are at the forefront of current whole-genome research, as the massive sequence data are evaluated and the information encoded within them extracted. Comparative genomic analyses promise new insights into the evolutionary forces that shape the size and structure of genomes. Furthermore, the intertwining of genetics, genomics, and bioinformatics makes for a strong force for identifying new genetic elements and for unraveling the mysteries of cellular processes in the most minute detail.

Appendix Cross-References. Whenever possible, cross references to the Appendices appear under the appropriate definition. The cross references provide information which complements that in the definition. For example, *nucleolus* is cross-referenced to entries in Appendix C, which indicate that this structure was first observed in the nucleus in 1838, that it was first shown to be divisible into subunits in 1934, that in 1965 the sex chromosomes of *Drosophila melanogaster* were found to contain multiple rRNA genes in their nucleolus organizers, and that in 1967 amplified rDNA was isolated from *Xenopus* oocytes. Furthermore, nucleolar Miller trees were discovered in 1969, in 1976 ribosomal proteins were found to attach to precursor rRNAs in the nucleolus, and in 1989 the cDNA for human nucleolin was isolated. Another example is *Streptomyces*, which is cross-referenced to Appendices A, E, and F. In this case, the material in the Appendices indicates that this organism is a prokaryote belonging to the phylum Actinobacteria, that there is web-based information pertaining to *S. coelicolor* at http://www.sanger.ac.uk, and that the genome of this species has 12.07×10^6 base pairs and contains 7,825 predicted genes. The cross-referenced information in the Appendices thus greatly broadens the reader's perspective on a particular term or concept.

Genetics has clearly entered an exciting new era of exploration and expansion. It is our sincere hope that *A Dictionary of Genetics* will become a helpful companion for those participating in this marvelous adventure.

Rules Regarding the Arrangement of Entries

The arrangement of entries in the current edition has not changed since the publication of the previous edition. Each term appears in boldface and is placed in alphabetical order using the letter-by-letter method, ignoring

spaces between words. Thus, *Homo sapiens* is placed between *homopolymer tails* and *homosequential species*, and *H-Y antigen* appears between *hyaluronidase* and *hybrid*. In the case of identical alphabetical listings, lowercase letters precede uppercase letters. Thus, the *p* entry is found before the *P* entry. In entries beginning with a Greek letter, the letter is spelled out. Therefore, β *galactosidase* appears as *beta galactosidase*. When a number is found at the beginning of an entry, the number is ignored in the alphabetical placement. Therefore, *M5 technique* is treated as *M technique* and *T24 oncogene* as *T oncogene*. However, numbers are used to determine the order in the series. For example, *P1 phage* appears before *P22 phage*. For two- or three-word terms, the definition sometimes appears under the second or third word, rather than the first. For example, definitions for *embryonic stem cells* and *germ line transformation* occur under *stem cells* and *transformation*, respectively.

Acknowledgments

We owe the greatest debt to Ellen Rasch, whose critical advice at various stages during the evolution of the dictionary provided us with wisdom and encouragement. We also benefited by following wide-ranging suggestions made by Lloyd Davidson, Joseph Gall, Natalia Shiltsev, Igor Zhimulev, and the late Hampton Carson. Rodney Adam, Bruce Baldwin, Frank Butterworth, Susanne Gollin, Jon Moulton, and Patrick Storto suggested changes that improved the quality of many definitions. Atsuo Nakata kindly brought to our attention many typographical errors that we had missed.

We are grateful to the many scientists, illustrators, and publishers who kindly provided their illustrations to accompany various entries. Robert S. King, who took over secretarial functions from his mother, Suja, and elder brother Tom, worked cheerfully and tirelessly throughout the project. Vikram K. Mulligan suggested various terms and modified others, and Rob and Vikram's drawings illustrate eight of the entries.

Robert C. King
William D. Stansfield
Pamela K. Mulligan

Contents

A Dictionary of Genetics

A

A **1.** mass number of an atom; **2.** haploid set of autosomes; **3.** ampere; **4.** adenine or adenosine.

Å Angstrom unit (*q.v.*).

A_2 *See* hemoglobin.

A 23187 *See* ionophore.

AA-AMP amino acid adenylate.

A, B antigens mucopolysaccharides responsible for the ABO blood group system. The A and B antigens reside on the surface of erythrocytes, and differ only in the sugar attached to the penultimate monosaccharide unit of the carbohydrate chain. This minor chemical difference makes the macromolecule differentially active antigenically. The I^A, I^B, and *i* are alleles of a gene residing on the long arm of chromosome 9 between bands 34.1 and 34.2. The I^A and I^B alleles encode A and B glycotransferases, and the difference in their specificities is due to differences in their amino acid sequences at only four positions. These in turn result from different missense mutations in the two alleles. The A and B transferases add *N*-acetyl galactosamine or galactose, respectively, to the oligosaccharide terminus. The *i* allele encodes a defective enzyme, so no additional monosaccharide is added to the chain. Glycoproteins with properties antigenically identical to the A, B antigens are ubiquitous, having been isolated from bacteria and plants. Every human being more than 6 months old possesses those antibodies of the A, B system that are not directed against its own blood-group antigens. These "preexisting natural" antibodies probably result from immunization by the ubiquitous antigens mentioned above. The A and B antigens also occur on the surfaces of epithelial cells, and here they may mask receptors that serve as binding sites for certain pathogenic bacteria. *See* Appendix C, 1901, Landsteiner; 1925, Bernstein; 1990, Yamomoto *et al.*; blood group, *Helicobacter pylori*, H substance, Lewis blood group, MN blood group, null allele, oligosaccharide, P blood group, *Secretor* gene.

ABC model *See* floral identity mutations.

ABC transporters a family of proteins that span the plasma membranes of cells and function to transport specific molecules into or out of the cell. The name is an abbreviation of *A*TP-*B*inding Cassette. ABC transporters all contain an ATP binding domain, and they utilize the energy of ATP to pump substrates across the membrane against a concentration gradient. The substrates may be amino acids, sugars, polypeptides, or inorganic ions. The product of the cystic fibrosis gene is an ABC transporter. *See* Bacillus, cystic fibrosis (CF), *Escherichia coli*.

Abelson murine leukemia virus an oncogenic virus identified in 1969 by Dr. H. T. Abelson. The transforming gene *v-abl* has a cellular homolog *c-abl*. This is actively transcribed in embryos at all stages and during postnatal development. A homolog of *c-abl* occurs in the human genome at 9q34, and it encodes a protein kinase (*q.v.*). It is this gene which is damaged during the reciprocal interchange that occurs between chromosome 9 at q34 and chromosome 22 at q11, resulting in myeloid leukemia. *See* Philadelphia (Ph^1) chromosome, myeloproliferative disease.

aberrations *See* chromosomal aberration, radiation-induced chromosomal aberration.

ABM paper aminobenzyloxy methyl cellulose paper, which when chemically activated, reacts covalently with single-stranded nucleic acids.

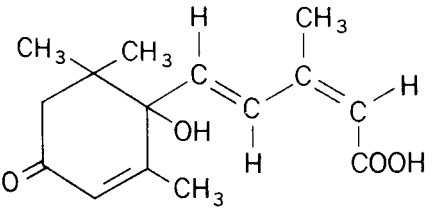
ABO blood group system system of alleles residing on human chromosome 9 that specifies certain red cell antigens. *See* AB antigens, blood groups, Bombay blood group.

abortion **1.** The expulsion of a human fetus from the womb by natural causes, before it is able to survive independently; this is sometimes called a miscarriage (*q.v.*). **2.** The deliberate termination of a human pregnancy, most often performed during the first 28 weeks of pregnancy. **3.** The termination of development of an organ, such as a seed or fruit.

abortive transduction failure of a transducing exogenote to become integrated into the host chromosome, but rather existing as a nonreplicating particle in only one cell of a clone. *See* transduction.

abortus a dead fetus born prematurely, whether the abortion was artificially induced or spontaneous. Over 20% of human spontaneous abortions show chromosomal abnormalities. *See* Appendix C, 1965, Carr.

abscisic acid a plant hormone synthesized by chloroplasts. High levels of abscisic acid result in the abscission of leaves, flowers, and fruits. The hormone also causes the closing of stomata in response to dehydration.



abscission the process whereby a plant sheds one of its parts, such as leaves, flowers, seeds, or fruits.

absolute plating efficiency the percentage of individual cells that give rise to colonies when inoculated into culture vessels. *See* relative plating efficiency.

absorbance (*also* absorbancy) a measure of the loss of intensity of radiation passing through an absorbing medium. It is defined in spectrophotometry by the relation log (I_o/I), where I_o = the intensity of the radiation entering the medium and I = the intensity after traversing the medium. *See* Beer-Lambert law, OD_{260} unit.

abundance in molecular biology, the average number of molecules of a specific mRNA in a given cell, also termed *representation*. The abundance, $A = NRf/M$, where N = Avogadro's number, R = the RNA content of the cell in grams, f = the fraction the specific RNA represents of the total RNA, and M = the molecular weight of the specific RNA in daltons.

abzymes catalytic antibodies. A class of monoclonal antibodies that bind to and stabilize molecules in the transition state through which they must pass to form products. *See* enzyme.

acatalasemia the hereditary absence of catalase (*q.v.*) in humans. Mutations in the structural gene on chromosome 11 at p13 result in the production of an unstable form of the enzyme. The gene is 34 kb in length and contains 13 exons.

acatalasia synonym for acatalasemia (*q.v.*).

acceleration *See* heterochrony.

accelerator an apparatus that imparts kinetic energy to charged subatomic particles to produce a high-energy particle stream for analyzing the atomic nucleus.

acceptor stem the double-stranded branch of a tRNA molecule to which an amino acid is attached (at the 3′, CCA terminus) by a specific aminoacyl-tRNA synthetase. *See* transfer RNA.

accessory chromosomes *See* B chromosomes.

accessory nuclei bodies resembling small nuclei that occur in the oocytes of most Hymenoptera and those of some Hemiptera, Coleoptera, Lepidoptera, and Diptera. Accessory nuclei are covered by a double membrane possessing annulate pores. They are originally derived from the oocyte nucleus, but they subsequently form by the amitotic division of other accessory nuclei.

***Ac, Ds* system** *Activator–Dissociation* system (*q.v.*).

ace *See* symbols used in human cytogenetics.

acentric designating a chromatid or a chromosome that lacks a centromere. *See* chromosome bridge.

Acer the genus of maple trees. *A. rubrum*, the red maple, and *A. saccharum*, the sugar maple, are studied genetically because of their commercial importance.

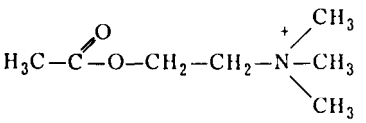
Acetabularia a genus of large, unicellular green algae. Each organism consists of a base, a stalk, and a cap. The base, which contains the nucleus, anchors the alga to the supporting rocks. The stalk, which may be 5 cm long, joins the base and the cap. The cap carries out photosynthesis and has a species-specific shape. For example, the disc-shaped cap of *A. mediterranea* is smooth, whereas the cap of *A. crenulata* is indented. Hammerling cut the base and cap off a *crenulata* alga and then grafted the stalk on a *mediterranea* base. The cap that regenerated was smooth, characteristic of the species that provided the nucleus. Heterografts like these provided some of the earliest evidence that the nucleus could send messages that directed developmental programs at distant regions of the cell. *See* Appendix A, Protoctista, Chlorophyta; Appendix C, 1943, Hammerling; graft.

Acetobacter a genus of aerobic bacilli which secure energy by oxidizing alcohol to acetic acid.

aceto-orcein a fluid consisting of 1% orcein (*q.v.*) dissolved in 45% acetic acid, used in making squash preparations of chromosomes. *See* salivary gland squash preparation.

acetylcholine a biogenic amine that plays an important role in the transmission of nerve impulses across synapses and from nerve endings to the muscles innervated. Here it changes the permeability of

the sarcolemma and causes contraction. Acetylcholine is evidently a very ancient hormone, since it is present even in protists.



acetylcholinesterase the enzyme that catalyses the hydrolysis of acetylcholine (*q.v.*) into choline and acetate. Also called *cholinesterase*.

acetyl-coenzyme A *See* coenzyme A.

acetyl serine *See N*-acetyl serine.

achaete-scute complex a complex locus in *Drosophila* first identified by mutations that affected the development of adult bristles. Lack of the entire complex results in the failure of neurogenesis during the embryo stage. The complex contains four ORFs that encode DNA-binding proteins that contain helix-turn-helix motifs (*q.v.*).

achiasmate referring to meiosis without chiasmata. In those species in which crossing over is limited to one sex, the achiasmate meiosis generally occurs in the heterogametic sex.

Achilles' heel cleavage (AHC) a technique that allows a DNA molecule to be cut at a specified site. The name comes from the legend in Greek mythology where Achilles' mother dipped him in the river Styx. The waters made him invulnerable, except for the heel by which she held him. In the AHC procedure a sequence-specific DNA-binding molecule is complexed with the DNA under study. A methyltransferase is then added to methylate all CpG sequences except those hidden under the sequence-specific DNA-binding molecule. Next, this molecule and the methyltransferases are removed, and a restriction endonuclease is added. This will cut the DNA only in the region where methylation was blocked, i.e., the "Achilles' heel."

achondroplasia a form of hereditary dwarfism due to retarded growth of the long bones. It is the most common form of dwarfism in humans (1 in 15,000 live births) and is inherited as an autosomal dominant trait. Homozygotes die at an early age. The gene responsible has been mapped to chromosome 4p16.3. The *ACH* gene has been renamed *FGFR3*, since it encodes the *F*ibroblast *G*rowth *F*actor *R*eceptor *3*, a protein containing 806 amino acids. The gene contains 14,975 bp of DNA and produces two alternative transcripts. Homologous genes have been identified in rat, mouse, *Xenopus*, and zebrafish. The genes are expressed in the chondrocytes of developing bones. *See* bovine achondroplasia, *de novo* mutation, fowl achrondroplasia, positional candidate approach.

achromatic figure the mitotic apparatus (*q.v.*).

