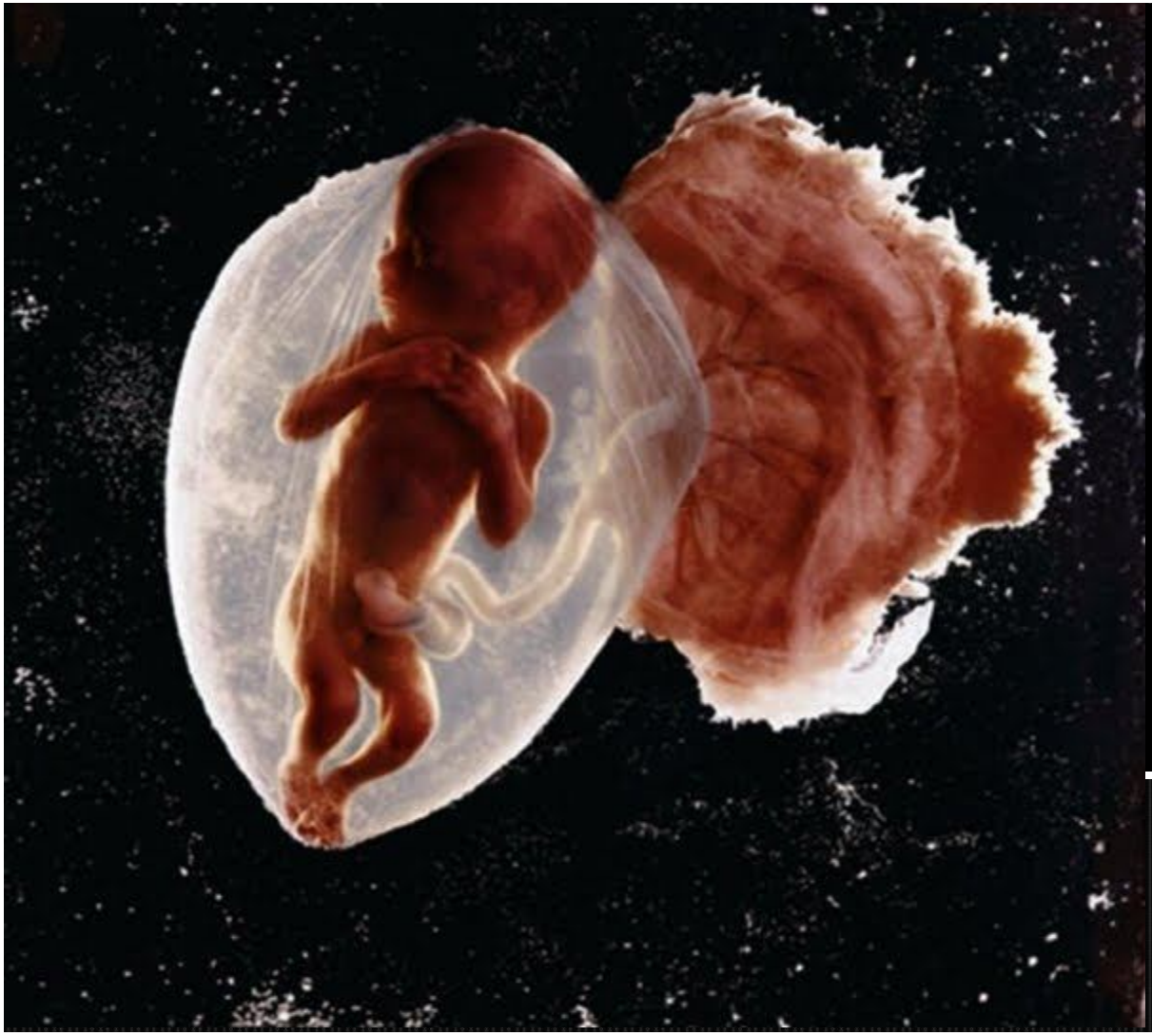


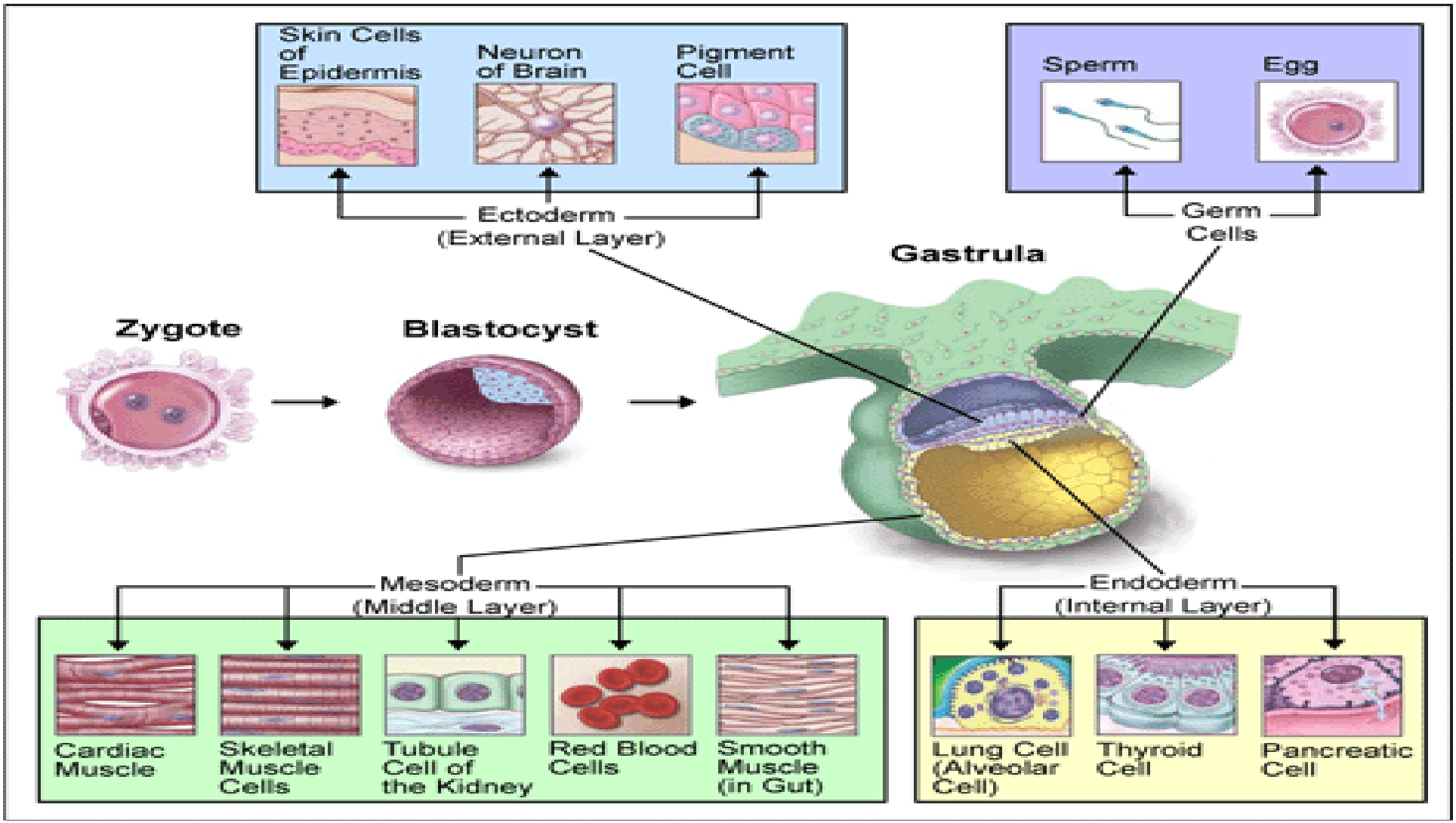
Embryology RESUMED

