Definitions and concepts of some terms

(1) Traits: A feature or character that results from gene action or interaction (with another gene or environment), and transmitted from one generation to another

[Talk about Central Dogma of Molecular Biology]

(2) Genotype: Specific combination of genes or alleles (alternate forms of a gene) that express and function for a particular trait or a set of traits of a cell or organism.

(3) Phenotype: An observable or detectable (visible or non-visible) and measurable aspect of a trait or set of traits of a cell or an organism, and the trait or traits are expressed or produced by the interaction between genotype and the environment (internal and external).

For example, **Color** of a flower is a trait; **Red** & **Purple** are phenotypes of the flower

(2) & (3) Genotype & Phenotype:

Contd..

In ideal sense,

- **genotype** means complete set of genes that are inherited by the individuals from the parents and
- **phenotype** means observable and measurable aspects of all traits i.e., morphology, physiology and behavior of that organism.

- Genotype has the potential to give rise to a set or range or possible combination of phenotypes, but the developmental events and the environmental conditions determine what actually the phenotype ends up.

- Sometimes the environment plays a major role and in other cases environmental influence is minor or even non-existent. In this sense, two individuals may have the identical genotype, but not the same phenotype as there are always differences (slight to large extent) in morphology, physiology and behavior between two members of the same species.

- Also we need to keep in mind that (except for asexual reproduction), any two individual progeny of the same species differ at least a little in genotype as there is naturally occurring crossing over of the segments between homologous chromosomes followed by random segregation of chromosomes before gamete formation and random fusion of (male and female) gametes to form zygote in sexual reproduction.

- However, in practice we use these two terms in partial or restricted sense.

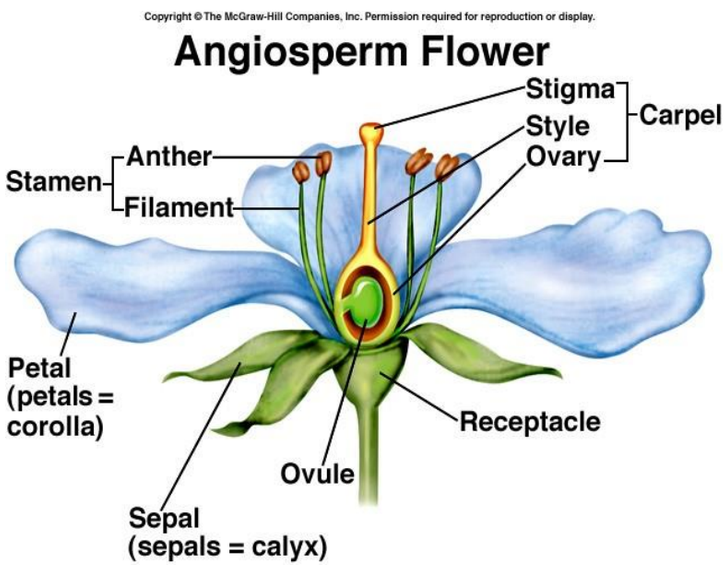
Definitions and concepts of some terms

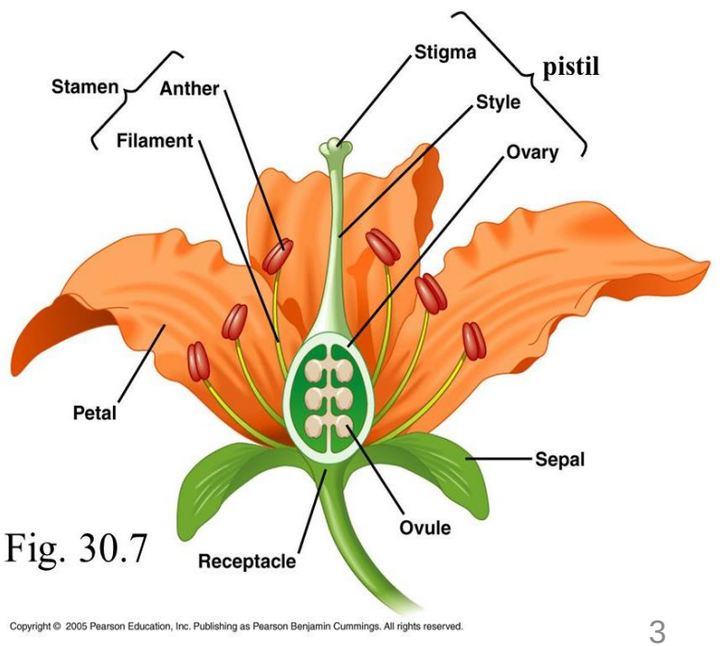
Contd..

