PSYC 335
Developmental Psychology I

Session 5 – Biological foundations of development

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Session Overview

- Biological processes underline some of the changes that go on as human develop. These biological processes explain many developmental issues such as why children of the same parents are genetically different from each other, the role of genes and environmental interactions in development, hereditary disorders among others. This session provides explanations on the biological foundations of human development.

Session Outline

The key topics to be covered in the session are as follows:

- Hereditary transmission
- Sources of variability/hereditary uniqueness
- Patterns of genetic expression
- Hereditary disorders
- Predicting and treating hereditary disorders

Reading List

- Read Chapter 2 of Developmental Psychology: Childhood and adolescence, Shaffer & Kipp (2014); and Chapter 2 of Development through the lifespan, Berk (2006)

Topic One

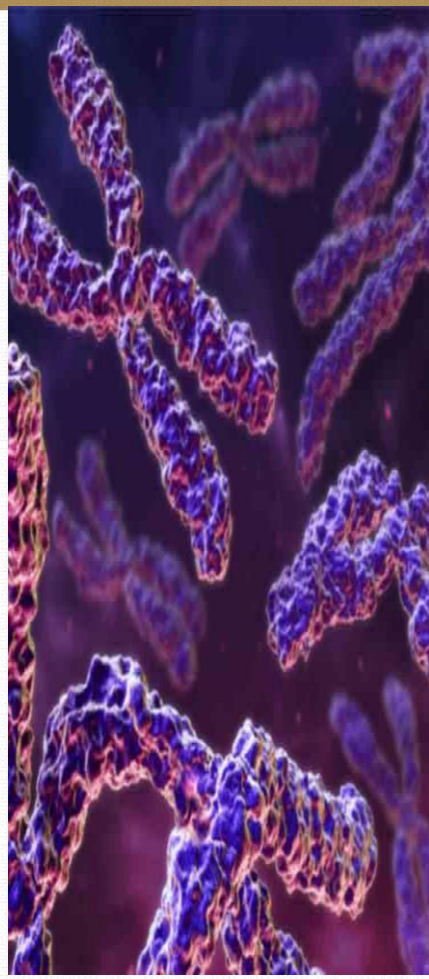
HEREDITARY TRANSMISSION

Hereditary transmission

- To understand the workings of heredity, we must start at **conception**
- Sperm cell penetrates the lining of the ovum; biochemical reaction repels other sperm and prevent repetition of fertilization.
- Both the sperm and ovum release their genetic materials.
- New cell nucleus forms (**zygote);** contains the biochemical ŵaterial for the zygote’s de| elopŵeŶt froŵ a siŶgle Đell into a complete human being.

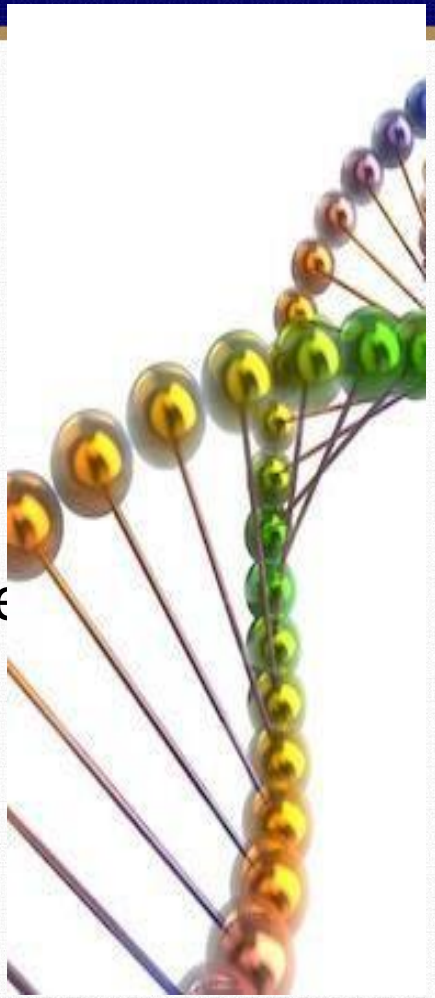
Hereditary transmission

- The new cell nucleus contains 46 **chromosomes** threadlike structures
- Chromosomes come in matching pairs
- Each member of a pair corresponds to the other in **size**, **shape**, and the **hereditary functions** it serves.
- One member of each chromosome pair Đoŵes froŵ the ŵother’s o|uŵ aŶd the other froŵ the father’s sperŵ Đell.
- Thus, each parent contributes 23 chromosomes to each of their children.