D.H.MD

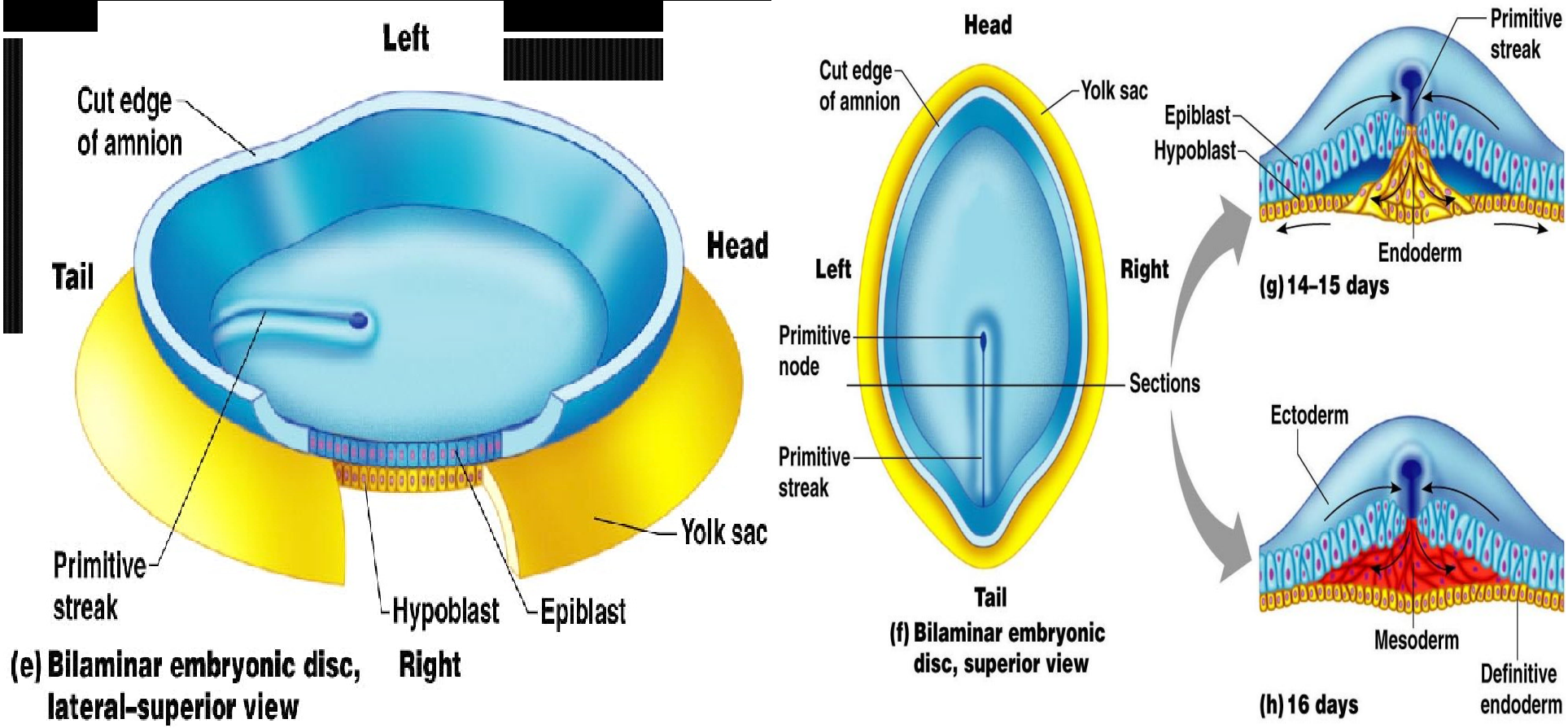

Of hundreds of sperms, only one can fertilize the ovum.