(4) True-breeding or pure line: A strain /variety or cultivar /stock in which mating of individuals yields progeny with the same genotype and phenotype as the parents

[Show & dissect 2/3 flowers in the class, discuss about its parts]

Flower parts: 1) Sepals/Calyx, 2) Petals/Corolla, 3) Carpel/Pistil (Stigma, Style and Ovary), 4) Stamen (Anther, Filaments)





Definitions and concepts of some terms

Contd..

(5) Self-fertilization or self-pollination (Autogamy):

- During sexual reproduction, when the two (male and female) gametes come from the same individual (bisexual organism) is called self-fertilization.
- When pollination (transfer of pollen into stigma) occurs within the flowers of the same plant it is called self-pollination.
- For example, rice, wheat, tomato, potato
- Many self-pollinating plants have certain %-age of cross-pollination

Definitions and concepts of some terms

Contd..

(5) Self-fertilization or self-pollination (Autogamy) (contd..):

- Hermaphrodite animals (having both male and female gonads in the same individuals) are mostly cross-fertilized due to their adaptation.
- Hermaphrodite flower has both male organ (stamen) and female organ (carpel/pistil); e.g., many common garden plants.
- A monoecious plant (bisexual organism) has both male and female reproductive organs in the same individual (either bearing the hermaphrodite flowers or bearing both the male and female flowers)
- A dioecious plant has either male or female flowers, not both; meaning the male and female organs are in separate individuals

Definitions and concepts of some terms

Contd..

(6) Cross-fertilization or cross-pollination (Allogamy):

- During sexual reproduction, when the two (male and female) gametes come from the different individuals of the same species is called cross-fertilization.
- When pollination (transfer of pollen into stigma) occurs between two different plants of the same species it is called cross-pollination.
- For example, pumpkin, grape, maize [although maize has both male and female flowers in the same plant, normally it is cross-pollinated but 5-10% self-pollination can occur in field condition]

Definitions and concepts of some terms

Contd..

(7) Crossing or hybridization:

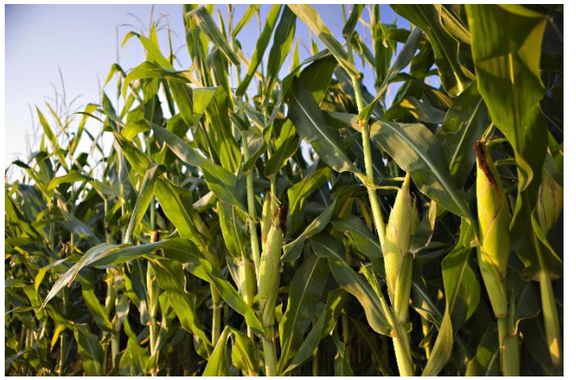
- Deliberate mating of two selected parental types of organisms in genetic analysis, i.e. fusion of male gamete from one selected individual and female gamete from another selected one.
- The resultant progeny or offspring is known as **hybrid** (or also known as crossbred in animal) which has important role in basic and applied genetic research

Examples of hybrid flowers from hybrid swarms of *Aquilegia pubescens* and *Aquilegia formosa*

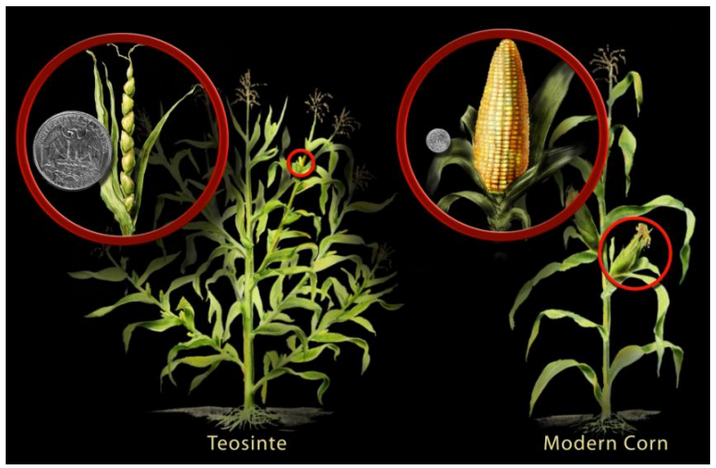
https://en.wikipedia.org/wiki/Hybrid_(biology)

Examples of hybrid food materials









Credit: Nicolle Rager Fuller, National Science Foundation

The food we eat comes from plants already extensively genetically modified from their original form through hybridization.