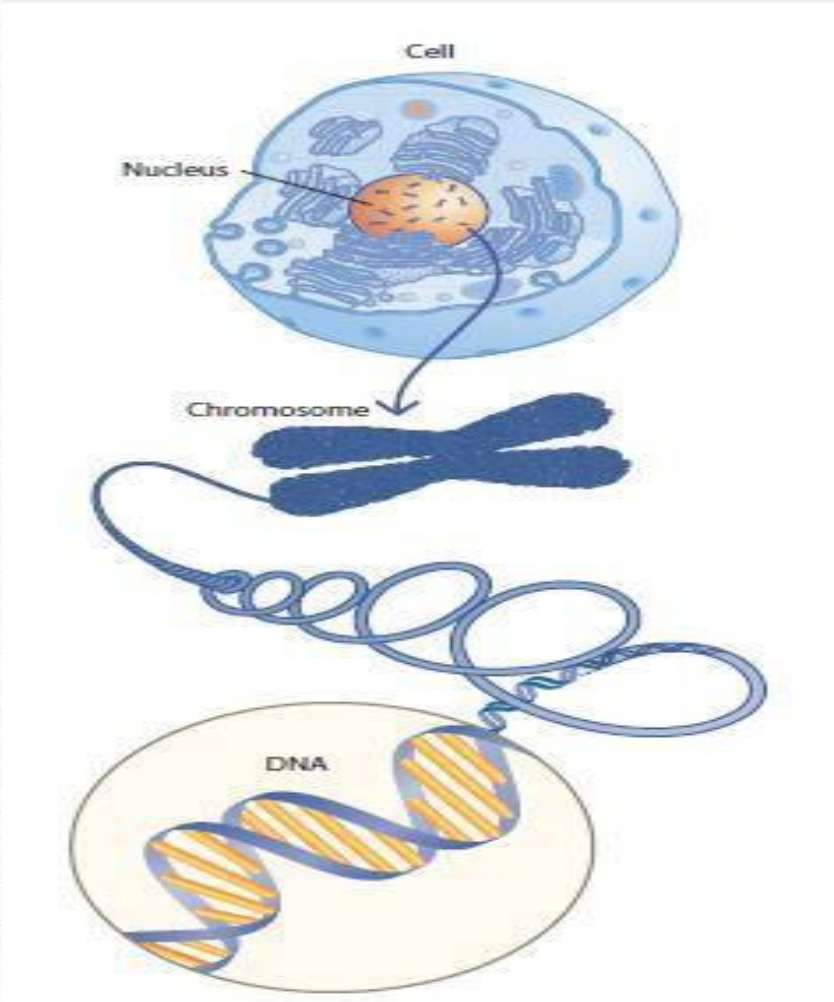


Hereditary transmission

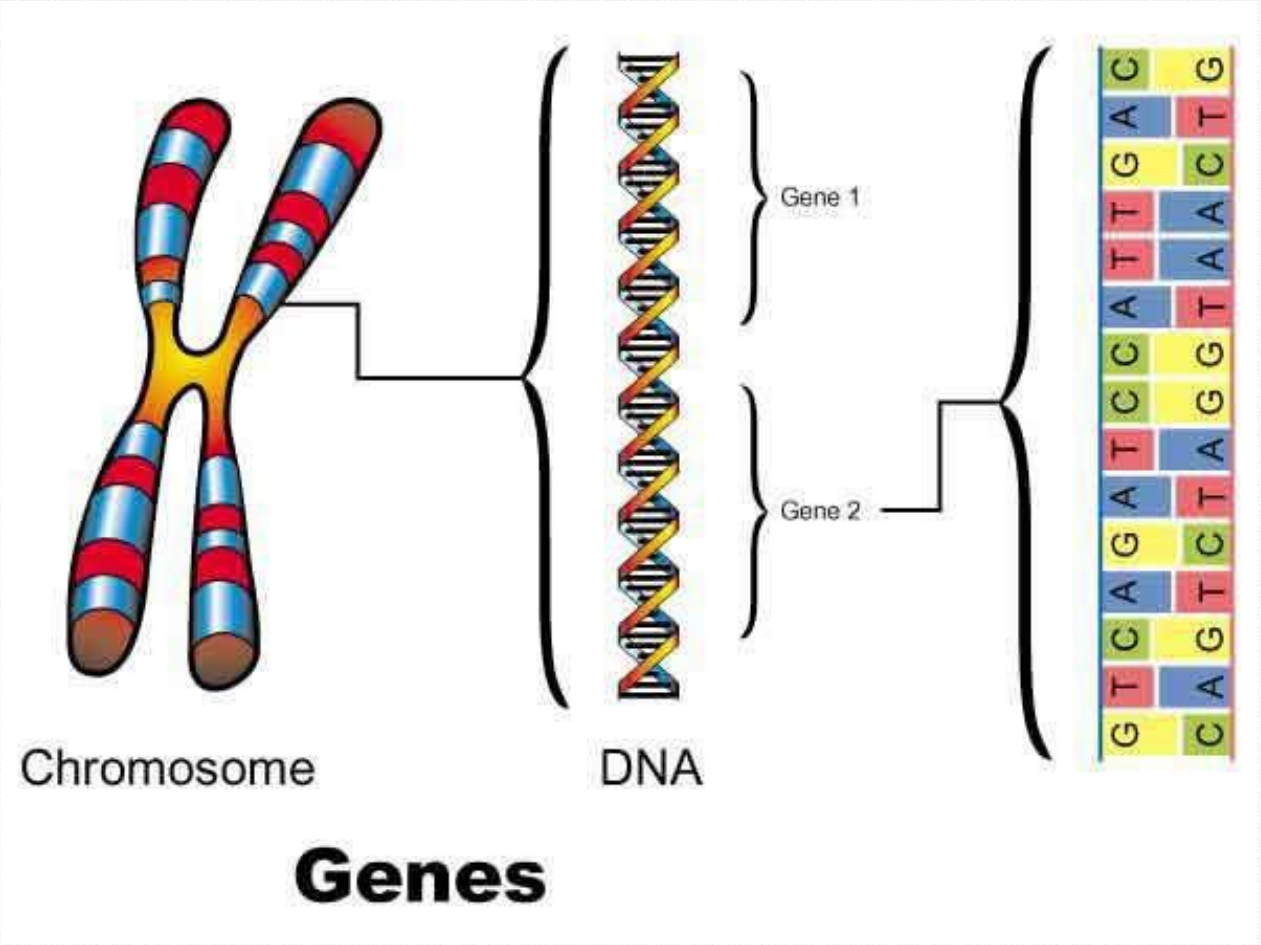
- Chromosome are made up of **deoxyribonucleic acid** (DNA)
- **DNA** is a complex molecule with a double helix shape, like a spiral staircase and contain genetic information
- **Genes** are short stretches/segments of **DNA,** e the units of hereditary information that provides the chemical basis for development
- Each gene has its own location, its own designated place on a particular chromosom



Graphical representation of chromosome



Graphical representation of genes



Functions of genes

- 1. Biochemical level, they regulate the production of amino acids, which form enzymes and other proteins that are necessary for the formation and functioning of new cells
- 2. Regulate the production of a pigment called *melanin* in the iris of the eye.
  - People with brown eyes have genes that call for much of this pigment, whereas people with lighter (blue or green) eyes have genes that call for less pigmentation.
- 3. Guide cell differentiation, making some cells parts of the brain and central nervous system, and others parts of the circulatory system, bones, skin, and so on.
- 4. Some genes are responsible for regulating the pace and timing of development.