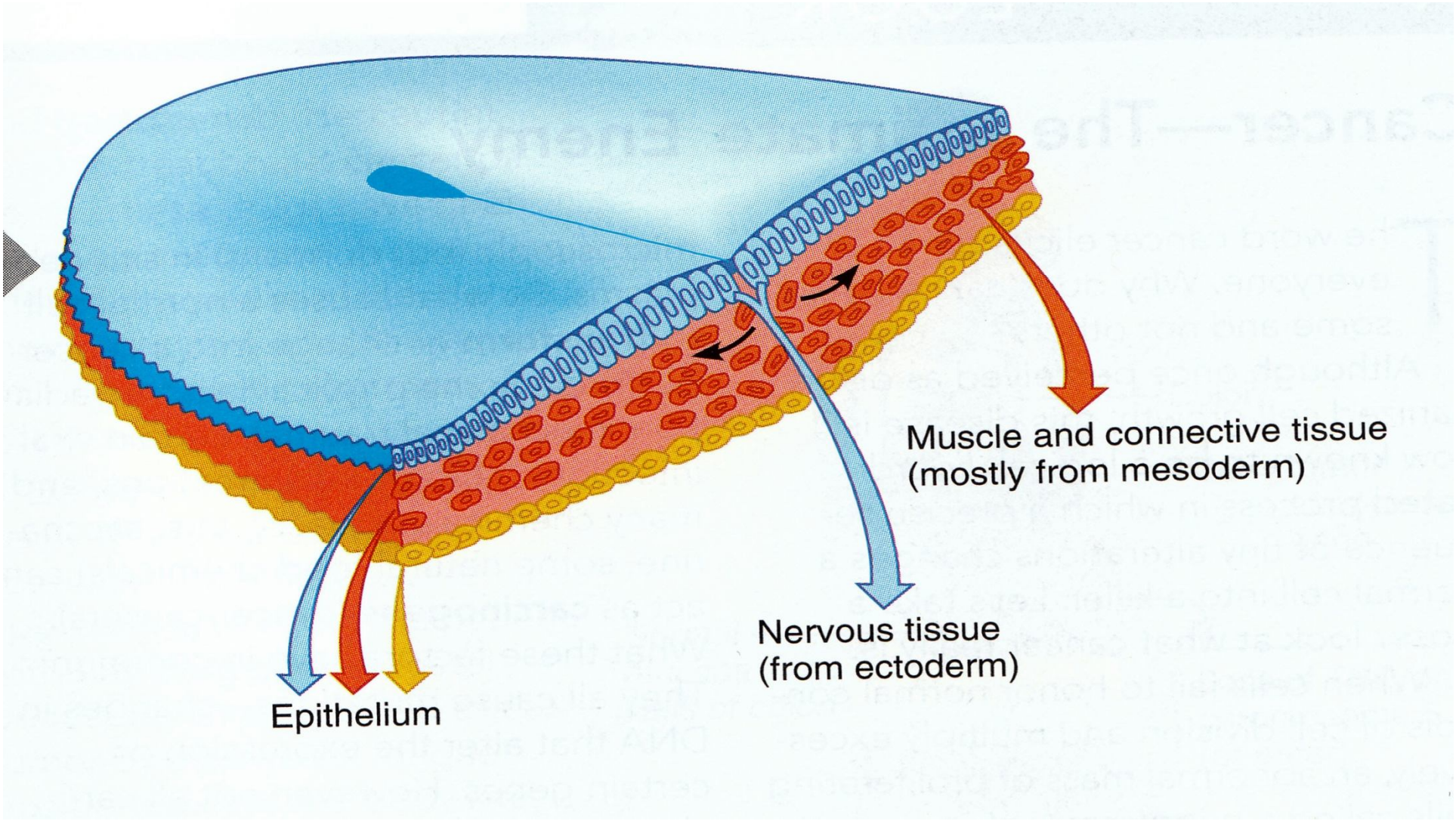
Prenatal period

- **Embryonic period** – first 8 weeks
- **Fetal period** – remaining 30 weeks

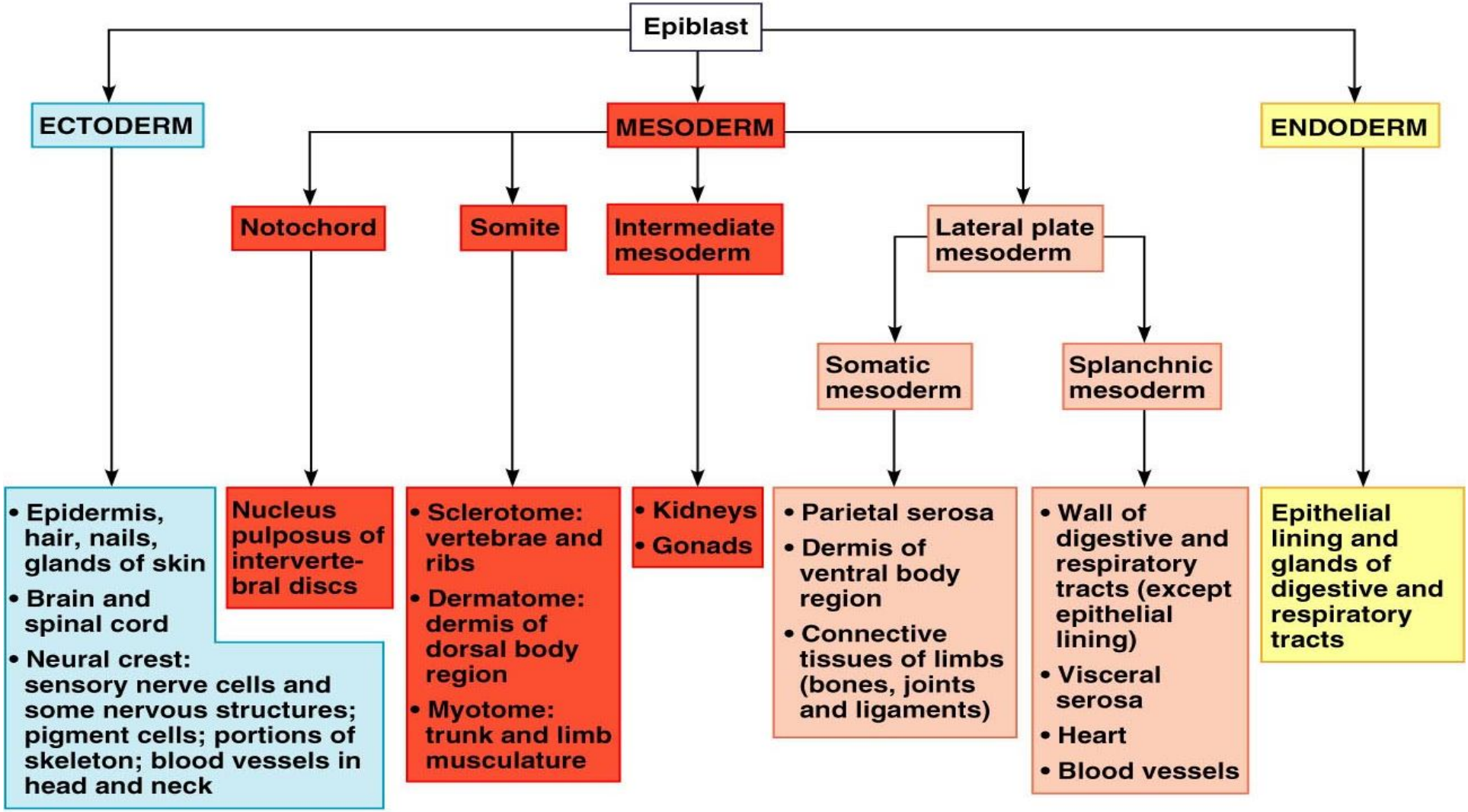

Skin Cells of Epidermis
Neuron of Brain
Pigment Cell
Ectoderm (External Layer)
Sperm
Egg
Germ Cells
Gastrula
Zygote
Blastocyst
Mesoderm (Middle Layer)
Endoderm (Internal Layer)
Cardiac Muscle
Skeletal Muscle Cells
Tubule Cell of the Kidney
Red Blood Cells
Smooth Muscle (in Gut)
Lung Cell (Alveolar Cell)
Thyroid Cell
Pancreatic Cell

The Primitive Streak

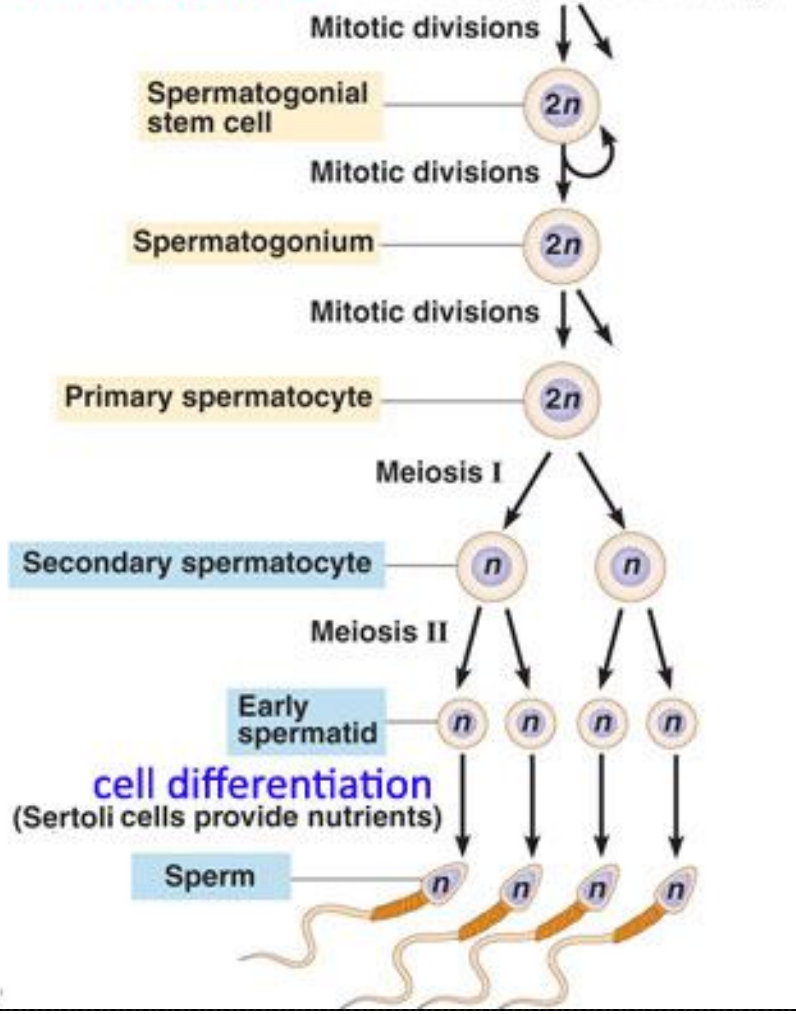



Muscle and connective tissue
(mostly from mesoderm)
Nervous tissue
(from ectoderm)
Epithelium