Definitions and concepts of some terms

Contd..

(8) Haploid (N): A cell having one chromosome set (or basic genomic set), i.e. one copy of each chromosome in the nucleus or an individual organism composed of such cells.

(9) Diploid (2N): A cell having two sets of chromosomes, i.e. two copies of each chromosome in the nucleus or an individual organism composed of such cells.

Thus, **haploid or diploid can be nucleus or cell or organism**

Haploid or diploid phases alternate in sexual life cycle of higher eukaryotes

(= meiosis and fertilization alternate)

Figure 13.5 The human life cycle. In each generation, the number of chromosome sets is halved during meiosis but doubles at fertilization. For humans, the number of chromosomes in a haploid cell is 23, consisting of one set ($n = 23$); the number of chromosomes in the diploid zygote and all somatic cells arising from it is 46, consisting of two sets ($2n = 46$).

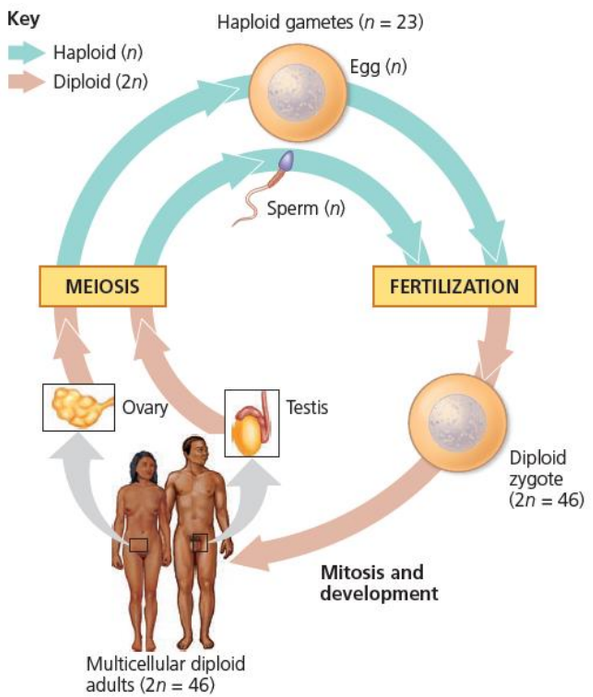
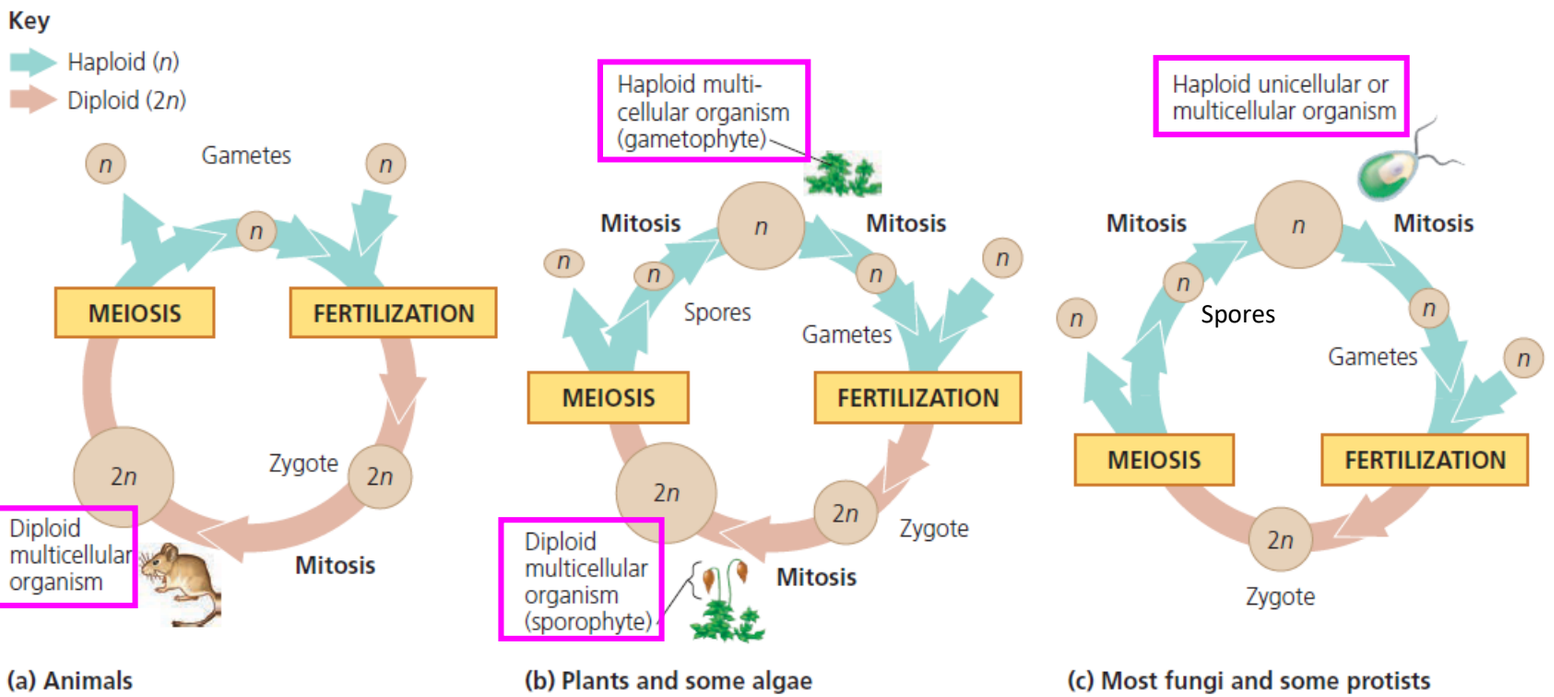