A chromosomes *See* B chromosomes.

acid fuchsin an acidic dye used in cytochemistry.

acidic amino acid an amino acid (*q.v.*) having a net negative charge at neutral pH. Those universally found in proteins are aspartic acid and glutamic acid, which bear negatively charged side chains in the pH range generally found in living systems.

acidic dye an organic anion that binds to and stains positively charged macromolecules.

Acinonyx jubatus the cheetah, a carnivore that has the distinction of being the world's fastest land animal. Cheetahs are of genetic interest because, while most other species of cats show heterozygosity levels of 10–20%, cheetahs have levels close to zero. This high degree of homozygosity is correlated with low fecundity, high mortality of cubs, and low disease resistance.

Acoelomata a subdivision of the Protostomia-containing species in which the space between the epidermis and the digestive tube is occupied by a cellular parenchyma. *See* classification.

acquired characteristics, inheritance of inheritance by offspring of characteristics that arose in their parents as responses to environmental influences and are not the result of gene action. *See* Lamarckism.

acquired immunodeficiency syndrome *See* AIDS, HIV.

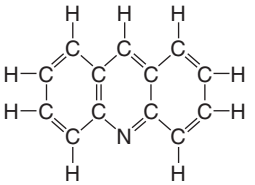
Acraniata a subphylum of Chordata containing animals without a true skull. *See* Appendix A.

acrasin a chemotactic agent produced by *Dictyostelium discoideum* that is responsible for the aggregation of the cells. Acrasin has been shown to be cyclic AMP (*q.v.*).

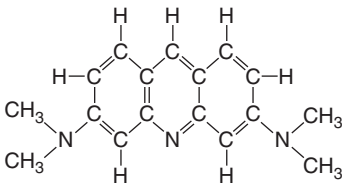
Acrasiomycota the phylum containing the cellular slime molds. These are protoctists that pass through a unicellular stage of amoebas that feed on bacteria. Subsequently, these amoebas aggregate to form a fruiting structure that produces spores. The two most extensively studied species from this phylum

are *Dictyostelium discoideum* and *Polysphondylium pallidum*.

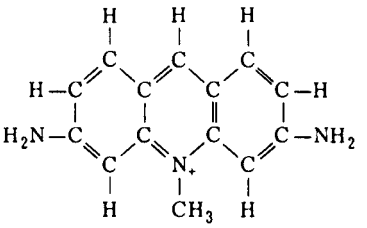
acridine dyes heterocyclic compounds that include acridine (shown below) and its derivatives. These molecules bind to double-stranded DNAs as intercalating agents. Examples of acridine dyes are acridine organe, acriflavin, proflavin, and quinicrine (*all of which see*).



acridine orange an acridine dye that functions both as a fluorochrome and a mutagen.



acriflavin an acridine dye that produces reading frame shifts (*q.v.*).



acritarchs spherical bodies thought to represent the earliest eukaryotic cells, estimated to begin in the fossil record about 1.6 billion years ago. Most acritarchs were probably thick-walled, cyst-forming protists. *See* Proterozoic.

acrocentric designating a chromosome or chromatid with a nearly terminal centromere. *See* telocentric chromosome.

acromycin *See* tetracycline.

acron the anterior nonsegmented portion of the embryonic arthropod that produces eyes and antennae. *See* maternal polarity mutants.

acrosome an apical organelle in the sperm head that is secreted by the Golgi material and that digests the egg coatings to permit fertilization.

acrostical hairs one or more rows of small bristles along the dorsal surface of the thorax of *Drosophila*.

acrosyndesis telomeric pairing by homologs during meiosis.

acrotrophic *See* meroistic.

acrylamide *See* polyacrylamide gel.

ACTH adrenocorticotropic hormone (*q.v.*).

actidione cycloheximide.

actin a protein that is the major constituent of the 7-nanometer-wide microfilaments of cells. Actin microfilaments (F actin) are polymers of a globular subunit (G actin) of Mr 42,000. Each G actin molecule has a defined polarity, and during polymerization the subunits align "head to tail," so that all G actins point in the same direction. F actin grows by the addition of G actin to its ends, and cytochalasin B (*q.v.*) inhibits this process. All the actins that have been studied, from sources as diverse as slime molds, fruit flies, and vertebrate muscle cells, are similar in size and amino acid sequence, suggesting that they evolved from a single ancestral gene. In mammals and birds, there are four different muscle actins. α_1 is unique to skeletal muscle; α_2, to cardiac muscle; α_3, to smooth vascular muscle; and α_4, to smooth enteric muscle. Two other actins (β and γ) are found in the cytoplasm of both muscle and nonmuscle cells. *See* alternative splicing, contractile ring, fibronectin, *hu-li tai shuo (hts)*, isoform, *kelch*, myosin, ring canals, spectrin, stress fibers, tropomyosin, vinculin.

actin-binding proteins a large family of proteins that form complexes with actin. Such proteins include certain heat-shock proteins, dystrophin, myosin, spectrin, and tropomyosin (*all of which see*).

actin genes genes encoding the various isoforms of actin. In *Drosophila*, for example, actin genes have been localized at six different chromosomal sites. Two genes encode cytoplasmic actins, while the other four encode muscle actins. The amino acid–encoding segments of the different actin genes have very similar compositions, but the segments specifying the trailers (*q.v.*) differ considerably in nucleotide sequences.

actinomycete any prokaryote placed in the phylum actinobacteria (*see* Appendix A). Actinomycetes belonging to the genus *Streptomyces* produce a large number of the antibiotics, of which actinomycin D (*q.v.*) is an example.

actinomycin D an antibiotic produced by *Streptomyces chrysomallus* that prevents the transcription of messenger RNA. *See* RNA polymerase.

activated macrophage a macrophage that has been stimulated (usually by a lymphokine) to enlarge, to increase its enzymatic content, and to increase its nonspecific phagocytic activity.

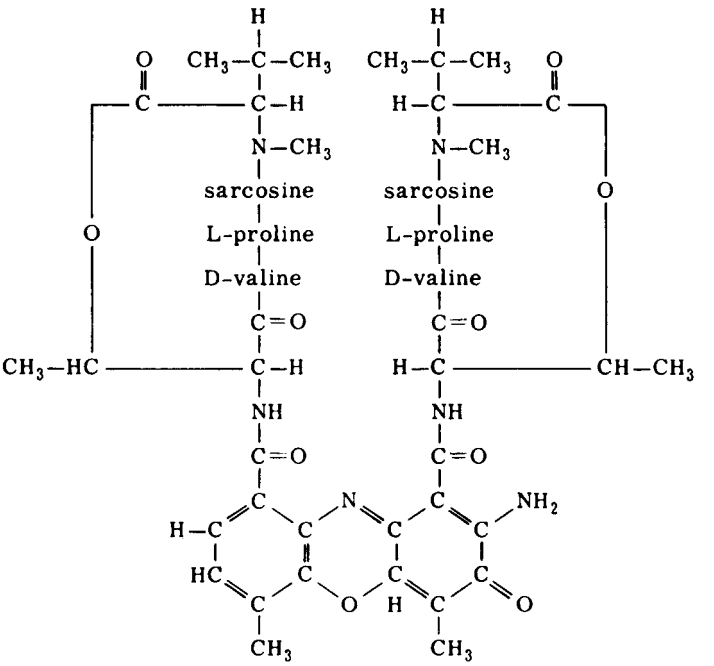
activating enzyme an enzyme that catalyzes a reaction involving ATP and a specific amino acid. The product is an activated complex that subsequently reacts with a specific transfer RNA.

activation analysis a method of extremely sensitive analysis based on the detection of characteristic radionuclides produced by neutron activation.

activation energy the energy required for a chemical reaction to proceed. Enzymes (*q.v.*) combine transiently with a reactant to produce a new complex that has a lower activation energy. Under these circumstances the reaction can take place at the prevailing temperature of the biological system. Once the product is formed, the enzyme is released unchanged.

activator a molecule that converts a repressor into a stimulator of operon transcription; e.g., the repressor of a bacterial arabinose operon becomes an activator when combined with the substrate.

***Activator-Dissociation* system** a pair of interacting genetic elements in maize discovered and analyzed by Barbara McClintock. *Ac* is an *autonomous* element that is inherently unstable. It has the ability to excise itself from one chromosomal site and to transpose to another. *Ac* is detected by its activation of *Ds*. *Ds* is *nonautonomous* and is not capable of excision or transposition by itself. *Ac* need not be adjacent to *Ds* or even on the same chromosome in order to activate *Ds*. When *Ds* is so activated, it can alter the level of expression of neighboring genes, the structure of the gene product, or the time of development when the gene expresses itself, as a consequence of nucleotide changes inside or outside of a given cistron. An activated *Ds* can also cause chromosome breakage, which may yield deletions or generate a breakage-fusion-bridge cycle (*q.v.*). It is now known that *Ac* is a 4,500 bp segment of DNA that encodes a transposable element (*q.v.*) which contains within it the locus of a functional transposase (*q.v.*). The transposase gives *Ac* the ability to detach



Actinomycin D

from one chromosome and then insert into another. The excision of *Ac* may cause a break in the chromosome, and this is what generated the breakage-fusion-bridge cycles that McClintock observed. *Ds* is a defective transpon that contains a deletion in its transposase locus. Therefore the *Ds* transposon can move from chromosome to chromosome only if *Ac* is also in the nucleus to supply its transposase. *Ac* and *Ds* were originally classified as mutator genes, since they would sometimes insert into structural genes and modify their functioning. *See* Appendix C, 1950, McClintock; 1984, Pohlman *et al.*; *Dotted*, genomic instability, mutator gene, terminal inverted repeats (TIRs), transposon tagging.

active center in the case of enzymes, a flexible portion of the protein that binds to the substrate and converts it into the reaction product. In the case of carrier and receptor proteins, the active center is the portion of the molecule that interacts with the specific target compounds.

active immunity immunity conferred on an organism by its own exposure and response to antigen. In the case of immunity to disease-causing agents, the antigenic pathogens may be administered in a dead or attenuated form. *See also* passive immunity.

active site that portion(s) of a protein that must be maintained in a specific shape and amino acid content to be functional. Examples: **1.** in an enzyme, the substrate-binding region; **2.** in histones or repressors, the parts that bind to DNA; **3.** in an antibody, the part that binds antigen; **4.** in a hormone, the portion that recognizes the cell receptor.

active transport the movement of an ion or molecule across a cell membrane against a concentration or electrochemical gradient. The process requires specific enzymes and energy supplied by ATP.

activin a protein first isolated from the culture fluid of *Xenopus* cell lines. Activin is a member of the transforming growth factor-β (*q.v.*) family of intercellular signaling molecules. It acts as a diffusible morphogen for mesodermal structures, and the type of differentiation is determined by the concentration of actin (i.e., high concentrations produce head structures, low concentrations tail structures).

actomyosin *See* myosin.

***acute myeloid leukemia 1* gene (*AML1*)** a gene that maps to 21q22.3 and is one of the most frequent targets of chromosome translocations associated with leukemia. The involvement of *AML1* with the oncogenic transformation of blood cells is worth noting, since acute myeloid leukemia is hundreds of times more common in children with trisomy 21 than in other children. *See* Down syndrome, *lozenge*, myeloproliferative disease.

acute transfection infection of cells with DNA for a short period of time.

acylated tRNA a transfer RNA molecule to which an amino acid is covalently attached. Also referred to as an activated tRNA, a charged tRNA, or a loaded tRNA.

adaptation **1.** the process by which organisms undergo modification so as to function more perfectly in a given environment. **2.** any developmental, behavioral, anatomical, or physiological characteristic of an organism that, in its environment, improves its chances for survival and of leaving descendants.

adaptive enzyme an enzyme that is formed by an organism in response to an outside stimulus. The term has been replaced by the term *inducible enzyme*. The discovery of adaptive enzymes led eventually to the elucidation of the mechanisms that switch gene transcription on and off. *See* Appendix C, 1937, Karström; regulator gene.

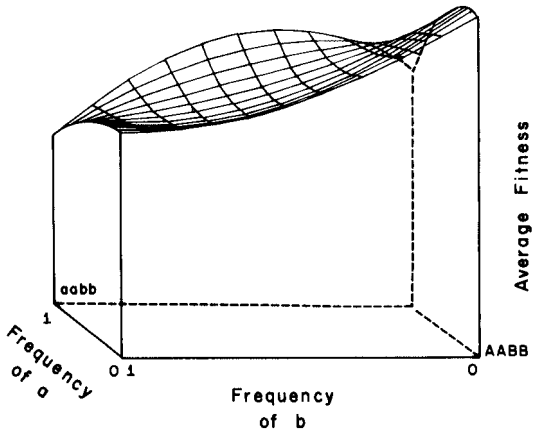
adaptive immunity the immunity that develops in response to an antigens (*q.v.*), as opposed to innate or natural immunity. C*ontrast with* innate immunity.

adaptive landscape a three-dimensional graph that shows the frequencies of two genes, each present in two allelic forms (aA and bB in the illustration) plotted against average fitness for a given set of environmental conditions, or a comparable conceptual plot in multidimensional space to accommodate more than two loci.

adaptive melanism hereditary changes in melanin production that cause the darkening in color of populations of animals in darkened surroundings. By improving their camouflage, this makes them less conspicuous to predators. For example, desert mice are preyed upon by owls, hawks, and foxes. The mice that live among sand and light-colored rocks are tan and blend in well with their surroundings. However, the fur from populations of the same species that live among outcrops of dark, ancient lava flows is much darker. *See Chaetodipus intermedius.*

adaptive norm the array of genotypes (compatible with the demands of the environment) possessed by a given population of a species.

adaptive peak a high point (perhaps one of several) on an adaptive landscape (*q.v.*), from which movement in any planar direction (changed gene frequencies) results in lower average fitness.