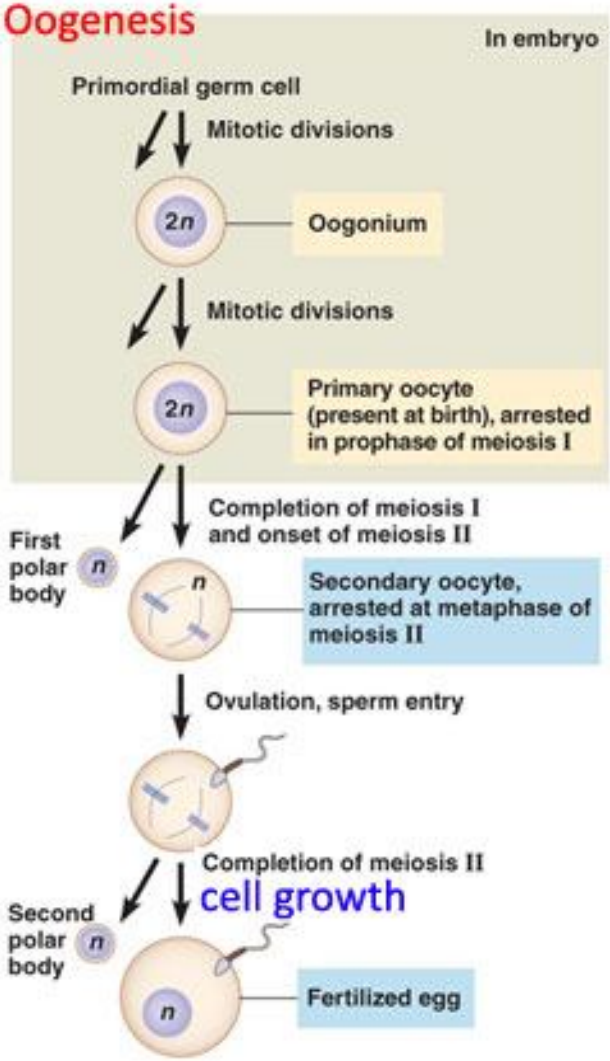
ECTODERM	MESODERM	ENDODERM
• Epidermis of skin and its derivatives (including sweat glands, hair follicles) • Epithelial lining of mouth and anus • Cornea and lens of eye • Adrenal medulla • Tooth enamel • Epithelium of pineal and pituitary glands	• Notochord • Skeletal system • Muscular system • Muscular layer of stomach and intestine • Excretory system • Circulatory and lymphatic systems • Reproductive system (except germ cells) • Dermis of skin • Lining of body cavity • Adrenal cortex	• Epithelial lining of digestive tract • Epithelial lining of respiratory system • Lining of urethra, urinary bladder, and reproductive system • Liver • Pancreas • Thymus • Thyroid and parathyroid glands

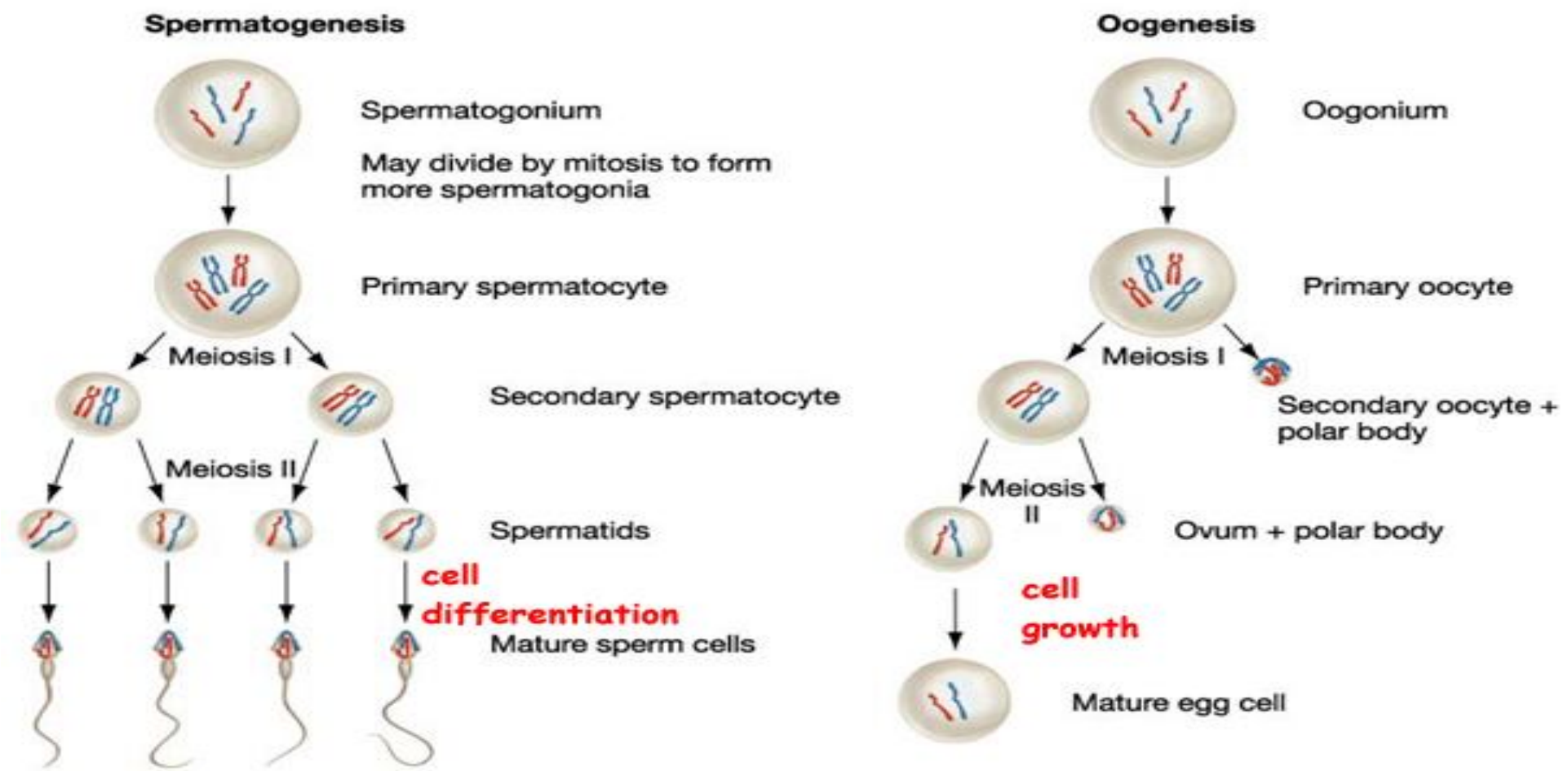

Epiblast
ECTODERM
MESODERM
ENDODERM
Notochord
Somite
Intermediate mesoderm
Lateral plate mesoderm
Somatic mesoderm
Splanchnic mesoderm
• Epidermis, hair, nails, glands of skin
• Brain and spinal cord
• Neural crest: sensory nerve cells and some nervous structures; pigment cells; portions of skeleton; blood vessels in head and neck
Nucleus pulposus of interverte-bral discs
• Sclerotome: vertebrae and ribs
• Dermatome: dermis of dorsal body region
• Myotome: trunk and limb musculature
• Kidneys
• Gonads
• Parietal serosa
• Dermis of ventral body region
• Connective tissues of limbs (bones, joints and ligaments)
• Wall of digestive and respiratory tracts (except epithelial lining)
• Visceral serosa
• Heart
• Blood vessels
Epithelial lining and glands of digestive and respiratory tracts

Spermatogenesis



Oogenesis




Spermatogenesis
Spermatogonium
May divide by mitosis to form more spermatogonia
Primary spermatocyte
Meiosis I
Secondary spermatocyte
Meiosis II
Spermatids
cell differentiation
Mature sperm cells
Oogenesis
Oogonium
Primary oocyte
Meiosis I
Secondary oocyte + polar body
Meiosis II
Ovum + polar body
cell growth
Mature egg cell