Figure 13.6 Three types of sexual life cycles. The common feature of all three cycles is the alternation of meiosis and fertilization, key events that contribute to genetic variation among offspring. The cycles differ in the timing of these two key events. (Small circles are cells; large circles are organisms.)



Biology: A Global Approach by Campbell et al. (11th Ed.)

Up to this slide, Lecture # 3 delivered on 22.08.2023

Definitions and concepts of some terms

Contd..

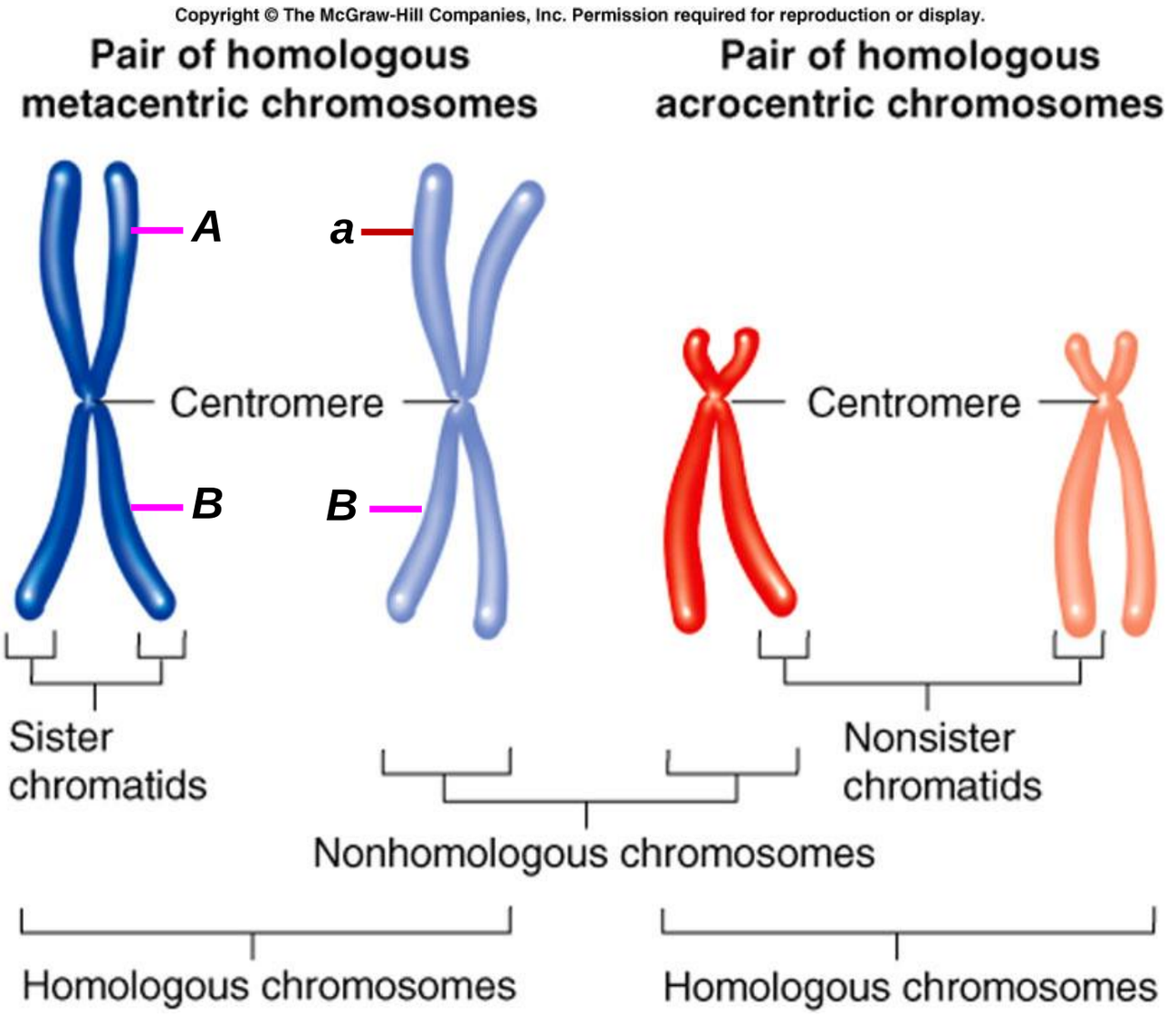
(10) Locus (pl. loci**)**: The specific place or position in the chromosome, wherein a **particular gene** (**Mendel's particulate factor**) is located or its alternate alleles are located in the homologous chromosomes. Also refers to the position of a gene on the genetic map.