Adaptive landscape

adaptive radiation the evolution of specialized species, each of which shows adaptations to a distinctive mode of life, from a generalized ancestral species. Darwin observed the adaptive radiation of finch species on the Galapagos islands. The Hawaiian archipelago shows perhaps the most spectacular examples of adaptive radiations. *See* Darwin's finches, Hawaiian Drosophilidae, silversword alliance.

adaptive surface, adaptive topography synonyms for adaptive landscape (*q.v.*).

adaptive value the property of a given genotype when compared with other genotypes that confers fitness (*q.v.*) to an organism in a given environment.

adaptor a short, synthetic DNA segment containing a restriction site that is coupled to both ends of a blunt-ended restriction fragment. The adaptor is used to join one molecule with blunt ends to a second molecule with cohesive ends. The restriction site of the adaptor is made identical to that of the other molecule so that when cleaved by the same restriction enzyme both DNAs will contain mutually complementary cohesive ends.

adaptor hypothesis the proposal that polynucleotide adaptor molecules exist that can recognize specific amino acids and also the regions of the RNA templates that specify the placement of amino acids in a newly forming polypeptide. *See* Appendix C, 1958, Crick; transfer RNA.

ADCC antibody-dependent cellular cytotoxicity; also known as antibody-dependent cell-mediated cytotoxicity. Cell-mediated cytotoxicity requires prior binding of antibody to target cells for killing to occur. It does not involve the complement cascade. *See* K cells.

additive factor one of a group of nonallelic genes affecting the same phenotypic characteristics and each enhancing the effect of the other in the phenotype. *See* quantitative inheritance.

additive gene action **1.** a form of allelic interaction in which dominance is absent; the heterozygote is intermediate in phenotype between homozygotes for the alternative alleles. **2.** the cumulative contribution made by all loci (of the kind described above) to a polygenic trait.

additive genetic variance genetic variance attributed to the average effects of substituting one allele for another at a given locus, or at the multiple loci governing a polygenic trait. It is this component of variance that allows prediction of the rate of response for selection of quantitative traits. *See* quantitative inheritance.

adducin a ubiquitously expressed protein found in the membranes of animal cells. Mammalian adducin is a heterodimeric protein whose subunits share sequence similarities and contain protease-resistant N-terminal and protease-sensitive C-terminal domains. Adducin has a high affinity for Ca^{2+}/calmodulin and is a substrate for protein kinases. *In vitro* it causes actin filaments to form bundles and promotes spectrin-actin associations in regions where cells contact one another. In *Drosophila*, a homolog of mammalian adducin is encoded by the *hts* gene. *See* calmodulin, fusome, *hu-li tai shao (hts)*, heterodimer, protein kinase, spectrosome.

adduct the product of a chemical reaction that results in the addition of a small chemical group to a relatively large recipient molecule. Thus the alkylating agent ethyl methane sulfonate (*q.v.*) can add

ethyl groups to the guanine molecules of DNA. These ethylated guanines would be examples of DNA adducts.

adenine *See* bases of nucleic acids.

adenine deoxyriboside *See* nucleoside.

adenohypophysis the anterior, intermediate, and tuberal portions of the hypophysis, which originate from the buccal lining in the embryo.

adenohypophysis hormone *See* growth hormone.

adenosine *See* nucleoside.

adenosine deaminase deficiency a rare immune deficiency disease due to mutations in a gene located on the long arm of human chromosome 20. The normal gene encodes an enzyme that controls the metabolism of purines, and ADA deficiency impairs the functioning of white blood cells. The division of T cells is depressed, and antibody production by B cells is reduced. As a result, ADA-deficient children die from viral, bacterial, and fungal infections. ADA deficiency is the first hereditary disease to be successfully treated by gene therapy. *See* Appendix C, 1990, Anderson; immune response.

adenosine phosphate any of three compounds in which the nucleoside adenosine is attached through its ribose group to one, two, or three phosphoric acid molecules, as illustrated here. AMP, ADP, and ATP are interconvertible. ATP upon hydrolysis yields the energy used to drive a multitude of biological processes (muscle contraction, photosynthesis, bioluminescence, and the biosynthesis of proteins, nucleic acids, polysaccharides and lipids). The most important process in human nutrition is the synthesis of ATP. Every day human beings synthesize, breakdown, and resynthesize an amount of ATP equaling their body weight. *See* Appendix C, 1929; Lohmann; ATPase, ATP synthase, cellular respiration, citric acid cycle, cytochromes, electron transport chain, glycolysis, mitochondria, oxidative phosphorylation, mitochondrial proton transport.

adenovirus any of a group of spherical DNA viruses characterized by a shell containing 252 capsomeres. Adenoviruses infect a number of mammalian species including humans. *See* human adenovirus 2 (HAdV-2), virus.

adenylcyclase the enzyme that catalyzes the conversion of ATP into cyclic AMP (*q.v.*). Also called adenylate cyclase. *See* adenosine phosphate.

adenylic acid *See* nucleotide.

ADH the abbreviation for alcohol dehydrogenase (*q.v.*).

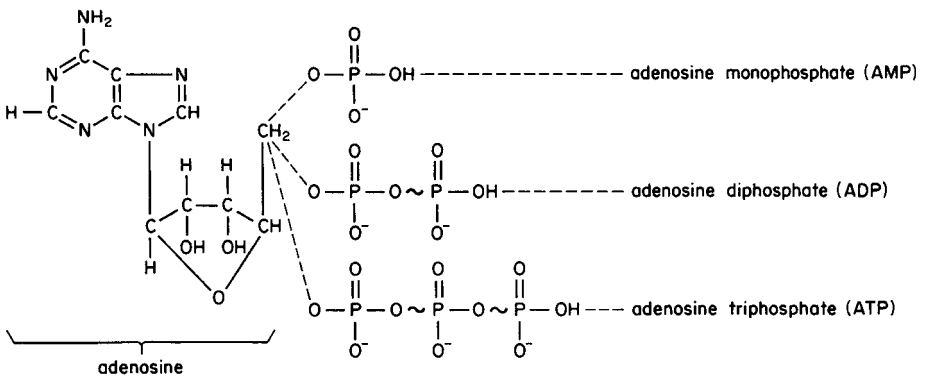
adhesion plaques *See* vincullin.

adhesive molecules any pair of complementary cell-surface molecules that bind specifically to one another, thereby causing cells to adhere to one another, as do carbohydrates and protein lectins (*q.v.*). Phenomena dependent on adhesive molecules include invasion of host cells by bacteria and viruses, species-specific union of sperms and eggs, and aggregation of specific cell types during embryological development. *See* cell affinity, hemagglutinins, P blood group, selectins.

adjacent disjunction, adjacent segregation *See* translocation heterozygote.

adjuvant a mixture injected together with an antigen that serves to intensify unspecifically the immune response. *See* Freund's adjuvant.

adoptive immunity the transfer of an immune function from one organism to another through the transfer of immunologically active or competent cells. Also called *adoptive transfer*.



Adenosine phosphate

ADP adenosine diphosphate. *See* adenosine phosphate.

adrenal corticosteroid a family of steroid hormones formed in the adrenal cortex. There are more than 30 of these hormones, and all are synthesized from cholesterol by cortical cells that have been stimulated by the adrenocorticotropic hormone (*q.v.*).

adrenocorticotropic hormone a single-chain peptide hormone (39 amino acids long) stimulating secretion by the adrenal cortex. It is produced by the adenohypophysis of vertebrates. Abbreviated ACTH. Also called *corticotropin*.

Adriamycin an antibiotic produced by *Streptomyces peucetius* that interacts with topoisomerase. DNA isolated from Adriamycin-poisoned cells contains single- and double-strand breaks. *See* gyrase, mitotic poison.

adult tissue stem cells *See* stem cells

advanced in systematics, the later or derived stages or conditions within a lineage that exhibits an evolutionary advance; the opposite of primitive.

adventitious embryony the production by mitotic divisions of an embryonic sporophyte from the tissues of another sporophyte without a gametophytic generation intervening.

Aedes a genus of mosquitoes containing over 700 species, several of which transmit important human diseases. *A. aegypti*, the vector of yellow fever, has a diploid chromosome number 6, and about 60 mutations have been mapped among its three linkage groups. Among these are genes conferring resistance to insecticides such as DDT and pyrethrins (*both of which see*).

Aegilops a genus of grasses including several species of genetic interest, especially *A. umbellulata*, a wild Mediterranean species resistant to leaf rust. A gene for rust resistance has been transferred from *A. umbellulata* to *Triticum vulgare* (wheat).

aerobe an organism that requires molecular oxygen and lives in an environment in contact with air.

aestivate to pass through a hot, dry season in a torpid condition. *See also* hibernate.

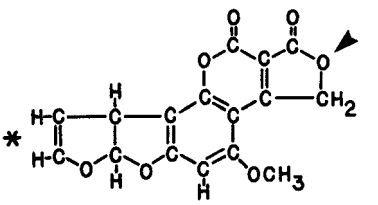
afferent leading toward the organ or cell involved. In immunology, the events or stages involved in activating the immune system. *Compare with* efferent.

affinity in immunology, the innate binding power of an antibody combining site with a single antigen binding site. *Compare with* avidity.

affinity chromatography a technique for separating molecules by their affinity to bind to ligands (e.g., antibodies) attached to an insoluble matrix (e.g., Sepharose). The bound molecules can subsequently be eluted in a relatively pure state.

afibrinogenemia an inherited disorder of the human blood-clotting system characterized by the inability to synthesize fibrinogen; inherited as an autosomal recessive.

aflatoxins a family of toxic compounds synthesized by *Aspergillus flavus* and other fungi belonging to the same genus. Aflatoxins bind to purines, making base pairing impossible, and they inhibit both DNA replication and RNA transcription. These mycotoxins are highly toxic and carcinogenic, and they often are contaminants of grains and oilseed products that are stored under damp conditions. The structure of aflatoxin G_1 is shown.



Aflatoxin B_1 has a CH_2 substituted for the O at the position marked by the arrow. Aflatoxin B_2 and G_2 are identical to B_1 and G_1, except that the ring labeled with an asterisk lacks a double bond.

African bees *Apis mellifera scutellata*, a race of bees, originally from South Africa, that was accidentally introduced into Brazil in 1957 and has spread as far as the southern United States. African bees are poor honey producers and tend to sting much more often than European bees. Because of daily differences in flight times of African queens and European drones, hybridization is rare. *See Apis mellifera.*

African Eve *See* mitochondrial DNA lineages.

African green monkey *See Cercopithecus aethiops.*

agamete a haploid, asexual reproductive cell resulting from meiosis in an agamont. Agametes disperse and grow into gamonts (*q.v.*).

agammaglobulinemia the inability in humans to synthesize certain immunoglobulins. The most common form is inherited as an X-linked recessive trait, which is symbolized XLA (X-linked agammaglobulinemia) in the early literature. When O. C. Bruton described the condition in 1952, it was the first hereditary immune disease to be reported. The disease

is now known to be caused by mutations in a gene at Xq21.3–q22. The gene is 36,740 bp long, and it encodes a protein containing 659 amino acids. The protein is a tyrosine kinase that has been named in Bruton's honor, and the gene is now symbolized *BTK*, for its product, the Bruton tyrosine kinase. The enzyme is a key regulator in the development of B lymphocytes. Boys with XLA lack circulating B cells. The bone marrow contains pre-B cells, but they are unable to mature. *See* antibody.

agamogony the series of cellular or nuclear divisions that generates agamonts.

agamont the diploid adult form of a protoctist that also has a haploid adult phase in its life cycle. An agamont undergoes meiosis and produces agametes. *See* gamont.

agamospermy the formation of seeds without fertilization. The male gametes, if present, serve only to stimulate division of the zygote. *See* apomixis.

agamous *See* floral identity mutations.