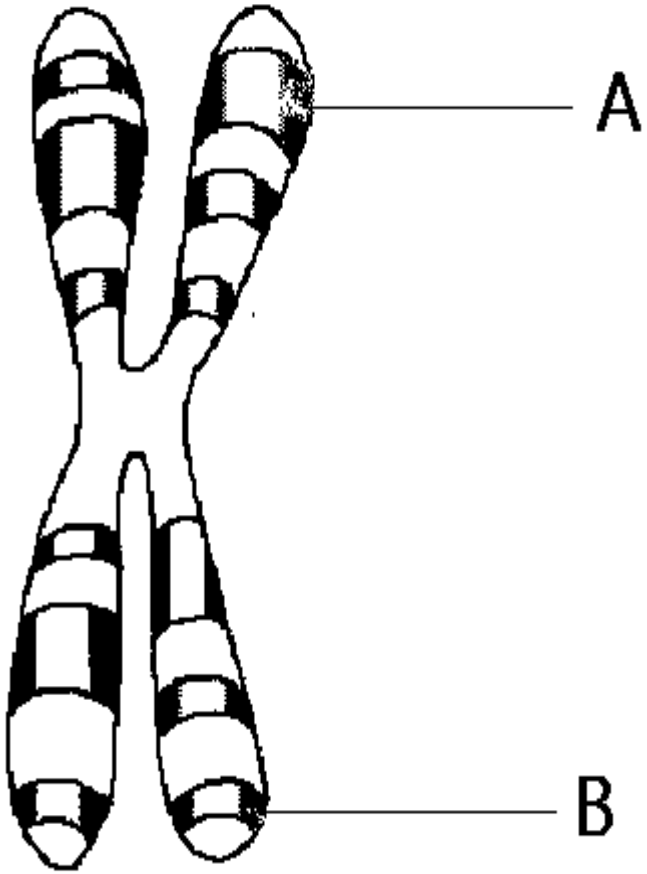
HEREDITARY BASIC

Genetics

- Diploid number of chromosomes
  - In all cells except gametes
  - Diploid number = 46 (23 pairs of homologous chromosomes)
    - 1 pair of sex chromosomes determines the genetic sex (XX = female, XY = male)
    - 22 pairs of autosomes guide expression of most other traits

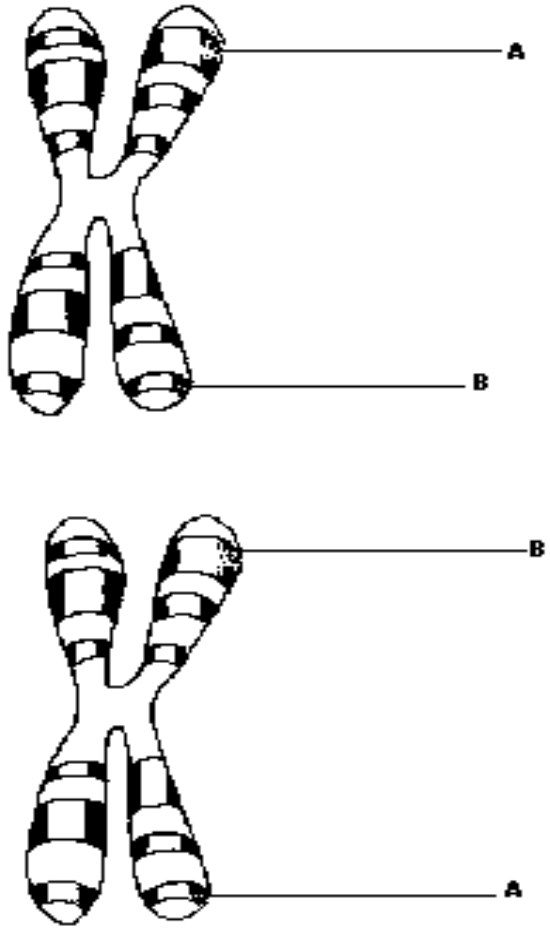
Phenotype

- all of the characteristics of an organism or to one phenotypic trait.
- Also, phenotype refers to any of the detectable attributes of a living thing.
- The phenotype is the result of the interaction of the gene and environmental components.



Alleles

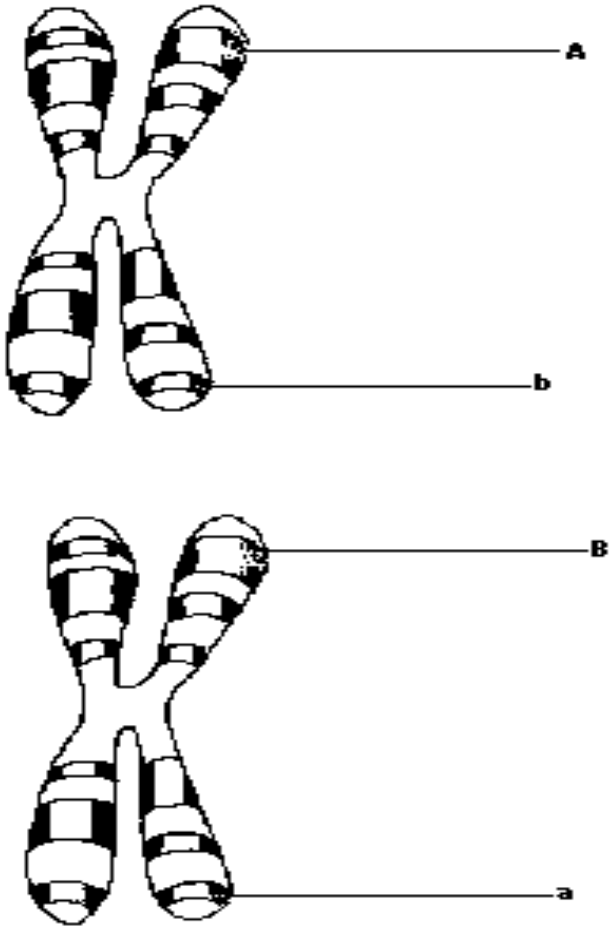
- Matched genes at the same locus on homologous chromosomes
- Homozygous: alleles controlling a single trait are the same
- Heterozygous: alleles for a trait are different
- Dominant: an allele that masks or suppresses its (recessive) partner



Homozygous Alleles

Homozygous

Refers to the state of carrying **identical alleles** at one or more **gene loci** (e.g. AABB or aabb).



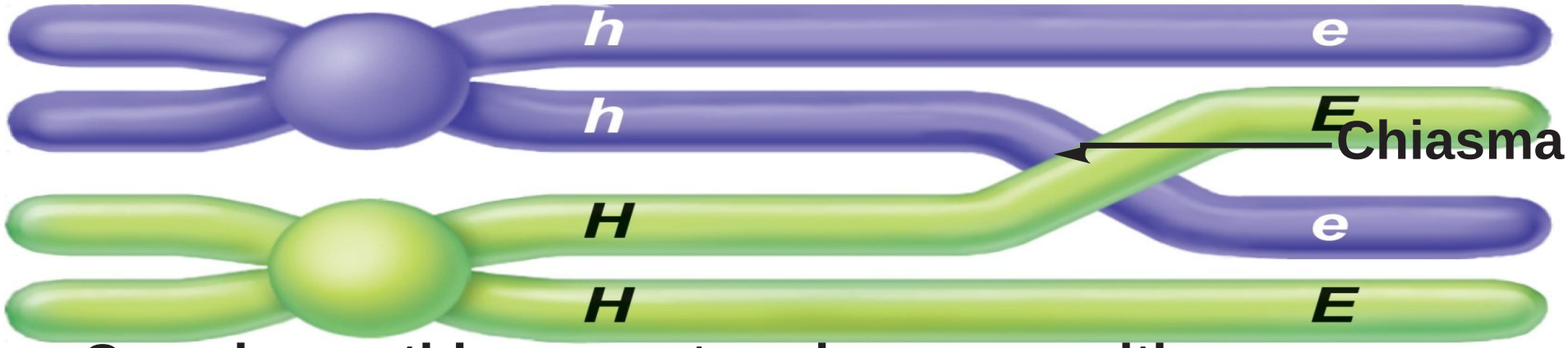
Heterozygous Alleles

Heterozygous

Refers to the state of carrying **different alleles** at one or **more gene loci** (e.g. AaBb).

Crossover and Genetic Recombination

- Genes on the same chromosome are linked
- Chromosomes can cross over, forming a chiasma, and exchange segments
- Recombinant chromosomes have mixed contributions from each parent



One chromatid segment exchanges positions with a homologous chromatid segment—in other words, crossing over occurs, forming a chiasma.