(11) Allele: One of the two or more alternate forms of a gene existing at a particular locus in the genome of an organism (e.g., pea plant), and alleles are responsible for different phenotypes (e.g., yellow, green) of the same hereditary trait (e.g., seed color).

Alleles are located on homologous chromosomes

Alleles of "***A***" gene can be also:
A1, A2, A3

"B" gene has no allele in this individual (but may be present in other individual) or in the population



Discuss blood group alleles. Ask the students about their blood group.

Definitions and concepts of some terms

Contd..

(12) DNA polymorphism leads to origin of alleles:

Alleles arise due to DNA polymorphism (i.e., alteration in nucleotide of the DNA sequence) in the genome, and DNA polymorphism may be essentially of **four types**:

i) single nucleotide polymorphism (**SNP**),

ii) **microsatellite** or simple sequence repeat (**SSR**) or short tandem repeat (**STR**) of 2-10 nucleotide units, repeated 5-50 times,

iii) **minisatellite** or simple sequence length polymorphism (**SSLP**) or variable number of tandem repeat (**VNTR**) of 11-100 nucleotide units, repeated 5-50 times and

iv) **indel** (short stretches of insertion and deletion in the DNA sequence)

We can have **alleles of genes and alleles of non-genes**:

In a population, alleles of DNA polymorphic markers can be found anywhere in the genome not necessarily as a part of gene which usually encodes for functional products (protein/RNA) for a phenotype.

Definitions and concepts of some terms

Contd..

(13) Concepts of alleles (gene and non-gene based DNA polymorphic markers) have several practical utilities:

a) Mapping of loci on chromosomes (distance between genes, between non-gene markers or between a gene and a non-gene marker)