Agapornis a genus of small parrots. The nest building of various species and their hybrids has provided information on the genetic control of behavior patterns.

agar a polysaccharide extract of certain seaweeds used as a solidifying agent in culture media.

agarose a linear polymer of alternating D-galactose and 3,6-anhydrogalactose molecules. The polymer, fractionated from agar, is often used in gel electrophoresis because few molecules bind to it, and therefore it does not interfere with electrophoretic movement of molecules through it.

agar plate count the number of bacterial colonies that develop on an agar-containing medium in a petri dish seeded with a known amount of inoculum. From the count, the concentration of bacteria per unit volume of inoculum can be determined.

age-dependent selection selection in which the values for relative fitness of different genotypes vary with the age of the individual.

agglutination the clumping of viruses or cellular components in the presence of a specific immune serum.

agglutinin any antibody capable of causing clumping of erythrocytes, or more rarely other types of cells.

agglutinogen an antigen that stimulates the production of agglutinins.

aggregation chimera a mammalian chimera made through the mingling of cells of two embryos. The resulting composite embryo is then transferred into the uterus of a surrogate mother where it comes to term. *See* allophenic mice.

aging growing old, a process that has a genetic component. Hereditary diseases are known in humans that cause premature aging, and mutations that speed up or delay aging have been isolated in *Saccharomyces*, *Caenorhabditis*, and *Drosophila*. *See* Appendix C, 1994, Orr and Sohal; 1995, Feng *et al.*; antioxidant enzymes, apoptosis, *daf-2*, free radical hypothesis of aging, *Indy*, *methuselah*, *Podospora anserina*, progeria, SGSI, telomerase, senescence, Werner syndrome.

agonistic behavior any social interaction between members of the same species that involves aggression or threat and conciliation or retreat.

agouti the grizzled color of the fur of mammals resulting from alternating bands of yellow (phaeomelanin) and black (eumelanin) pigments in individual hairs. The name is also given to the genes that control the hair color patterns. In the mouse more than 20 alleles have been described at the *agouti* locus on chromosome 2. The gene encodes a cysteine-rich, 131 amino acid protein that instructs the melanocytes in the hair follicle when to switch from making black to yellow pigment. The protein is translated by nearby follicle cells rather than in the melanocytes themselves. Therefore, the agouti protein acts as a paracrine-signaling molecule. *See* Appendix C, 1905, Cuénot; autocrine, *MC1R* gene, melanin.

agranular reticulum *See* smooth endoplasmic reticulum (SER).

agranulocytes white blood cells whose cytoplasm contains few or no granules and that possess an unlobed nucleus; mononuclear leucocytes including lymphocytes and monocytes.

agriculturally important species *See* Appendix B.

Agrobacterium tumefaciens the bacterium responsible for crown gall disease (*q.v.*) in a wide range of dicotyledonous plants. The bacterium enters only dead, broken plant cells and then may transmit a tumor-inducing plasmid into adjacent living plant cells. This infective process is a natural form of genetic engineering, since the bacterium transfers part of its DNA to the infected plant. This

is integrated into the plant genome, and here it induces changes in metabolism and tumor formation. The genome of strain 58 of *A. tumefaciens* was sequenced in 2001 by a group led by E. W. Nestor. Its genome contained 5.67 mbp of DNA distributed among four replicons: a circular chromosome (CC), a linear chromosome (LC), and two plasmids (pAt and pTi). The plasmids are DNA circles, and most of the genes responsible for crown galls are on pTi. The Table lists the general features of the four replicons.

Replicon	Size (kbp)	ORFs	tRNAs	rRNAs
CC	2,841	2,789	40	2
LC	2,076	1,882	13	2
pAt	543	550	0	0
pTi	214	198	0	0

Strains of *A. tumefaciens* carrying the plasmid may be artificially genetically engineered to introduce foreign genes of choice into plant cells, and then by growing the cells in tissue culture, whole plants can be regenerated, every cell of which contains the foreign gene. *See* Appendix A, Bacteria, Proteobacteria; Appendix C, 1907, Smith and Townsend; 1981, Kemp and Hall; 2001, Wood *et al.*; Appendix E; Ti plasmid.

Agropyron elongatum a weed related to crabgrass noted for its resistance to stem rust. Genes conferring rust resistance have been transferred from this species to *Triticum aestivum* (wheat).

AHC Achilles' heel cleavage (*q.v.*).

AHF antihemophilic factor. *See* blood clotting.

AI, AID, AIH *See* artificial insemination.

AIA anti-immunoglobulin antibodies, produced in response to foreign antibodies introduced into an experimental animal.

AIDS the *a*cquired *i*mmuno*d*eficiency *s*yndrome, a disease caused by the human immunodeficiency virus (HIV). This virus attacks lymphocytes of helper T subclass and macrophages. The depletion of these cells makes the patient susceptible to pathogens that would easily be controlled by a healthy immune system. The infection is transmitted by sexual intercourse, by direct contamination of the blood (as when virus-contaminated drug paraphernalia is shared), or by passage of the virus from an infected mother to her fetus or to a suckling baby. AIDS was first identified as a new infectious disease by the U.S. Centers for Disease Control and Prevention in 1981. *See* Appendix C, 1983, Montagnier and Gallo. HIV, lymphocyte, retroviruses.

akinetic acentric (*q.v.*).

ala alanine. *See* amino acid.

albinism **1.** deficiency of chromoplasts in plants. **2.** the inability to form melanin (*q.v.*) in the eyes, skin, and hair, due to a tyrosinase deficiency. In humans the condition is inherited as an autosomal recessive. Tyrosinase (TYR) is an essential enzyme for melanin synthesis, and some mutations in the tyrosinase gene (*tyr*) result in oculocutaneous albinism (OCA). The *TYR* gene is located in 11q14-21; it contains five exons, and its mRNA is 2,384 nucleotides long. More than 90 mutations have been identified, most of the missense type. One such mutation in codon 422 results in the substitution of glutamine for arginine. The changed enzyme is heat-sensitive and so mimics the temperature-sensitive enzymes known for the Himalayan strains of mice, rabbits, and other species. *See* Himalayan mutant, ocular albinism, temperature-sensitive mutation, tyrosinase.

albino **1.** a plant lacking chromoplasts. **2.** an animal lacking pigmentation. *See* melanin.

albomaculatus referring to a variegation consisting of irregularly distributed white and green regions on plants resulting from the mitotic segregation of genes or plastids.

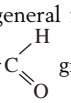
albumin a water-soluble 70-kilodalton protein that represents 40–50% of the plasma protein in adult mammals. It is important both as an osmotic and as a pH buffer and also functions in the transport of metal ions and various small organic molecules. Albumin is synthesized and secreted by the liver. In the mouse the albumin gene resides on chromosome 5, separated from the alpha fetoprotein gene by a DNA segment about 13.5 kilobases long. In humans, these two genes are in the long arm of chromosome 4. *See* Appendix C, 1967, Sarich and Wilson; alpha fetoprotein.

alcaptonuria alkaptonuria (*q.v.*).

alcohol any hydrocarbon that carries one or more hydroxyl groups. The term is often used to refer specifically to ethyl alcohol, the product of yeast-based fermentations. Hereditary differences in alcohol preference are known to exist in mice. *See* Appendix C, 1962, Rodgers and McClearn.

alcohol dehydrogenase (ADH) a zinc-containing enzyme found in bacteria, yeasts, plants, and animals that reversibly oxidizes primary and secondary alcohols to the corresponding aldehydes and ketones. In the case of yeast, ADH functions as the last enzyme in alcoholic fermentation. In *Drosophila melanogas-*

ter, ADH is a dimeric protein. By suitable crosses between null activity mutants it is possible to generate heteroallelic individuals that exhibit partial restoration of enzyme activity. This is often due to the production of a heterodimer with improved functional activity. The gene is of interest to developmental geneticist because its expression is controlled by *two* promotors. The proximal promotor lies adjacent to the initiation codon and switches the gene on during the larval stage. The distal promotor lies 700 base pairs upstream and controls the production of ADH in the adult. *See* allelic complementation, promotor.

aldehyde any of a class of organic compounds having the general formula $C_nH_{2n}O$ and containing a terminal  group.

aldosterone an adrenal corticosteroid hormone that controls the sodium and potassium balance in the vertebrates.

aleurone the outer layer of the endosperm of a seed. Genes controlling the inheritance of aleurone color in maize provided early examples of epistasis (*q.v.*) and parental imprinting (*q.v.*). *See* kernel.

aleurone grain a granule of protein occurring in the aleurone.

Aleutian mink an autosomal recessive mutation in *Mustela vison* producing diluted pigmentation of the fur and eyes. The homozygotes show a lysosomal defect similar in humans to the Chédiak-Higashi syndrome (*q.v.*).

alga (*plural* algae) any of a large group of aquatic, chlorophyll-bearing organisms ranging from single cells to giant seaweeds. *See* Appendix A: Cyanobacteria, Dinoflagellata, Euglenophyta, Xanthophyta, Chrysophyta, Bacillariophyta, Phaecophyta, Rhodophyta, Gamophyta, Chlorophyta.

algorithm a set of simple mathematical procedures that are followed in a specified order to solve a problem in a finite period of time. Computers are instructed to perform tasks with programs containing one or more algorithms.

alien addition monosomic a genome that contains a single chromosome from another species in addition to the normal complement of chromosomes.

alien substitution replacement of one or more chromosomes of a species by those from a different species.

aliphatic designating molecules made up of linear chains of carbon atoms.

aliquot a part, such as a representative sample, that divides the whole without a remainder. Two is an aliquot of six because it is contained exactly three times. Loosely used for any fraction or portion.

alkali metal any of five elements in Group IA of the periodic table: lithium (Li), sodium (Na), potassium (K), rubidium (Rb), and cesium (Cs).

alkaline earth any element of Group IIA of the periodic table: beryllium (Be), magnesium (Mg), calcium (Ca), strontium (Sr), barium (Ba), and radium (Ra).

alkaline phosphatase an enzyme that removes 5′-P termini of DNA and leaves 5′-OH groups. The alkaline phosphatase of *E. coli* is a dimer made up of identical protein subunits encoded by a single structural gene. *In vitro* complementation was demonstrated using this enzyme. *See* allelic complementation.

alkaloid any member of a group of over 3,000 cyclic, nitrogenous, organic compounds, many of which have pharmacological properties. They occur mainly in plants, but are also synthesized by some fungi, amphibians, and arthropods. They include caffeine, cocaine, quinine, morphine, nicotine, reserpine, strychnine, and theobromine.

alkapton 2,5-dihydroxyphenylacetic acid. *See* homogentisic acid.

alkaptonuria (*also* alcaptonuria) a relatively benign hereditary disease in humans due to a recessive gene located on the long arm of chromosome 3. Alkaptonurics cannot make the liver enzyme homogentisic acid oxidase. Therefore, homogentisic acid (*q.v.*) is not broken down to simpler compounds but is excreted in the urine. Since the colorless homogentisic acid is readily oxidized to a black pigment, the urine of alkaptonurics darkens when exposed to air. This disease enjoys the historic distinction of being the first metabolic disease studied. *See* Appendix C, 1909, Garrod.

alkylating agent a compound causing the substitution of an alkyl group (usually methyl or ethyl) for an active hydrogen atom in an organic compound. According to the number of reactive groups they contain, alkylating agents are classified as mono-, bi-, or polyfunctional. Many chemical mutagens are alkylating agents. *See* busulfan, chlorambucil, cyclophosphamide, epoxide, ethylmethane sulfonate, melphalan, Myleran, nitrogen mustard, sulfur mustard, TEM, Thio-tepa, triethylenethiophosphoramide.

alkyl group a univalent radical having the general formula C_nH_{2n+1} derived from a saturated aliphatic hydrocarbon by removal of one atom of hydrogen. Named by replacing the ending *-ane* of the hydrocarbon with *-yl* (e.g., meth*ane* becomes meth*yl*).

allantois a saclike outgrowth of the ventral side of the hindgut present in the embryos of reptiles, birds, and mammals. The allantois represents a large and precocious development of the urinary bladder.

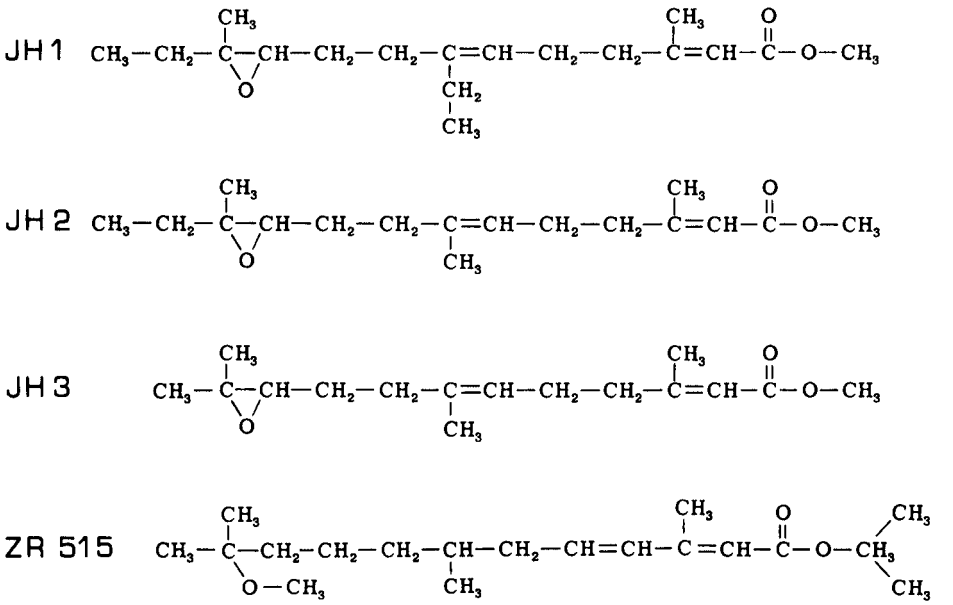
allatum hormones hormones synthesized by the insect corpus allatum. The titer of allatum hormones influences the qualitative properties of each molt in holometabolous insects. At high concentrations, larval development ensues; at lower levels, the insect undergoes pupal metamorphosis, and in the absence of the allatum hormones adult differentiation takes place. The allatum hormones thus have a juvenilizing action and for this reason have also been called juvenile hormones (JHs). The structural formulas for three of the juvenile hormones are illustrated on page 14. In adult females, the allatum hormone is required for vitellogenesis. The JH analog, ZR515 (*q.v.*), is often used as a substitute for natural JHs in *Drosophila* experiments. *See* Appendix C, 1966, Röller *et al.*; ring gland, *status quo* hormones.