H **Allele for brown hair**

h **Allele for blond hair**

E **Allele for brown eyes**

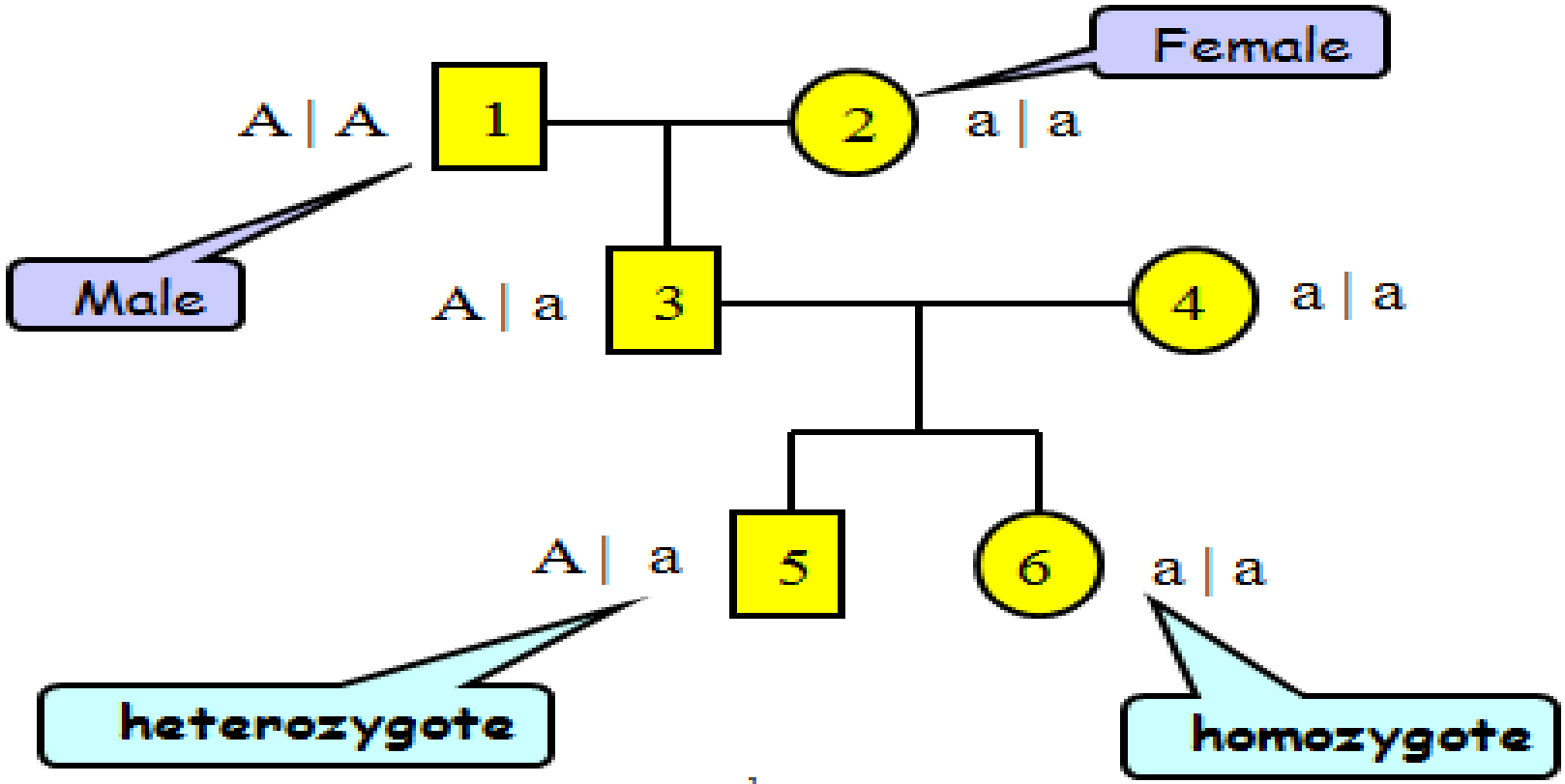
e **Allele for blue eyes**

Paternal chromosome

Maternal chromosome

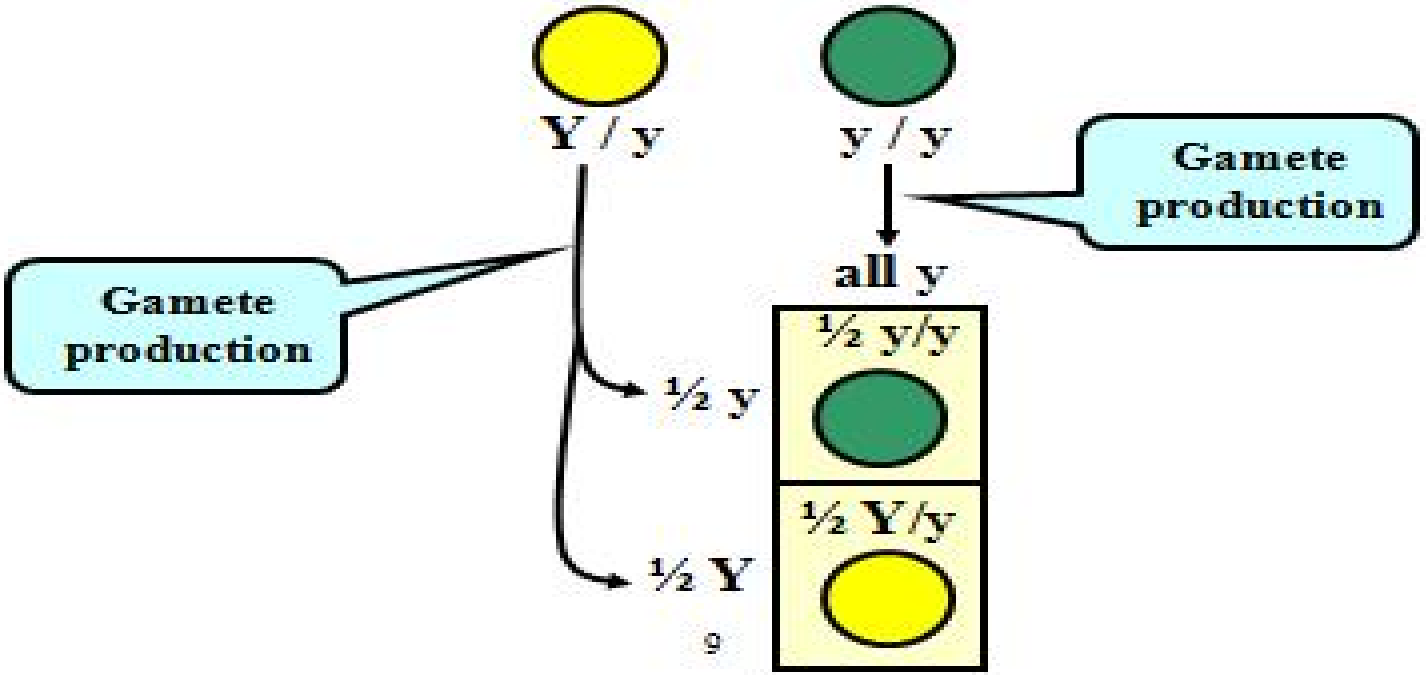
Homologous pair

One Locus Inheritance



Mendel's 1st Law

Two members of a gene pair segregate from each other into the gametes, so half the gametes carry one member of the pair and the other half carry the other member of the pair.



