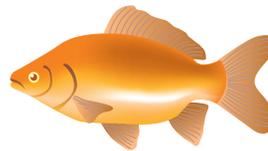## GENES AND PHENOTYPES

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

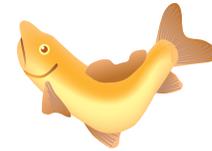**Genome:** all of an organism's DNA sequences.

locus: the site of the gene in the genome

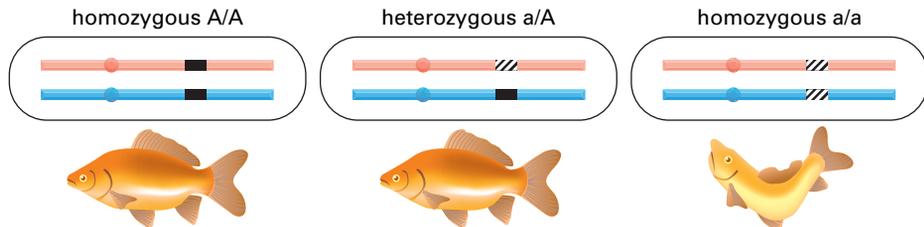**alleles:** alternative forms of a gene

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

homozygous A/A   heterozygous a/A   homozygous a/a

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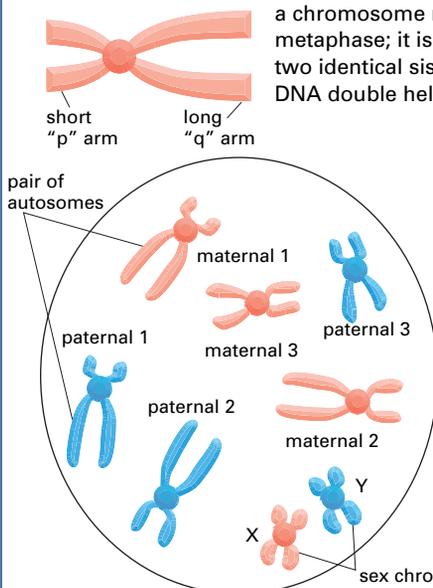
In the example above, the phenotype of the heterozygote is the same as that of one of the homozygotes; in cases where it is different from both, the two alleles are said to be co-dominant.

CHROMOSOMES

a chromosome at the beginning of the cell cycle, in G_1 phase; the single long bar represents one long double helix of DNA

centromere

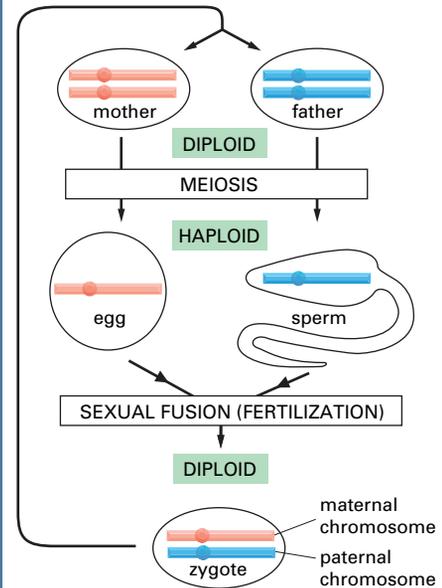short "p" arm   long "q" arm

a chromosome near the end of the cell cycle, in metaphase; it is duplicated and condensed, consisting of two identical sister chromatids (each containing one DNA double helix) joined at the centromere.

short "p" arm   long "q" arm

pair of autosomes

maternal 1

paternal 3

paternal 1   maternal 3

paternal 2

maternal 2

Y

X

sex 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.
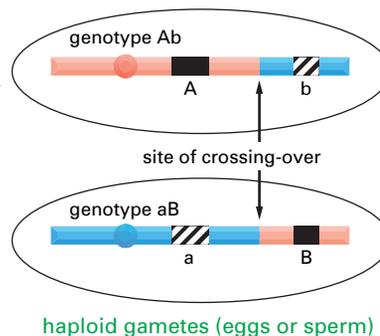
THE HAPLOID–DIPLOID CYCLE OF SEXUAL REPRODUCTION

mother   father

DIPLOID

MEIOSIS

HAPLOID

egg   sperm

SEXUAL FUSION (FERTILIZATION)

DIPLOID

maternal chromosome

paternal chromosome

zygote

For simplicity, the cycle is shown for only one chromosome/chromosome pair.