allele a shorthand form of **allelomorph,** one of a series of possible alternative forms of a given gene (cistron, *q.v.*), differing in DNA sequence, and affecting the functioning of a single product (RNA and/or protein). If more than two alleles have been identified in a population, the locus is said to show *multiple allelism. See* heteroallele, homoallele, isoallele, null allele, silent allele.

allele-specific oligonucleotide testing a technique used to identify a specific mutation in a collection of DNA fragments isolated from a mutant organism. An oligonucleotide is synthesized that has a base sequence complementary to the segment under study, and it is used as a probe. All segments binding to the probe are then collected and analyzed.

allelic complementation the production of nearly normal phenotype in an organism carrying two different mutant alleles in *trans* configuration. Such complementation is sometimes caused by the reconstruction in the cytoplasm of a functional protein from the inactive products of the two alleles. When such a phenomenon can be demonstrated by mixing extracts from individuals homozygous for each allele, the term *in vitro complementation* is used. Synonymous with intra-allelic complementation. *See* Appendix C, 1963, Schlesinger and Levinthal; alcohol dehydrogenase, alkaline phosphatase, transvection.

allelic exclusion the situation in a diploid nucleus where either the parental or the maternal allele, but not both, is expressed, even though both parental alleles are capable of being transcribed and may even be identical. This situation is seen during recombina-



Allatum hormones

tion within the segmented *Ig* genes of immature lymphocytes. In any one B lymphocyte (*q.v.*), a light chain or heavy chain can be synthesized from a maternal or paternal homolog, not both. *See* immunoglobulin genes, isotype exclusion, somatic recombination.

allelic frequency the percentage of all alleles at a given locus in a population gene pool represented by a particular allele. For example, in a population containing 20 *AA*, 10 *Aa*, and 5 *aa*, the frequency of the *A* allele is [2(20) + 1(10)]/2(35) = 5/7 = 0.714. *See* gene frequency.

allelism test complementation test (*q.v.*).

allelomorph commonly shortened to allele (*q.v.*). *See* Appendix C, 1900, Bateson.

allelopathy an interaction involving two different species in which chemicals introduced into the environment by one suppress the growth or reproduction of the other.

allelotype the frequency of alleles in a breeding population.

allergen a substance inducing hypersensitivity.

allergy an immune hypersensitivity response to an agent that is nonantigenic to most of the individuals in a population.

allesthetic trait any individual characteristic that has an adaptive function only via the nervous systems of other organisms, for example, odors, display of color patterns, mating calls, etc., which are important components of courtship in various species. *See* courtship ritual, pheromone.

Allium the genus that includes *A. cepa*, the onion; *A. porrum*, the leek; *A. sativum*, the garlic; and *A. schoenoprasum*, the chive—all classic subjects for cytological studies of mitotic chromosomes.

alloantigen an antigen (*q.v.*) that elicits an immune response (*q.v.*) when introduced into a genetically different individual of the same species. Antibodies produced in response to alloantigens are called *alloantibodies*. *See* histocompatibility molecules.

allochromacy the formation of other coloring agents from a given dye that is unstable in solution. Nile blue (*q.v.*) exhibits allochromacy.

allocycly a term referring to differences in the coiling behavior shown by chromosomal segments or whole chromosomes. Allocyclic behavior characterizes the pericentric heterochromatin, the nucleolus organizer, and in some species entire sex chromosomes. If a chromosome or chromosomal segment is tightly condensed in comparison with the rest of the chromosomal complement, the chromosome or chromosomal segment is said to show *positive heteropycnosis* (*q.v.*). Allocycly is also used to describe asynchronous separation of bivalents during the first anaphase in meiosis. In man, for example, the X and Y chromosomes segregate ahead of the autosomes and are said to show *positive allocycly*.

allogeneic disease *See* graft-versus-host reaction.

allogeneic graft a graft of tissue between genetically different members of the same species, especially with regard to alloantigens (*q.v.*). *See* allograft, heterograft. *Compare with* xenograft.

allograft a graft of tissue from a donor of one genotype to a host of a different genotype but of the same species.

allolactose *See* lactose.

allometry the relation between the rate of growth of a part of an individual and the growth rate of the whole or of another part. In the case of isometry, the relative proportions of the body parts remain constant as the individual grows; in all other cases, the relative proportions change as total body size increases. *See* heterauxesis.

allomone any chemical secreted by an organism that influences behavior in a member of another species, benefiting only the producer. If both species benefit, it is a *synamone*. If only the receiver benefits, it is a *kairomone*.

alloparapatric speciation a mode of gradual speciation in which new species originate through populations that are initially allopatric, but later become parapatric before completely effective reproductive isolation has evolved. Natural selection may enhance incipient reproductive isolating mechanisms in the zone of contact by character displacement (*q.v.*), and other mechanisms. *Compare with* parapatric speciation.

allopatric speciation the development of distinct species through differentiation of populations in geographic isolation. Such populations are called allopatric.

allopatry referring to species living in different geographic locations and separated by distance alone or by some barrier to migration such as a mountain range, river, or desert. *Compare with* sympatry.

allophene a phenotype not due to the mutant genetic constitution of the cells of the tissue in question. Such a tissue will develop a normal phenotype if transplanted to a wild-type host. *See* autophene.

allophenic mice chimeric mice produced by removing cleaving eggs from mice of different genotypes, fusing the blastomeres *in vitro*, and reimplanting the fused embryos into the uterus of another mouse to permit embryogenesis to continue. Viable mice containing cells derived from two or more embryos have been obtained and used in cell lineage studies. *See* Appendix C, 1967, Mintz.

alloplasmic referring to organisms or cells bearing chromosomes of one species and cytoplasm of a different species; for example, bread wheat (*Triticum aestivum*) chromosomes and rye (*Secale cereale*) cytoplasm. C*ompare with* heteroplasmic, heteroplastidy.

allopolyploid (*also* alloploid) a polyploid organism arising from the combination of genetically distinct chromosome sets. *See* isosyndetic alloploid, segmental alloploid.

alloprocoptic selection a mode of selection in which association of opposites increases the fitness of the associates. An example involves the loci governing alcohol dehydrogenase in *Drosophila melanogaster*. The fertility is greater than expected when two mating individuals are homozygous for different alleles and smaller than expected when they are homozygous for the same allele.

allostery the reversible interaction of a small molecule with a protein molecule, which leads to changes in the shape of the protein and a consequent alteration of the interaction of that protein with a third molecule.

allosteric effectors small molecules that reversibly bind to allosteric proteins at a site different from the active site, causing an allosteric effect.

allosteric enzyme a regulatory enzyme whose catalytic activity is modified by the noncovalent attachment of a specific metabolite to a site on the enzyme other than the catalytic site.

allosteric protein a protein showing allosteric effects.

allosteric site a region on a protein other than its active site (*q.v.*), to which a specific effector molecule may bind and influence (either positively or negatively) the functional activity of the protein. For example, in the lactose system of *E. coli*, the *lac* repressor becomes inactive (cannot bind to the *lac* operator) when allolactose is bound to the allosteric site of the repressor molecule. *See lac* operon.

allosyndesis the pairing of homoeologous chromosomes (*q.v.*) in an allopolyploid (*q.v.*). Thus if the genetic composition of an alloploid is given by AABB, where AA represent the chromosomes derived from one parent species and BB the chromosomes derived from the other parent species, then during meiotic prophase, A undergoes allosyndetic pairing with B. Such pairing indicates that the A and B chromosomes have some segments that are homologous, presumably because the two parent species have a common ancestry. In the case of *autosyndesis*, A pairs only with A, and B with B. *Segmental alloploids* form both bivalents and multivalents during meiosis because of allosyndesis.

allotetraploid an organism that is diploid for two genomes, each from a different species; synonymous with amphidiploid (*q.v.*).

allotypes proteins that are products of different alleles of the same gene. The term is often used to refer to serologically detectable variants of immunoglobins and other serum proteins.

allotype suppression the systematic and long-term suppression of the expression of an immunoglobulin allotype in an animal induced by treatment with antibodies against the allotype.

allotypic differentiation *See in vivo* culturing of imaginal discs.

allozygote an individual homozygous at a given locus, whose two homologous genes are of independent origin, as far as can be determined from pedigree information. *See* autozygote.

allozymes allelic forms of an enzyme that can be distinguished by electrophoresis, as opposed to the more general term *isozyme* (*q.v.*). *See* Appendix C, 1966, Lewontin and Hubby.

alpha amanitin *See* amatoxins.

alpha chain one of the two polypeptides found in adult and fetal hemoglobin (*q.v.*).

alpha fetoprotein the major plasma protein of fetal mammals. AFP is a 70-kilodalton glycoprotein that is synthesized and secreted by the liver and the yolk sac. The genes encoding AFP and serum albumen arose in evolution as the result of a duplication of an ancestral gene $(3–5) \times 10^8$ years ago. *See* albumen.

alpha galactosidase an enzyme that catalyzes the hydrolysis of substrates that contain α-galactosidic residues, including glycosphingolipids and glycopro-

teins. In humans, α-galactosidase exists in two forms, A and B. The A form is encoded by a gene on the X chromosome. Fabry disease (*q.v.*) is caused by mutations at this locus. The B form is encoded by a gene on chromosome 22.

alpha helix one of two common, regularly repeating structures seen in proteins (*compare with* beta pleated sheet). The alpha helix is a compact spiral with the side chains of the amino acids in the polypeptide extending outward from the helix. The helix is stabilized by hydrogen bonds that form between the CO group of each amino acid and the NH group of the amino acid, which lies four residues ahead in the sequence. All main-chain CO and NH groups are hydrogen-bonded according to this pattern. One turn of the helix occurs for each 3.6 amino acid residues. Alpha helices are built from a continuous sequence that contains as few as 4 to as many as 50 amino acids. *See* Appendix C, 1951, Pauling and Corey; 1958, Kendrew *et al.*; protein structure.

alpha particle a helium nucleus consisting of two protons and two neutrons, and having a double positive charge.

alpha tocopherol vitamin E (*q.v.*).

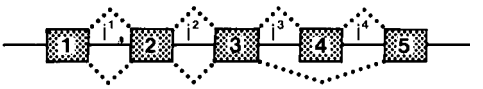
alphoid sequences a complex family of repetitive DNA sequences found in the centromeric heterochromatin of human chromosomes. The alphoid family is composed of tandem arrays of 170 base pair segments. The segments isolated from different chromosomes show a consensus sequence, but also differences with respect to individual bases, so that the 170 base pair units may vary in sequence by as much as 40%. The repeats are organized in turn into groups containing several units in tandem, and these groups are further organized into larger sequences 1 to 6 kilobases in length. These large segments are then repeated to generate segments 0.5 to 10 megabase pairs in size. Such larger, or "macro," DNA repeats are chromosome-specific. Since alphoid sequences are not transcribed, they play an as yet undefined structural role in the chromosome cycle. The variation in the sequences within the alphoid DNA results in a high frequency of RFLPs. These are inherited and can be used to characterize the DNAs of specific individuals and their relatives. *See* DNA fingerprint technique, restriction fragment length polymorphisms.

alteration enzyme a protein of phage T4 that is injected into a host bacterium along with the phage DNA; this protein modifies host RNA polymerase by linking it to ADP-ribose. RNA polymerase modified in this way renders it incapable of binding to sigma factor and thus unable to initiate transcription at host promoters. *See* RNA polymerase.

alternate disjunction, alternate segregation *See* translocation heterozygote.

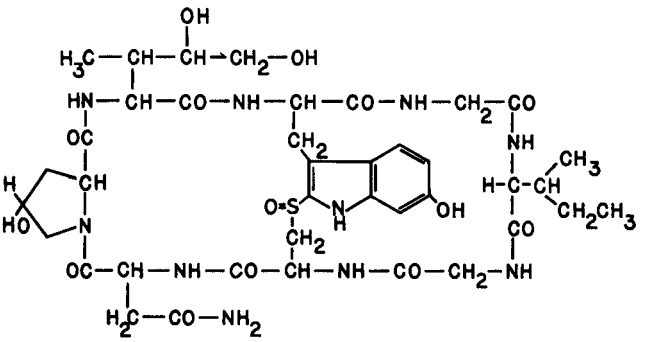
alternation of generations reproductive cycles in which a haploid phase alternates with a diploid phase. In mosses and vascular plants, the haploid phase is the gametophyte, the diploid the sporophyte.

alternative splicing a mechanism for generating multiple protein isoforms from a single gene that involves the splicing together of nonconsecutive exons during the processing of some, but not all, transcripts of the gene. This is illustrated in the diagram, where a gene is made up of five exons joined by introns i^1–i^4. The exons may be spliced by the upper pathway shown by the dotted lines to generate a mature transcript containing all five exons. This type of splicing is termed *constitutive*. The alternative mode of splicing shown generates a mature transcript that lacks exon 4. If each exon encodes 20 amino acids, the constitutive splicing path would result in a polypeptide made up of 100 amino acids. The alternative path would produce a polypeptide only 80 amino acids long. If the amino acid sequences of the two proteins were determined, the first 60 and the last 20 would be identical. The premessenger RNAs (*q.v.*) of at least 40% of all human genes undergo alternative splicing. This removes the intron RNAs and joins the adjacent exon RNAs by phosphodiester linkages. The splicing takes place in spliceosomes (*q.v.*) that reside within the nucleus. Therefore the number of proteins encoded by the human genome is many times larger than the number of structural genes it contains. *See* Appendix C, 1977, Weber *et al.*; adenovirus, DSCAM, fibronectin, Human Genome Project, isoforms, posttranscriptional processing, myosin genes, RNA splicing, tropomyosin.



altricial referring to the type of ontogeny seen in vertebrate species characterized by large litters, short gestations, and the birth of relatively undeveloped, helpless young. *Compare with* precocial.

altruism behavior of an individual that benefits others. To the extent that the "others" are related to the altruist (the one exhibiting altruistic behavior), such actions may actually be an expression of fitness. *See* inclusive fitness.