Calculating Probabilities

- We want to predict patterns of inheritance of traits and diseases in pedigrees.
- E.g., we want to know the likelihood that a dog chosen at random from the study population will have blue eyes.

blue eyed

+

brown eyed

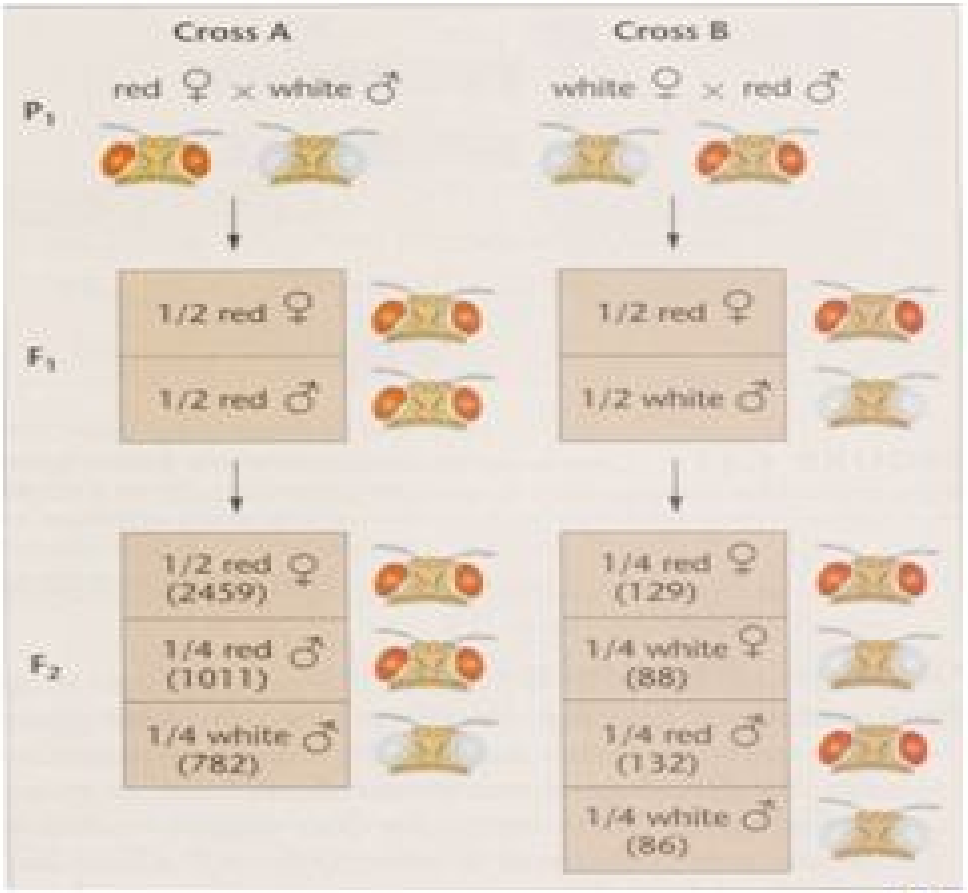
= 30 dogs

$$P(\text{blue eyed}) = \frac{10}{30}$$

$$P(\text{brown eyed}) = \frac{20}{30}$$

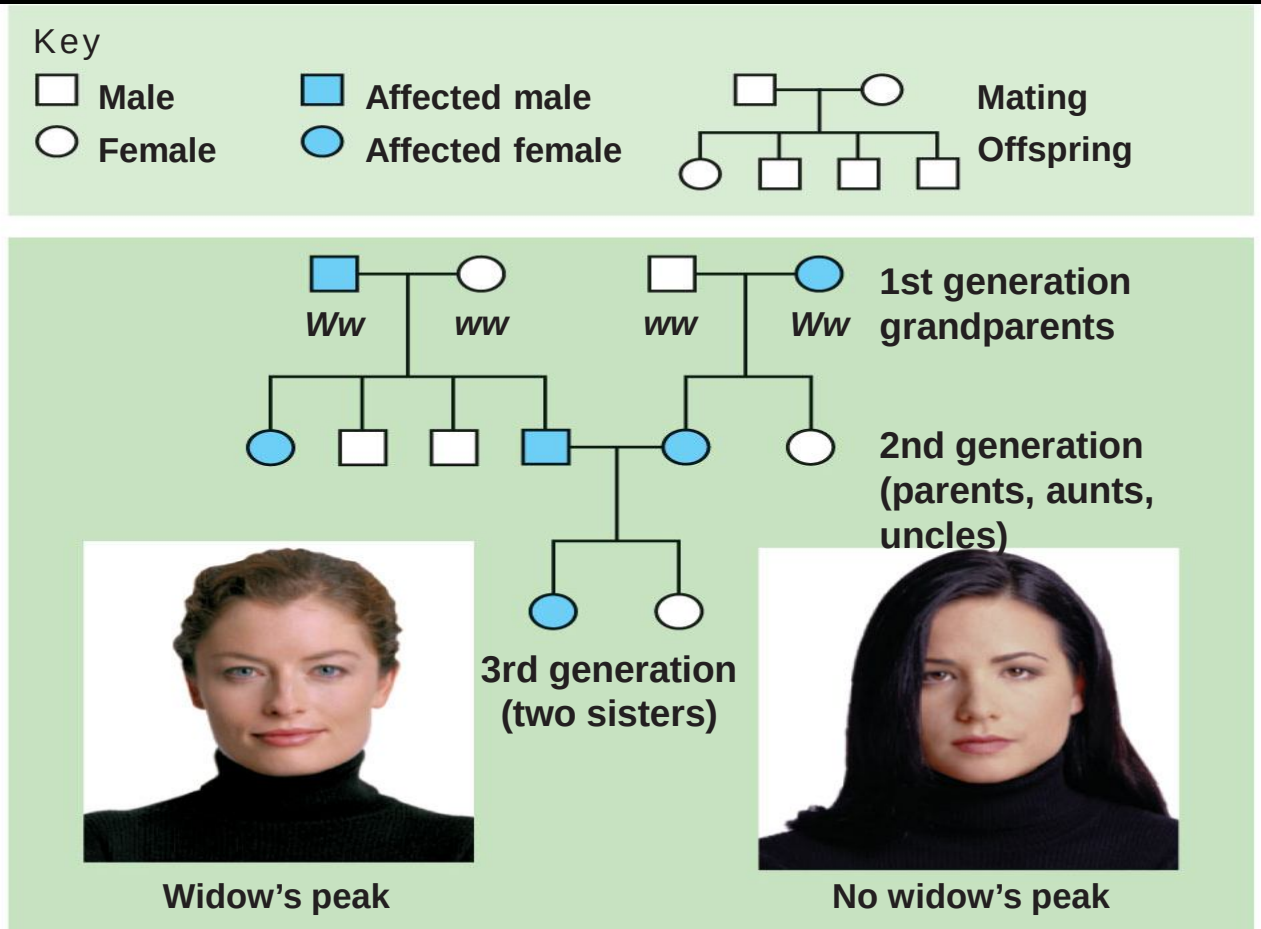
$$P(\text{blue eyed}) + P(\text{brown eyed}) = 1$$

X-linked Inheritance



Different results obtained from reciprocal crosses between red-eyed and white-eyed Drosophila.

Explanation: The gene responsible for eye-color is **X-linked**. Females have 2 X-chromosomes, while males have 1 X-chromosome and 1 Y-chromosome.