MEIOSIS AND GENETIC RECOMBINATION

maternal chromosome

A   B

paternal chromosome

a   b

diploid germ cell

genotype $\dfrac{AB}{ab}$
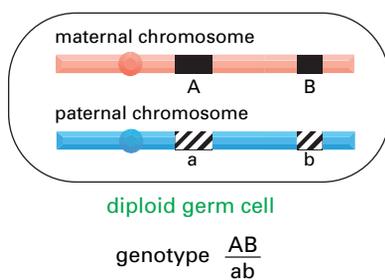
MEIOSIS AND RECOMBINATION

genotype Ab

A   b

site of crossing-over

genotype aB

a   B

haploid gametes (eggs or sperm)

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).

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
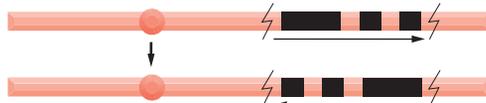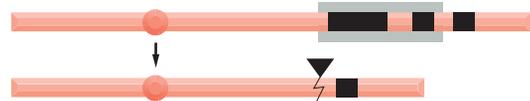

INVERSION: inverts a segment of a chromosome

**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.

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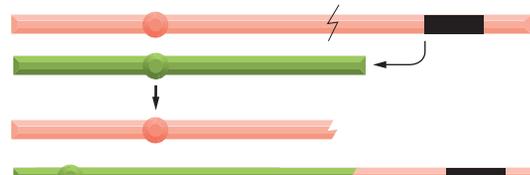
TRANSLOCATION: breaks off a segment from one chromosome and attaches it to another

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.

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	NONCOMPLEMENTATION: TWO INDEPENDENT MUTATIONS IN THE SAME GENE
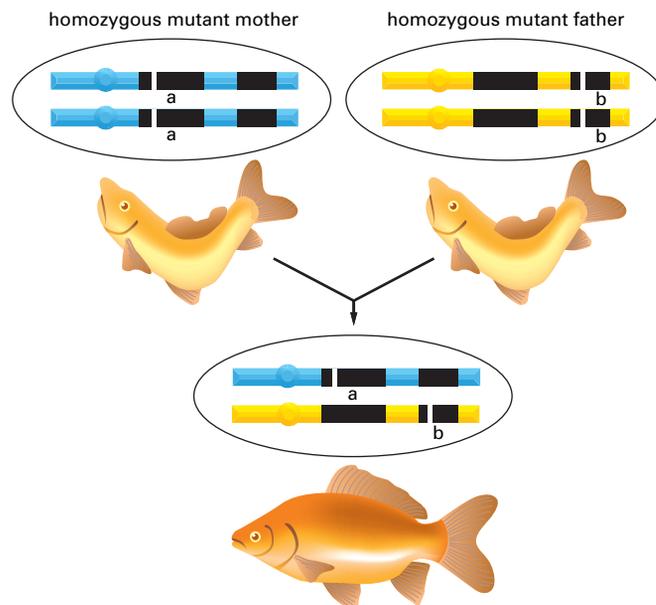
hybrid offspring shows normal phenotype: one normal copy of each gene is present
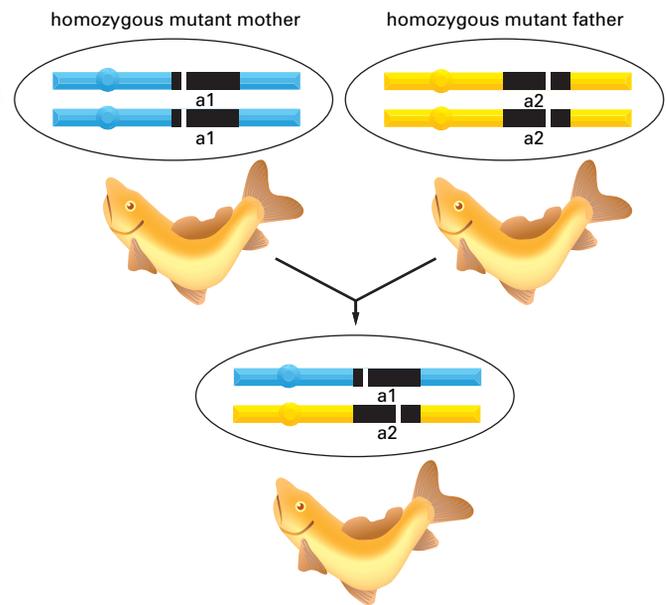
hybrid offspring shows mutant phenotype: no normal copies of the mutated gene are present