GENES AND PHENOTYPES

**Gene**: a functional unit of inheritance, usually corresponding to the segment of DNA coding for a single protein.

**Genome**: all of an organism’s DNA sequences.

- **locus**: the site of the gene in the genome
- **alleles**: alternative forms of a gene

**Wild-type**: the normal, naturally occurring type

**Mutant**: differing from the wild-type because of a genetic change (a mutation)

**GENOTYPE**: the specific set of alleles forming the genome of an individual

**PHENOTYPE**: the visible character of the individual

- allele A is dominant (relative to a); allele a is recessive (relative to A)

In the example above, the phenotype of the heterozygote is the same as that of one of the homozygotes; in cases where it is different from both, the two alleles are said to be co-dominant.

CHROMOSOMES

A normal diploid chromosome set, as seen in a metaphase spread, prepared by bursting open a cell at metaphase and staining the scattered chromosomes. In the example shown schematically here, there are three pairs of autosomes (chromosomes inherited symmetrically from both parents, regardless of sex) and two sex chromosomes—an X from the mother and a Y from the father. The numbers and types of sex chromosomes and their role in sex determination are variable from one class of organisms to another, as is the number of pairs of autosomes.

THE HAPLOID–DIPLOID CYCLE OF SEXUAL REPRODUCTION

The greater the distance between two loci on a single chromosome, the greater is the chance that they will be separated by crossing over occurring at a site between them. If two genes are thus reassorted in x% of gametes, they are said to be separated on a chromosome by a genetic map distance of x map units (or x centimorgans).

MEIOSIS AND GENETIC RECOMBINATION

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TYPES OF MUTATIONS

POINT MUTATION: maps to a single site in the genome, corresponding to a single nucleotide pair or a very small part of a single gene.

lethal mutation: causes the developing organism to die prematurely.
conditional mutation: produces its phenotypic effect only under certain conditions, called the restrictive conditions. Under other conditions—the permissive conditions—the effect is not seen. For a temperature-sensitive mutation, the restrictive condition typically is high temperature, while the permissive condition is low temperature.
loss-of-function mutation: either reduces or abolishes the activity of the gene. These are the most common class of mutations. Loss-of-function mutations are usually recessive—the organism can usually function normally as long as it retains at least one normal copy of the affected gene.
null mutation: a loss-of-function mutation that completely abolishes the activity of the gene.

gain-of-function mutation: increases the activity of the gene or makes it active in inappropriate circumstances; these mutations are usually dominant.
dominant-negative mutation: dominant-acting mutation that blocks gene activity, causing a loss-of-function phenotype even in the presence of a normal copy of the gene. This phenomenon occurs when the mutant gene product interferes with the function of the normal gene product.
suppressor mutation: suppresses the phenotypic effect of another mutation, so that the double mutant seems normal. An intragenic suppressor mutation lies within the gene affected by the first mutation; an extragenic suppressor mutation lies in a second gene—often one whose product interacts directly with the product of the first.

INVERSION: inverts a segment of a chromosome

DELETION: deletes a segment of a chromosome

TRANSLOCATION: breaks off a segment from one chromosome and attaches it to another

TWO GENES OR ONE?

Given two mutations that produce the same phenotype, how can we tell whether they are mutations in the same gene? If the mutations are recessive (as they most often are), the answer can be found by a complementation test.

In the simplest type of complementation test, an individual who is homozygous for one mutation is mated with an individual who is homozygous for the other. The phenotype of the offspring gives the answer to the question.

COMPLEMENTATION: MUTATIONS IN TWO DIFFERENT GENES

homozygous mutant mother

homozygous mutant father

hybrid offspring shows normal phenotype: one normal copy of each gene is present

NONCOMPLEMENTATION: TWO INDEPENDENT MUTATIONS IN THE SAME GENE

homozygous mutant mother

homozygous mutant father

hybrid offspring shows mutant phenotype: no normal copies of the mutated gene are present