Key
Male
Female
Affected male
Affected female
Mating
Offspring
Ww
ww
ww
Ww
1st generation grandparents
2nd generation (parents, aunts, uncles)
3rd generation (two sisters)
Widow's peak
No widow's peak

Medical Genetics

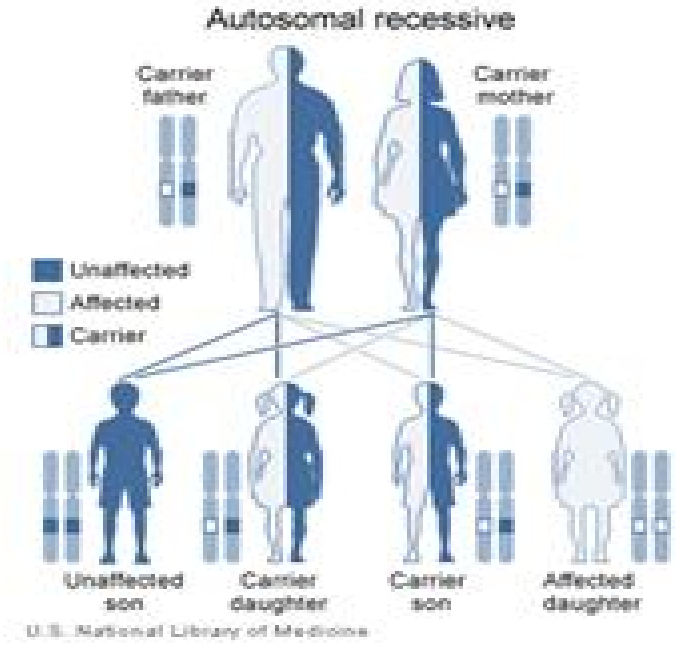
When studying rare disorders, 6 general patterns of inheritance are observed:

- Autosomal recessive
- Autosomal dominant
- X-linked recessive
- X-linked dominant
- Codominant
- Mitochondrial

Medical Genetics (cont.)

Autosomal recessive

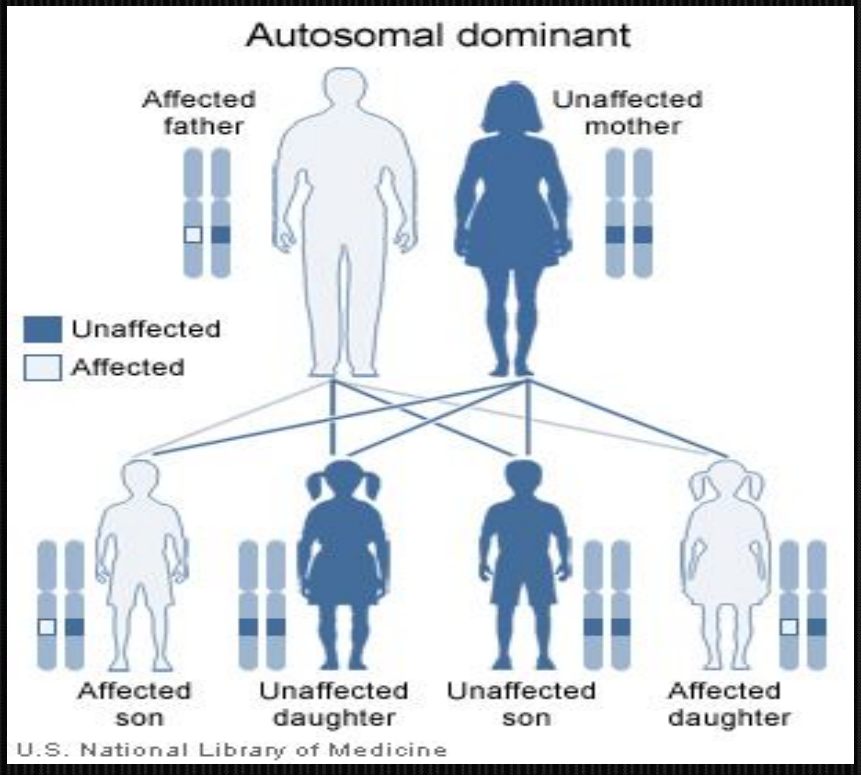
- The disease appears in male and female children of unaffected parents.
- e.g., cystic fibrosis



Medical Genetics (cont.)

Autosomal dominant

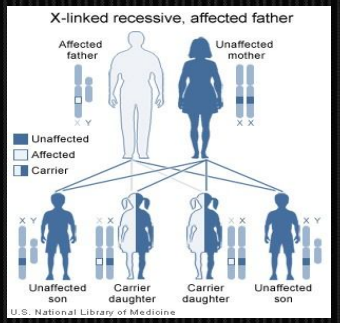
- Affected males and females appear in each generation of the pedigree.
- Affected mothers and fathers transmit the phenotype to both sons and daughters.
- e.g., Huntington disease.

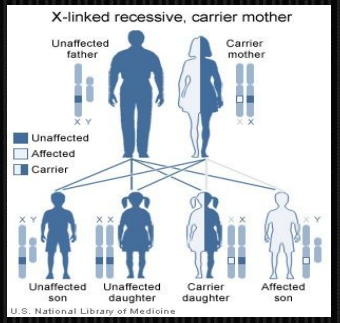


Medical Genetics (cont.)

X-linked recessive

- Many more males than females show the disorder.
- All the daughters of an affected male are "carriers".
- None of the sons of an affected male show the disorder or are carriers.
- e.g., hemophilia

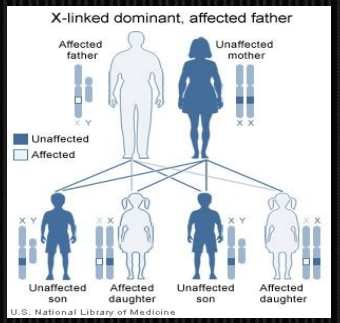


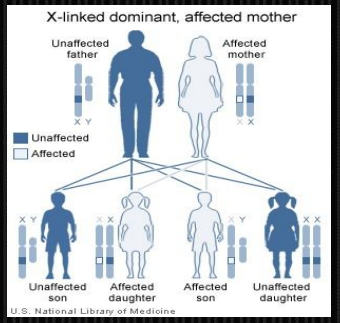


Medical Genetics (cont.)

X-linked dominant

- Affected males pass the disorder to all daughters but to none of their sons.
- Affected heterozygous females married to unaffected males pass the condition to half their sons and daughters
- e.g. fragile X syndrome

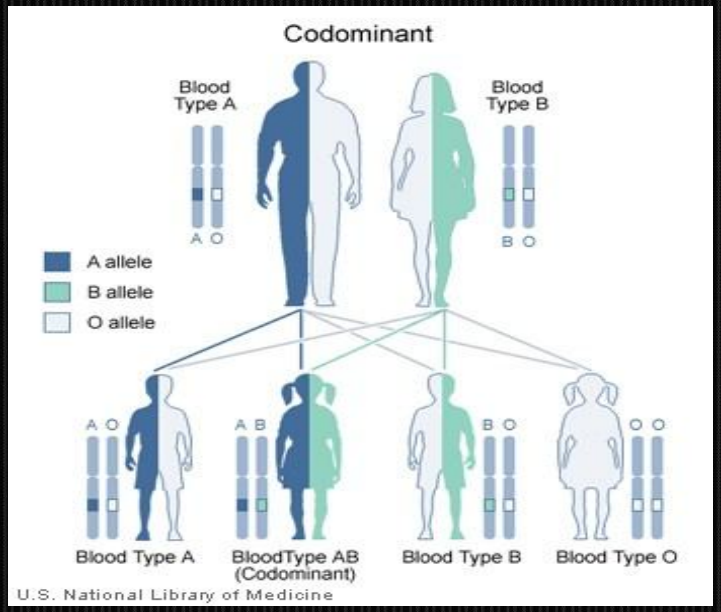




Medical Genetics (cont.)

Codominant inheritance

- Two different versions (alleles) of a gene can be expressed, and each version makes a slightly different protein
- Both alleles influence the genetic trait or determine the characteristics of the genetic condition.
- E.g. ABO locus



Medical Genetics (cont.)

Mitochondrial inheritance

- This type of inheritance applies to genes in mitochondrial DNA
- Mitochondrial disorders can appear in every generation of a family and can affect both males and females, but fathers do not pass mitochondrial traits to their children.
- E.g. Leber's hereditary optic neuropathy (LHON)