b) Pedigree or progeny analysis in breeding in animals or plants, respectively

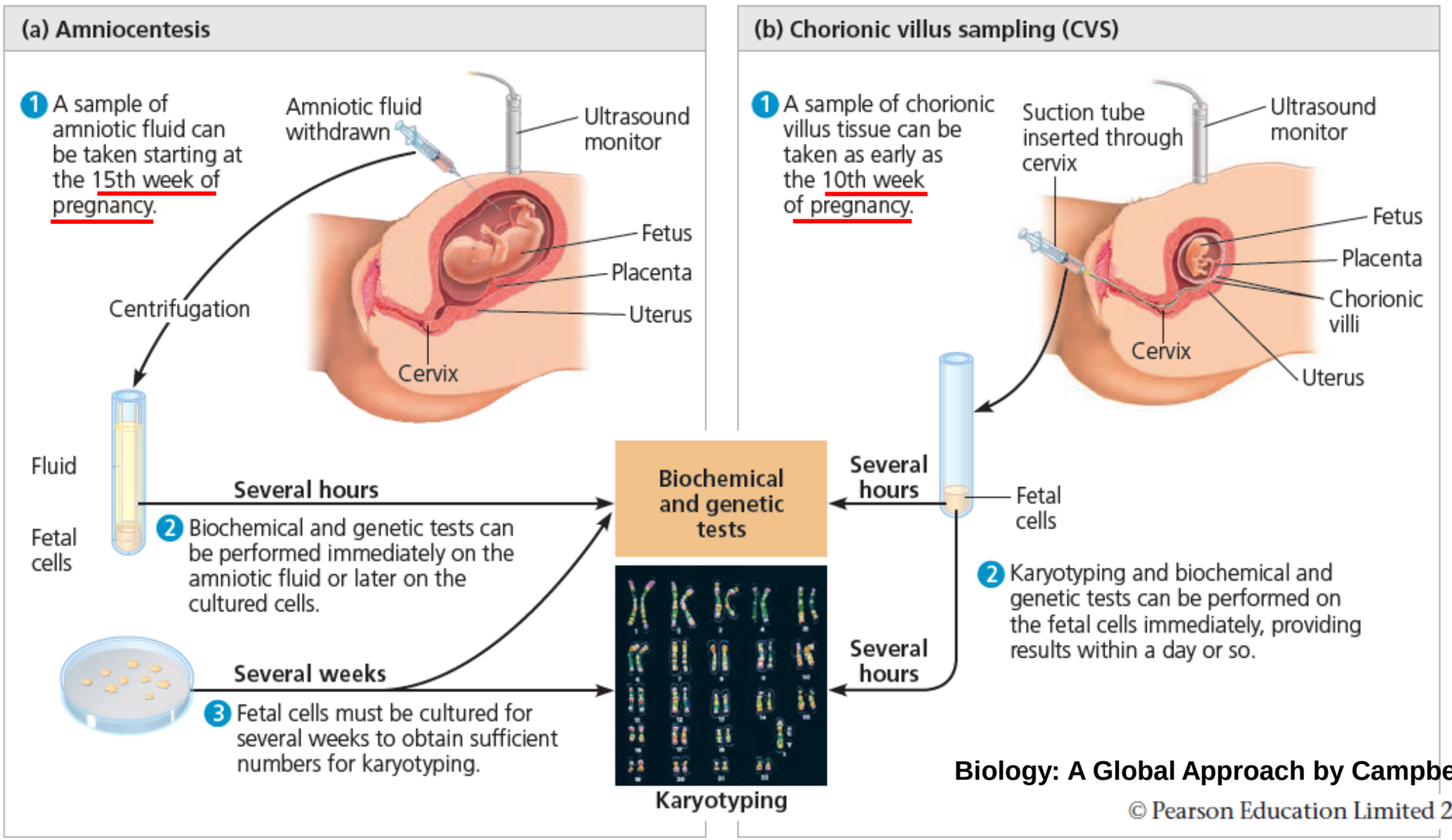
c) DNA fingerprinting or typing in parental dispute for paternity determination

d) Forensic analysis in crime scene investigation to find out the suspect

e) Disease diagnosis at an early life stage (fetus or new born)

Diagnosis of genetic disease at an early life stage (fetus or new born)

Figure 14.19 Testing a fetus for genetic disorders. Biochemical tests may detect substances associated with particular disorders, and genetic testing can detect many genetic abnormalities. Karyotyping shows whether the chromosomes of the fetus are normal in number and appearance.



Biology: A Global Approach by Campbell et al. (11th Ed.)

Definitions and concepts of some terms

Contd..

(13) Concepts of alleles (gene and non-gene based DNA polymorphic markers) have several practical utilities (contd..):

f) In evolutionary genetics, phylogenetic tree construction

g) In population genetics, to establish variability among ethnic groups or population

h) Conservation biology: studies of endangered species to determine gene variability

i) Detection of contamination in a sample of mixed seeds or pathogenic microbes in food samples. [Bacteria do not have typical alleles as they do not have homologous chromosomes, but fungi have. However, conceptually alternate forms of genes or alleles are there in all microbial populations/communities]

Definitions and concepts of some terms

Contd..