Alpha amanitin

Alu family the most common dispersed, repeated DNA sequence in the human genome. There are at least 750,000 Alu elements, each consisting of about 300 base pairs, accounting for 11% of human DNA. Each element is made up of two 130 base pair sequences joined head to tail with a 32 base pair insert in the right-hand monomer. Alu sequences are targeted by cohesins (*q.v.*). The family name is derived from the fact that these sequences are cleaved by restriction endonuclease *Alu I*. *See* human gene maps, repetitious DNA.

Alzheimer disease (AD) a multifactorial syndrome that causes a devastating decline in mental ability and is accompanied by the appearance of amyloid plaques in the cerebral cortex. These deposits were first observed in 1906 by a German physician, Alois Alzheimer, in brain tissue from a woman who had died of an unusual mental illness. Amyloid plaques contain aggregates of amyloid-beta-peptides (AβPs), and these are derived from an amyloid beta precursor protein (AβPP), which is encoded by a gene on human chromosome 21. Patients with trisomy 21 (Down syndrome, *q.v.*) commonly develop AD by age 40. Familial, early-onset AD is often associated with mutations of genes that encode presenilins (PS1 and PS2). The genes for PS1 and PS2 are located at 14q24.3 and 1q42.1, respectively. The proteins they encode are made of 467 and 448 amino acids, respectively, and they contain seven to nine transmembrane domains. Both proteins are bound to membranes and play a role in cutting AβPP into AβPs, some of which are toxic. A gene homologous to the PS1 gene has been isolated from nematodes. *See* Appendix C, 1995, Sherrington, St. George-Hyslop *et al.*, Schellenberg *et al.*; *Caenorhabiditis elegans*, neuregulins (NRGs).

Amanita phalloides a poisonous mushroom which is the source of amatoxins and phallotoxins (*both of which see*). *See* Appendix A, Fungi, Basidiomycota.

amastigote *See* undulipodium.

amatoxins a group of bicyclic octapeptides that are among the poisons produced by *Amanita phalloides* (*q.v.*). These poisons inhibit transcription in eukaryotic cells because of their interaction with RNA polymerase II. However, they do not affect the RNA polymerases of mitochondria or chloroplasts. Alpha amanitin (formula, above) is an amatoxin most commonly used experimentally to inhibit transcription. *See* phallotoxins, RNA polymerase.

amaurosis blindness occurring without an obvious lesion in the eye, as from a disease of the optic nerve or brain. The term is sometimes found in the early descriptions of hereditary diseases leading to blindness (e.g., Leber congenital amaurosis). *See* Leber hereditary optic neuropathy (LHON).

amber codon the mRNA triplet UAG that causes termination of protein translation, one of three "stop" codons. The terms *amber* and *ochre* (*q.v.*) originated from a private laboratory joke and have nothing to do with colors.

Amberlite trade name for a family of ion-exchange resins.

amber mutation a mutation in which a polypeptide chain is terminated prematurely. Amber mutations are the result of a base substitution that converts a codon specifying an amino acid into UAG, which signals chain termination. In certain strains of *E. coli* amber mutations are suppressed. These strains contain a tRNA with an AUC anticodon, which inserts an amino acid at the UAG site and hence permits translation to continue. *See* ochre mutation, nonsense mutation.

amber suppressor any mutant gene coding for a tRNA whose anticodon can respond to the UAG stop codon by the insertion of an amino acid that renders the gene product at least partially functional.

For example, a mutant tyrosine-tRNA anticodon 3′AUC would recognize 5′UAG, tyrosine would be inserted, and chain growth would continue.

Ambystoma mexicanum the Mexican axolotl, a widely used laboratory species. The urodele for which the most genetic information is available. It was in the nuclei of oocytes of this species that the giant lampbrush chromosomes (*q.v.*) were first observed. *Ambystoma* has 14 pairs of chromosomes, and a genome size of about 35 gbp of DNA. It is one of the few vertebrates able to regenerate entire body structures. *See* Appendix A, Animalia, Chordata, Vertebrata, Amphibia, Urodela; Appendix C, 1882, Flemming; neoteny, regeneration.

amelogenins highly conserved proteins that are secreted by ameloblasts and constitute 90% of the organic matrix in the enamel of teeth. The amelogenins of humans come in a number of isoforms; the most common one contains 192 amino acids. The genes that encode amelogenins reside on the X and Y chromosomes. *AMELX* is at Xp22.22 and *AMELY* is at Yp11.2, and both are transcribed in the tooth buds of males. The X-linked gene contains 7,348 bases and the Y-linked gene 8,109. During forensic analyses, *amelogenin* primers from human X-Y DNA are often used in gender determination.

amelanogenesis imperfecta defects in the mineralization of teeth that affect the enamel layer. The teeth are small, pitted, and show yellow to brown discolorations. The condition often results from mutations in the genes that encode amelanogenins (*q.v.*).

amensalism a species interaction in which one is adversely affected and the other is unaffected.

Ames test a bioassay for detecting mutagenic and possibly carcinogenic compounds, developed by Bruce N. Ames in 1974. Reverse mutants to histidine independence are scored by growing *his*$^-$ *Salmonella typhimurium* on plates deficient in histidine in the presence of the chemical (test) and in its absence (control).

amethopterin methotrexate (*q.v.*).

amino acid activation a coupled reaction catalyzed by a specific aminoacyl synthetase that attaches a specific amino acid (AA) to a specific transfer RNA (tRNA) in preparation for translation (*q.v.*).

$$\text{AA} + \text{ATP} \rightarrow \text{AA-AMP} + 2\text{P}$$
$$\text{AA-AMP} + \text{tRNA} \rightarrow \text{AA-tRNA} + \text{AMP}$$

amino acid attachment site the 3′ end of a tRNA molecule to which an amino acid is covalently attached by an aminoacyl bond. *See* amino acid activation, aminoacyl-tRNA synthetases, transfer RNA.

amino acids aminocarboxylic acids that are components of proteins and peptides. They also occur in their free form or attached to transfer RNAs (*q.v.*). There are 20 different amino acids for which at least one specific codon exists in the DNA genetic code. These universal amino acids are illustrated on page 21. Their abbreviations and messenger RNA code designations are on page 22. Amino acids are joined together to form polypeptides. Polymers containing 50 or more amino acids are called proteins. All amino acids contain a central carbon atom (designated alpha) to which an amino group, a carboxyl group, and a hydrogen atom are attached. There is also a side chain or residue (R), and this gives each amino acid its characteristic properties. Note that proline is unique in that the alpha C and its amino group are incorporated into the side chain, which is in the form of a five-atom ring. At pH 7 the side chains of lysine, arginine, and histidine are positively charged and the side chains of aspartic acid and glutamic acid are negatively charged. Therefore, the net charge born by a protein is determined by the relative proportions of these five amino acids in it. Other amino acids control the shape of proteins. Amino acids like isoleucine, leucine, phenylalanine, and valine are repelled by water molecules and therefore tend to be found buried within the interior of the protein structure. *See* genetic code, peptide bond, translation.

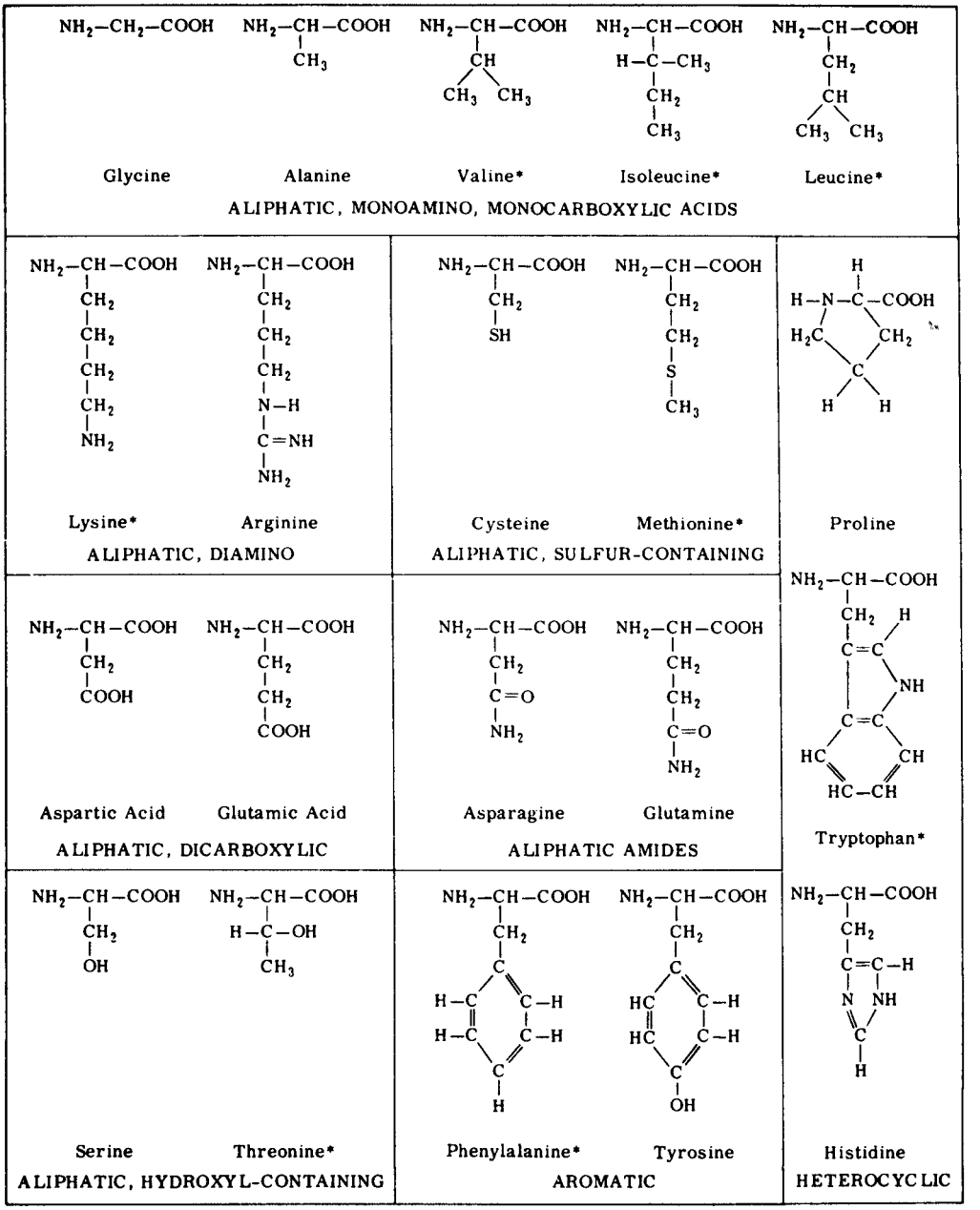
amino acid sequence the linear order of the amino acids in a peptide or protein. *See* protein structure.

amino acid side chain a group attached to an amino acid, represented by R in the general formula for an amino acid:

$$\begin{array}{l} \text{NH}_2-\text{CH}-\text{COOH} \\ \quad\quad\;\; | \\ \quad\quad\;\; \text{R} \end{array}$$

aminoaciduria the presence of one or more amino acids in the urine in abnormal quantities because of a metabolic defect.

aminoacyl adenylate the activated compound that is an intermediate in the formation of a covalent bond between an amino acid and its specific transfer RNA; abbreviated AA-AMP. *See* AMP, transfer RNA.



*required in the diet of mammals.

Structural formulas of the universal amino acids

aminoacyl site one of two binding sites for tRNA molecules on a ribosome; commonly called the *A site*. *See* translation.

aminoacyl-tRNA an aminoacyl ester of a transfer RNA molecule.

aminoacyl-tRNA binding site *See* translation.

aminoacyl-tRNA synthetases enzymes that activate amino acids and attach each activated amino acid to its own species of tRNA. These enzymes catalyze: (1) the reaction of a specific amino acid (AA) with adenosine triphosphate (ATP) to form AA-AMP, and (2) the transfer of the AA-AMP complex to a specific transfer RNA, forming AA-tRNA and

Amino Acids

AMINO ACID	ONE-LETTER SYMBOL	THREE-LETTER SYMBOL	mRNA CODE DESIGNATION
alanine	A	ala	GCU, GCC, GCA, GCG
arginine	R	arg	CGU, CGC, CGA, CGG, AGA, AGG
asparagine	N	asn	AAU, AAC
aspartic acid	D	asp	GAU, GAC
cysteine	C	cys	UGU, UGC
glutamic acid	E	glu	GAA, GAG
glutamine	Q	gln	CAA, CAG
glycine	G	gly	GGU, GGC, GGA, GGG
histidine	H	his	CAU, CAC
isoleucine	I	ile	AUU, AUC, AUA
leucine	L	leu	UUA, UUG, CUU,CUC, CUA, CUG
lysine	K	lys	AAA, AAG
methionine	M	met	AUG
phenylalanine	F	phe	UUU, UUC
proline	P	pro	CCU, CCC, CCA, CCG
serine	S	ser	UCU, UCC, UCA, UCG, AGU, AGC
threonine	T	thr	ACU, ACC, ACA, ACG
tryptophan	W	trp	UGG
tyrosine	Y	tyr	UAU, UAC
valine	V	val	GUU, GUC, GUA, GUG

free AMP (adenosine monophosphate). *See* adenosine phosphate.