(13) Concepts of alleles (gene and non-gene based DNA polymorphic markers) have several practical utilities (contd..):

Since, the alleles of non-genes usually do NOT encode for any functional product to give phenotype, they are codominant; unlike the alleles of genes which usually show dominance or recessiveness or codominance.

Thus, Genotype/gene (e.g., ***A***) > Traits/trait

Allele (e.g., ***a*** or A_1, A_2 etc.) > Phenotype

[When we know or do not know dominant-recessive status]

Definitions and concepts of some terms

Contd..

(14) Wild type: Wild type may be defined as the cell or individual having an allele or phenotype or function that can be designated **as normal or standard or natural**, because it is usually (but not always) **most prevalent in the natural population** of that species.

(15) Mutant: Mutant can be defined as the cell or individual having the **altered form** of the wild-type allele or phenotype or function. [wild-type = adjective, wild type = noun]

Mutant allele may function as/ show phenotype -

(i) **Null mutation** > complete loss of function;

(ii) **Leaky mutation** > partial loss of function;

(iii) **Conditional mutation** > wild-type phenotype under certain (permissive) environmental conditions and a mutant phenotype under other (restrictive) conditions]

Definitions and concepts of some terms

Contd..

(16) Homozygous: The state of diploid organism carrying a pair of identical alleles for a gene that determines a particular trait, and producing gametes of identical genotype.

E.g., ***AA***, ***aa*** *[How many types of gametes produced by these organisms?]*

(17) Heterozygous: The state of diploid organism carrying different alleles for a gene that determines particular trait, and producing gametes of different genotypes. E.g., ***Aa***. This is also sometimes called as **hybrid**.

A **monohybrid** is the heterozygous individual having two different alleles for a **single** gene. E.g., ***Aa*** *[How many types of gametes produced by this organism?]*

A **dihybrid** is the heterozygous individual having two pair of different alleles for the corresponding **two** genes. E.g., ***AaBb*** *[How many types of gametes produced by this organism?]*

Definitions and concepts of some terms

Contd..

(18) Dominant allele or phenotype: The parental allele of a gene or phenotype of a trait, which is expressed/ displayed in heterozygous (***Aa***) state as well as in one particular homozygous (***AA***) state. [*Gene is expressed, phenotype is displayed*]

(19) Recessive allele or phenotype: The parental allele of a gene or phenotype of a trait, which is NOT expressed/ displayed in the heterozygous (***Aa***) state but expressed/ displayed only in one particular homozygous (***aa***) state. [*Gene is expressed, phenotype is displayed*]

(20) Co-dominant allele or phenotype: When both the alleles or phenotypes are expressed/ displayed in heterozygous (AB) state. E.g., blood group AB.

Note: for any trait, the observed dominant/recessive relationship of alleles depends on the level (molecular, cellular, organismal) of analysis. We will discuss more about this while studying pleiotropic effect of gene/ allele

Definitions and concepts of some terms

Contd..

(21) Gene symbol normally used in Classical/ Transmission Genetics: There is no uniform system for gene symbols adopted by geneticists, but there are certain rules for gene symbol specific for the model organisms.

d) *Arabidopsis thaliana* (Thale cress, a member of the mustard family)

k) *Pisum sativum* (a garden pea)

l) *Zea mays* (corn)

b) *Drosophila melanogaster* (fruit fly)

e) *Mus musculus* (mouse)

f) *Homo sapiens* (human)

iGenetics

We will use uppercase and lowercase letter for dominant and recessive phenotypic allele, respectively in plant system; e.g. '***R***' for round (dominant) and '***r***' for wrinkle (recessive), shaped pea seeds.

Conventionally, the letter is chosen based on the phenotype of the wild-type/ dominant allele particularly in plant system

+ for wild-type allele (or lowercase letter with superscript **+**) and lowercase letter for mutant recessive allele of Drosophila; e.g., '**+**' or 'w^+' for red colored (dominant) and '***w***' for white colored (recessive) eyes. Certain other rules will be discussed later.

However, in Drosophila system the letter is chosen based on the phenotype of the mutant/recessive allele

Typically, (i) genes/alleles are designated as *italicized symbol*, but proteins are designated as normal symbol.

(ii) chromosome is represented as slash '*/*'. Thus, *R/r* or *+/w* indicates the two alleles are located in a single locus of the two homologous chromosomes.

Up to this slide, Lecture # 4 delivered on 22.08.2023