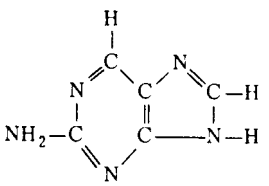
amino group a chemical group ($-NH_2$) which with the addition of a proton can form $-NH_3^+$.

p amino benzoic acid a component of folic acid (*q.v.*).

aminopeptidase an enzyme (in both prokaryotes and eukaryotes) that removes the formylated methionine (fMet) or methionine from the NH_2 terminus of growing or completed polypeptide chains.

aminopterin *See* folic acid.

aminopurine 2-aminopurine (2-AP) is a fluorescent analog of adenine (6-aminopurine). The fluorescence of 2-AP ia quenched once it is incorporated into the base-stacked structure of dsDNA. However, if the base stacking or base pairing of DNA is locally perturbed, the fluorescence of 2-AP is enhanced. Therefore the intensity of fluorescence emissions by 2-AP molecules can be used to monitor perturbations in DNA structure caused by interactions between DNA and proteins, such as DNA polymerases, helicases, repair enzymes, and methyl transferases *(all of which see)*.



amino terminal end the end of a polypeptide chain that has a free amino group.

Amish a human population descended from a limited number of founders who emigrated from southwestern Germany to the United States during the eighteenth century. The population is highly inbred, since marriage is allowed only within the community. Beneficial collaboration between geneticists and religious leaders has led to discoveries concerning certain genetic diseases that occur at unprecedentedly high frequencies among the Amish. *See* cartilage-hair hypoplasia (CHH), consanguinity, Ellis-van Creveld syndrome, inbreeding.

Amitochondriates a subkingdom of protoctists that includes the Archaeprotista and the Microspora (*see* Appendix A). These phyla contain anaerobic microorganisms that lack mitochondria and presumably were without them from the outset of their evolution.

amitosis the division of a nucleus into two parts by constriction without the participation of a mitotic apparatus. Accessory nuclei (*q.v.*) grow by amitosis.

amixis a reproductive cycle lacking meiosis and fertilization. Asexual reproduction. *Contrast with* amphimixis, apomixis.

***AML1* gene** *See acute myeloid leukemia 1* gene.

amniocentesis sampling of amniotic fluid for the prenatal diagnosis of fetal disorders. During the procedure, a hollow needle is inserted through the skin and muscle of the mother's abdomen, through the uterus, and into the amniotic sac that surrounds the fetus. Cells that have sloughed from the fetus are suspended in the fluid. Cells in the sample are cultured for about three weeks to raise their numbers to the point where chromosomal and biochemical analyses can be made. Amniocentesis cannot be done until about 16 weeks from the last menstrual period, since the sac containing the embryo is not large enough to permit safe withdrawal of the fluid until this time. *See* Appendix C, 1967, Jacobson and Barter; chorionic villi sampling, informed consent, genetic counseling, prenatal genetic testing.

amniocytes cells obtained by amniocentesis (*q.v.*).

amnion A fluid-filled sac within which the embryos of reptiles, birds, and mammals develop. The wall of this sac has a two-layered epithelium. The inner epithelium of the wall is the amnion, although the term is sometimes applied to the whole sac. The outer epithelium is usually called the chorion. Amniotic fluid within the sac provides a liquid environment for the embryo.

amniote a land-living vertebrate (reptile, bird, or mammal) whose embryos have an amnion and allantois.

Amoeba proteus a common species of rhizopod; a giant protozoan used for microsurgical nuclear transplantations. *See* Appendix C, 1967, Goldstein and Prescott.

amoeboid movement cellular motility involving cytoplasmic streaming into cellular extensions called *pseudopodia*.

amorphic mutation a mutation in which the altered gene product fails in its molecular function. Also called a *loss of function mutation* or a *null mutation*.

AMP adenosine monophosphate. *See* adenosine phosphate.

amphidiploid an organism that is diploid for two genomes, each from a different species; synonymous with allotetraploid. *See* Appendix C, 1925, Goodspeed and Clausen.

amphimixis sexual reproduction resulting in an individual having two parents: synonymous with mixis. *Contrast with* amixis, apomixis. The adjective form is *amphimictic*.

Amphioxus *See Branchiostoma*.

amphipathic descriptive of a molecule that has distinct polar and nonpolar segments (e.g., membrane phospholipids).

amphoteric compound (*also* ampholyte) a substance that can act both as an acid and a base. Thus a protein is amphoteric because it tends to lose protons on the more alkaline side of its isoelectric point and to gain protons on the acid side of its isoelectric point.

ampicillin *See* penicillin.

***amp*^{R}** a selectable gene which encodes the enzyme β-lactamase, which inactivates ampicillin (*q.v.*). Cells containing a plasmid vector (*q.v.*) which expresses *amp*^{R} can be selected from those that do not by growth in an ampicillin-containing medium. *See* penicillin, R (resistance) plasmid.

amplicon a segment of the genome that forms multiple linear copies after exposure of the organism to a compound that inhibits the functioning of a gene in the segment. For example, in mammals the enzyme dihydrofolate reductase (*q.v.*) is inhibited by methotrexate (*q.v.*). Exposure to this inhibitor causes amplification of the DHFR gene. More generally, the term *amplicons* is used for DNA fragments that have been generated in experiments utilizing the polymerase chain reaction (*q.v.*).

amplification *See* gene amplification, polymerase chain reaction, RNA amplification.

amplified RNA *See* RNA amplification.

amylase an enzyme that hydrolyzes glucosidic bonds in polyglucosans such as glycogen.

amyloid-beta-precursor protein (AβPP), amyloid-beta-peptides (AβPs), amyloid plaques. *See* Alzheimer disease (AD).

amyloplast a starch-rich plastid.

amyotrophic lateral sclerosis (ALS) a disease in humans resulting from the degeneration of motor neurons in the lateral columns of the spinal cord. The disease begins with an asymmetric weakness in the limbs and progresses to complete paralysis and death. ALS is sometimes called Lou Gehrig disease after the famous American baseball player who suffered from ALS. In familial cases of ALS, the condi-

tion was first shown to be due to mutations in a gene (*ALS1*) located at 21q22.1 which encoded the enzyme superoxide dismutase (SOD) (*q.v.*). Next *ALS2* was mapped to 2q33, and it encoded a protein (alsin) thought to be a GTPase regulator protein. Familial ALS due to *ALS1* is an adult-onset disease, and it shows dominant inheritance; whereas FALS due to *ALS2* causes a disease with juvenile onset which shows recessive inheritance. Two other forms of FALS occur: one caused by mutations in *ALS3* at 18q21 and the other by mutations in *ALS4* at 9q34. However, the cause of 90 percent of ALS cases (sporadic or non-familial) is unknown. *See* Appendix C, 1993, Rosen, Siddique *et al.*

anabolism the metabolic synthesis of complex molecules from simpler precursors, usually requiring the expenditure of energy and specific anabolic enzymes. *Contrast with* catabolism.

anaerobe a cell that can live without molecular oxygen. A strict anaerobe cannot live in the presence of oxygen. *See* Appendix C, 1861, Pasteur.

anagenesis phyletic evolution within a single lineage without subdivision or splitting; the opposite of cladogenesis.

analog a compound related to, but slightly different structurally from a biologically significant molecule, such as an amino acid (*see* azaserine), a pyrimidine or purine (*see* base analogs), or a hormone (*see* ZR515).

analogous referring to structures or processes that have evolved convergently, as opposed to the term *homologous* (*q.v.*). Analogous structures have similar functions but are different in evolutionary origin: e.g., the wing of a butterfly and of a bat. *See* homoplasy.

analysis of variance a statistical technique that allows the partitioning of the total variation observed in an experiment among several statistically independent possible causes of the variation. Among such causes are treatment effects, grouping effects, and experimental errors. Checking the absence of an effect due to the treatment is often the purpose of the inquiry. The statistical test of the hypothesis that the treatment had no effect is the *F* test, or variance-ratio test. If the ratio of the mean square for treatments to the mean square for error exceeds a certain constant that depends on the respective degrees of freedom of the two mean squares at a chosen significance level, then the treatments are inferred to have been effective. Analysis of variance is particularly useful in judging which sources of uncontrolled variation in an experiment need to be allowed for in testing treatment effects.

anamnestic response *See* immune response, immunological memory.

anaphase *See* mitosis.

anaphase lag delay in the movement of one or more chromosomes from the metaphase plate during anaphase, often resulting in chromosome loss (*q.v.*).

anaphylaxis a systemic allergic or hypersensitivity response leading to immediate respiratory and/or vascular difficulties.

Anas platyrhyncha the mallard duck, ancestor to the domestic or Pekin duck, *A.p. domestica*.

anastomosis the joining of two or more cell processes or tubular vessels to form a branching system.

anastral mitosis the type of mitosis characteristically found in plants. A spindle forms, but no centrioles or asters are observed.

anautogenous insect an adult female insect that must feed for egg maturation. *See* autogenous insect.

anchorage-dependent cells cells (or *in vitro* cell cultures) that will grow, survive, or maintain function only when attached to an inert surface such as glass or plastic; also known as *substrate-dependent cells*. The only normal animal cells that are designed to survive without attachment and spreading are cells that circulate in the blood. Some tumor cells acquire this ability to be anchorage-independent and leave their original tissue sites to form metastases. *See* microcarriers, suspension culture.

Anderson disease *See* Fabry disease.

androdioecy a sexual dimorphism in plants having bisexual and separate male individuals.

androecious referring to plants having only male flowers.

androecium the aggregate of the stamens in a flower.

androgen any compound with male sex hormone activity. In mammals, the most active androgens are synthesized by the interstitial cells of the testis. *See* testosterone.

androgenesis **1.** development from a fertilized egg followed by disintegration of the maternal nucleus prior to syngamy. The resulting individual possesses only paternal chromosomes and is haploid. **2.** production of an embryo having a diploid set of paternal chromosomes by nuclear transfer (*q.v.*). *Compare with* gynogenesis.

androgenic gland a gland found in most crustaceans belonging to the subclass Malacostraca. When implanted into maturing females, the gland brings about masculinization of primary and secondary sex characters.

androgen insensitivity syndrome a condition in which XY individuals develop as normal-appearing, but sterile, females. Spermatogenesis does not occur in the testes, which are generally located inside the abdomen. Also called *testicular feminization*. *See* Appendix C, 1988, Brown *et al.;* androgen receptor, androgen receptor gene.

androgenote a cell or embryo produced by androgenesis. *Compare with* gynogenote.

androgen receptor (AR) a protein belonging to a subfamily of steroid hormone receptors within a larger family of DNA-binding proteins. The human AR is made up of 919 amino acids and is subdivided into three domains. The N-terminal domain has a regulatory function, and the C-terminal domain binds dihydrotestosterone. The central domain, which binds to DNA, contains zinc fingers. The receptor binds to DNA as a homodimer. *See* androgen, testosterone, vitamin D receptor, zinc finger protein.

androgen receptor gene a gene symbolized *AR* that is located on the X chromosome at q12. It contains eight exons and is 180,245 bp long. *AR* encodes the androgen receptor (*q.v.*). Homologous genes occur in the rat and mouse. Mutations in critical portions of *AR* cause the loss of receptor activity, and this results in abnormalities in sexual phenotype referred to as androgen insensitivity syndrome (*q.v.*) or testicular feminization. See Appendix C, 1988, Brown *et al.*

androgynous **1.** being neither distinguishably masculine nor feminine in appearance or behavior. **2.** bearing staminate and pistillate flowers on distinct parts of the same inflorescence. *See* flower.

andromonecy a sexual condition in which plants develop both staminate flowers (that do not develop fruit) and hermaphroditic flowers.

androphages "male-specific" bacteriophages that absorb on the surface of F pili. Examples are MS2, R17, and Qβ (*all of which see*). *See* F factor (fertility factor).

anemia a disorder characterized by a decrease in hemoglobin per unit volume of blood. In the case of *hemolytic* anemia, there is a destruction of red blood cells. In the case of *hypochromic* anemia, there is a reduction in the hemoglobin content of the erythrocyte.

anemophily pollination by the wind.

anergy the lack of an expected immune response.

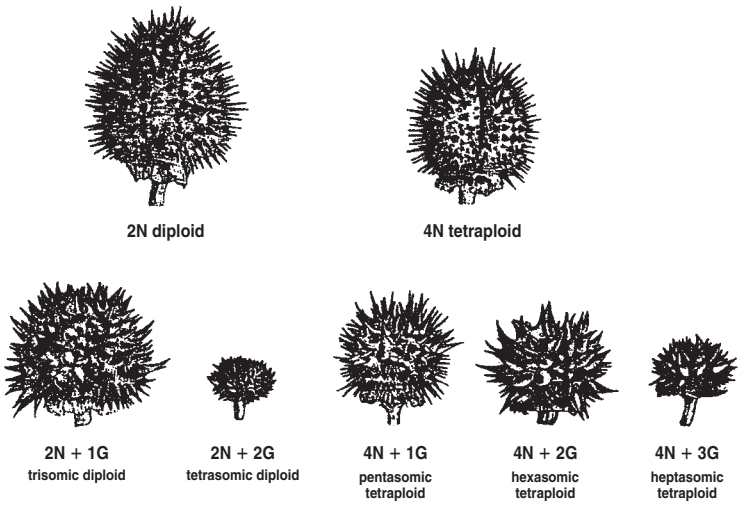
aneucentric referring to an aberration generating a chromosome with more than one centromere.

aneuploidy the condition in which the chromosome number of the cells of an individual is not an exact multiple of the typical haploid set for that species. The nomenclature employs the suffix *somic,* as the following examples illustrate. Down syndrome (*q.v.*) and Turner syndrome (*q.v.*) are examples of a human trisomic and monosomic, respectively. Nullisomics result from the loss (2N – 2) and tetrasomics from the gain (2N + 2) of a chromosome pair. If more than one different chromosome is lost or gained, the condition is described as doubly monosomic (2N – 1 – 1) or doubly trisomic (2N + 1 + 1). Early studies of aneuploids led to the conclusion that genes carried by specific chromosomes controlled morphological traits. For example, in *Datura stramonium* (*q.v.*) extra doses of chromosome G broaden and reduce the seed capsule and increase the size of the spines (see page 26). *See* Appendix C, 1934, Blakeslee; hyperploid, hypoploid, symbols used in human cytogenetics, polyploid.

aneurin vitamin B_1; more commonly known as *thiamine*.

aneusomy the condition in which an organism is made up of cells that contain different numbers of chromosomes. Aneusomy is widespread in flowering plants possessing B chromosomes (*q.v.*). In animals, the term generally refers to a diploid organism with subpopulations of aneuploid, somatic cells. The term aneusomy has been misused in the recent literature of human cytogenetics to refer to a genetic imbalance within a chromosome pair. For example, an individual heterozygous for a deficiency including one or more genes is hemizygous for those genes on the normal homolog. To call such an individual a *segmental aneusomic* is confusing, since aneusomy traditionally implies mosaicism. *See* aneuploidy.

Angelman syndrome (AS) children with this condition are hyperactive and are unable to develop normal speech. Because they show impaired motor control and tend to laugh excessively, the condition is sometimes called "happy puppet syndrome." The British pediatrician Harry Angelman gave the first description of children with the disease in 1965. Later the condition was found to be the result of a deletion in the long arm of chromosome 15. *See* Prader-Willi syndrome (PWS).



Aneuploidy in the Jimson weed, *Datura stramonium.* Extra doses of G chromosomes make the seed capsules smaller and broader and their spines larger.

angiosperm a flowering plant. Any species in the Superclass Angiospermae (*see* Appendix A, Kingdom Plantae) characterized by having seeds enclosed in an ovary. Almost all agriculturally important plants (apart from conifers) belong to the Angiospermae.

Angstrom unit a unit of length equal to one ten-thousandth of a micron (10^{-4} micron; a micron being 10^{-6} meter); convenient for describing atomic dimensions; also equivalent to 10^{-1} nanometers (nm) or 10^{-10} meter. Abbreviated A, A°, Å, Å.U., or A.U. Named in honor of the Swedish physicist Anders Jonas Ångstrom.

Animalia the kingdom containing animals (heterotrophic organisms developing from a blastula). *See* Appendix A, Kingdom 4; opisthokonta.

animal pole that pole of an egg which contains the most cytoplasm and the least yolk.

anion a negatively charged ion. *Contrast with* cation.

Aniridia a dominant mutation of the *Pax-6* gene located at 11p13, which causes defects in the iris, lens, cornea, and retina of humans. The *Pax-6* gene of humans, the *Sey* gene of mice and rats, and the *ey* gene of *Drosophila* are homologous. *See* eyeless.

anisogamy that mode of sexual reproduction in which one of the sex cells, the egg, is large and nonmobile, whereas the other (the sperm) is small and motile. In most anisogamous eukaryotes, the centriole is paternally inherited. *See* isogamy, parthenogenesis.

anisotropy a directional property of crystals and fibers having a high degree of molecular orientation. Anisotropic substances have different physical properties when tested in different directions. When a ray of plane polarized light passes through anisotropic material, it is split into two rays polarized in mutually perpendicular planes. This property of anisotropic material is called *birefringence.* Muscle fibers and the metaphase spindle are examples of living materials exhibiting birefringence. Materials showing no birefringence are said to be *isotropic. See* polarization microscope.

ankylosing spondylitis an arthritic disease resulting in a stiffening and bending of the spine; inherited as an autosomal dominant with reduced penetrance. Over 90% of patients with this disease carry the B27 HLA antigen. *See* histocompatibility.

ankyrin a protein that binds to β-spectrin as well as to the cytoplasmic domains of a variety of integral membrane proteins, and that is thought to interconnect the spectrin-based membrane cytoskeleton and the overlying lipid bilayer. Based on sequence similarity, ankyrins have been identified in various tissues and cell types from a variety of organisms. These proteins generally have three structural domains: a conserved, N-terminal region containing membrane-binding properties, a highly conserved

spectrin-binding region, and a variable, regulatory C-terminal domain. In humans, three forms of ankyrin have been characterized. Ankyrin-1 (also called ankyrin-R) is encoded by the *ANK1* gene at chromosomal map position 8p11 and expressed in erythrocytes and the brain. Ankyrin-2 (also called ankyrin-B) is encoded by the *ANK2* gene at position 4q25-q27 and expressed primarily in the brain. A third protein, ankyrin-3 (also known as ankyrin-G), is encoded by the *ANK3* gene mapping to 10q21, and alternatively spliced isoforms are expressed in nervous tissue, muscle, and other tissues. Mutations in the erythrocytic ankyrin gene, *ANK1*, are associated with hereditary spherocytosis (*q.v.*). *See* fusome, integral protein, spectrin, spectrosome.

anlage the embryonic primordium from which a specific part of the organism develops.

anneal to subject first to heating then to cooling. In molecular genetics experiments, annealing is used to produce hybrid nucleic acid molecules containing paired strands, each from a different source. Heating results in the separation of the individual strands of any double-stranded, nucleic-acid helix, and cooling leads to the pairing of any molecules that have segments with complementary base pairs.

annidation the phenomenon where a mutant is maintained in a population because it can flourish in an available ecological niche that the parent organisms cannot utilize. A wingless mutant insect, for example, might be poorly adapted in its ancestral habitat but able to live in tunnels and crevices that a winged form could not occupy.

annotation *See* genome annotation.

annulate lamellae paired membranes arranged in stacks and possessing annuli resembling those of the nuclear membrane. Annulate lamellae may serve to transfer nuclear material to the cytoplasm by the replication of the nuclear envelope and may be a mechanism for storing gene-derived information to be used for cytoplasmic differentiation during early embryogenesis. During insect oogenesis, annulate lamellae occur alongside nurse cell nuclei and germinal vesicles, and they are abundant in the ooplasm.

annulus a ring. Applied to any of a number of ring-shaped parts of animals and plants. Used in cytology to refer to the ring-shaped nuclear pores.

anode the positive electrode; the electrode to which negative ions are attracted. *Contrast with* cathode.

anodontia the congenital absence of teeth. *Hypodontia* is currently the preferred term.

anonymous DNA a segment of DNA of unknown gene content that has been localized to a specific chromosome.

Anopheles a genus containing about 150 species of mosquitoes, many of which are of medical importance. Africa's principal malaria vector is *A. gambiae*. Other vector species are *A. funestus*, *A. quadrimaculatus*, *A. atroparvus*, *A. nili*, *A. moucheti*, and *A. pharoensis*. Polytene chromosomes occur in both larval salivary gland cells and adult ovarian nurse cells. Sibling species can often be separated by differences in the banding patterns of their polytene chromosomes. The genome of *A. gambiae* has been shown to contain 278 mbp of DNA and about 13,700 genes. *See* Appendix A, Arthropoda, Insecta, Diptera; Appendix C, 1898, Ross; 1899, Grassi; 2002, Holt *et al.*; Appendix E; intron dynamics, malaria, mariner elements, Minos element, shotgun sequencing.

anosmia the absence or loss of the sense of smell. Anosmia may be caused by injury to or loss of olfactory receptor neurons (*q.v.*) or by injury to region(s) in the brain or elsewhere where olfactory signals are processed. Anosmia may also result due to defects in any element of the olfaction signaling pathway. For example, mice and nematodes with defects in G proteins (*q.v.*) found in olfactory receptor neurons exhibit olfactory and chemosensory defects, respectively.

Anser anser the Gray Lag goose, a favorite experimental organism for students of animal behavior and its hereditary components.

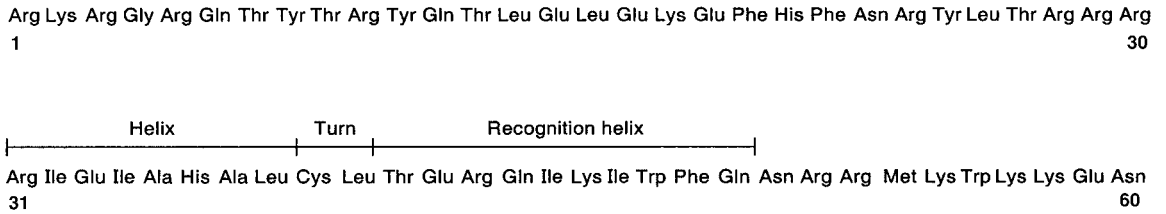
antagonist a molecule that bears sufficient structural similarity to a second molecule to compete with that molecule for binding sites on a third molecule. *See* competition.

antagonistic pleiotropy a phenomenon in which alleles (that are detrimental late in life) improve fitness earlier in life.

antenatal before birth; during pregnancy.

antennae the first paired appendages on the head of arthropods.

Antennapedia a gene residing at 47.9 on the genetic map and within segment 84B of the salivary map of *Drosophila melanogaster*. The *Antp* gene is one of a cluster of three genes that specify the type of differentiation that cells in the segments from the



Antennapedia homeobox

head to the anterior portion of the second thoracic segment will undergo. Mutations in the *Antp* gene cause the transformation of the segment that normally produces the antenna into one that produces a middle leg. The gene encodes a protein characterized by a homeobox (*q.v.*). This is a segment of 60 amino acids that lies close to the C terminus of the *Antp* protein. The amino acid sequence of this segment is shown above. It binds to target DNA sequences by its helix-turn-helix motif (*q.v.*). The complete three-dimensional structures of the *Antp* homeodomain as well as of the homeodomain-target DNA complex have been determined using NMR spectroscopy (*q.v.*) and x-ray crystallography (*q.v.*). *See* Appendix C, 1983, Scott *et al.*; 1989, Qian *et al.*; 1990, Maliki, Schughart, and McGinnis; *bithorax*, homeotic mutants, *Hox* genes, *Polycomb*, *proboscipedia*, segment identity genes.

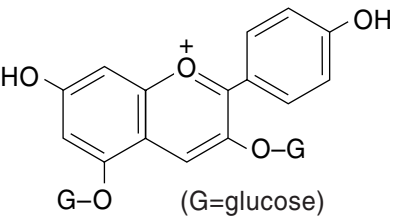
anther the terminal portion of a stamen bearing pollen sacs.

anther culture a technique that utilizes anthers or pollen cells to generate haploid tissue cultures or even plants. *See* Appendix C, 1973, Debergh and Nitsch, haploid sporophytes.

antheridium the male gametangium of algae, fungi, bryophytes, and pteridophytes. *Contrast with* oogonium.

anthesis the time of flowering.

anthocyanins the red, violet, or blue glycosidic pigments that give color to flowers, fruits, seeds, stems, and leaves of plants. The common structural unit is a 15-carbon flavone skeleton to which sugars are attached. An example is pelargonidin, a scarlet pigment produced by geraniums. Unlike the carotenoids and chlorophylls, which are lipid-soluble pigments of plastids, anthocyanins are water-soluble and are found dissolved in the vacuoles (*q.v.*) of plant cells. A primary function of anthocyanins is to attract insect pollinators to plants. *See* kernel, pelargonidin monoglucoside, *R* genes of maize.



Pelargonidin, an anthocyanin

anthrax a disease caused by *Bacillus anthracis*, a spore-forming bacterium. The condition is seen in cows, pigs, goats, horses, and sheep. In humans the condition was first called *wool-sorter's disease*, and it resulted from inhalation of dust that contained spores. The first successful artificially produced vaccine (*q.v.*) was against anthrax. *See* Appendix A, Bacteria, Endospora; Appendix C, 1881, Pasteur.

anthropocentrism also called *anthropomorphism*. **1.** explanation of natural phenomena or processes in terms of human values. **2.** assuming humans to be of central importance in the universe or ultimate end of creation. **3.** ascribing human characteristics to a non-human organism.

anthropoid designating the great apes of the family Pongidae, including the gibbons, orangutans, gorillas, and chimpanzees.

anthropometry the science that deals with the measurements of the human body and its parts.

antiauxin a molecule that competes with an auxin (*q.v.*) for auxin receptor sites. A well-known antiauxin is 2,6-dichlorophenoxyacetic acid.

antibiotic a bacteriocidal or bacteriostatic substance produced by certain microorganisms, especially species of the genera *Penicillium*, *Cephalosporium*, and *Streptomyces*. *See* actinomycin D, ampicillin, chloramphenicol, cyclohexamide, erythromycin, ka-