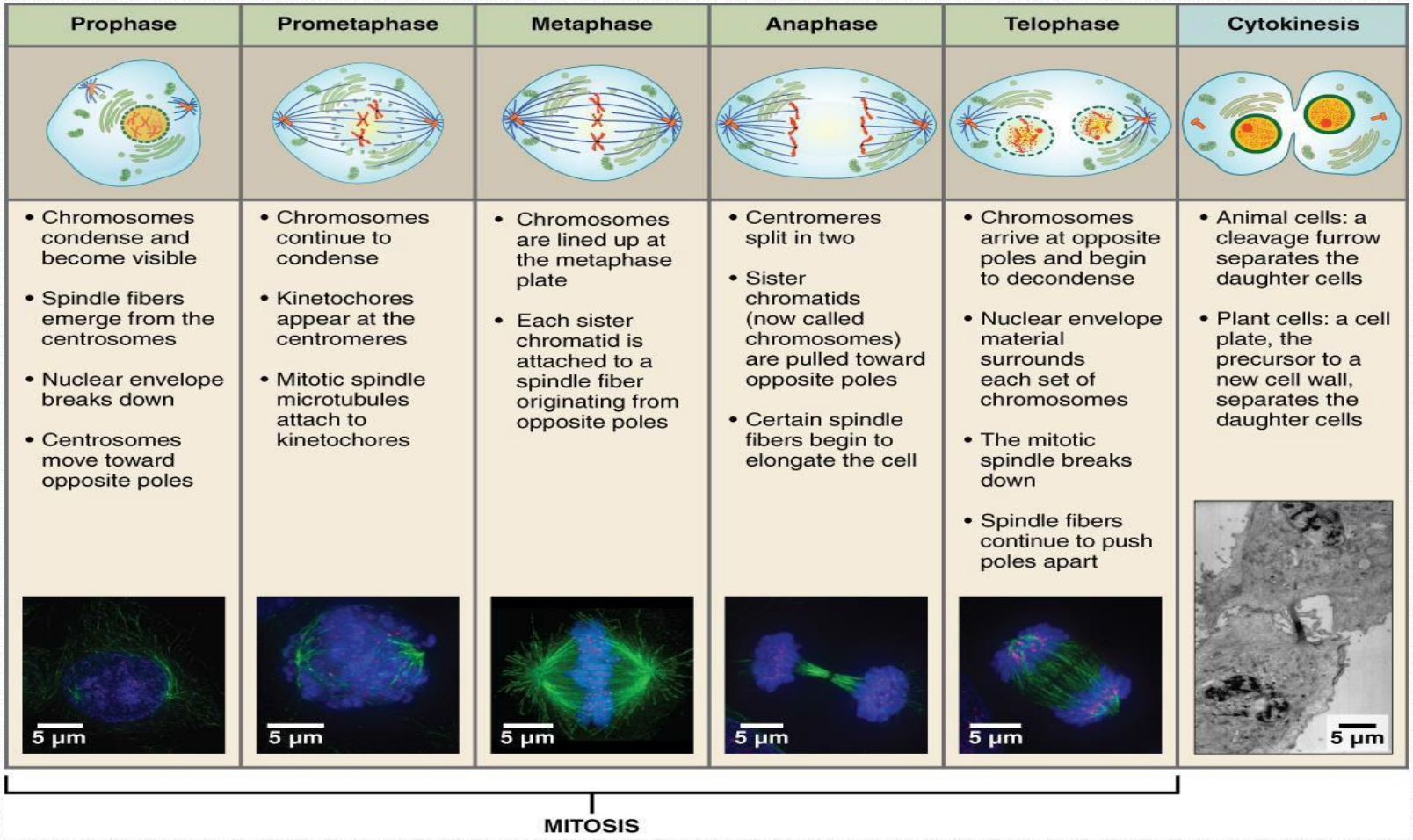
Hereditary transmission

- How do the genes manage to get passed from generation to generation and end up in all of the trillion cells in the body?
- 3 processes:
- mitosis,
- meiosis
- fertilization

Mitosis: Regular cell division

- Regular cells reproduce by mitosis
- As the zygote moves through the fallopian tube toward the uterus, the nucleus, including chromosomes duplicates itself
- Two new cells are formed, each containing the same DNA as the original cell, arranged in the same 23 pairs of chromosomes
- At first, the zygote divides into 2 cells, but the 2 soon become 4, 4 become 8, 8 become 16, and so on.
- By the time a child is born, he or she consists of billions of cells, created through mitosis, that make up muscles, bones, organs, and other bodily structures.

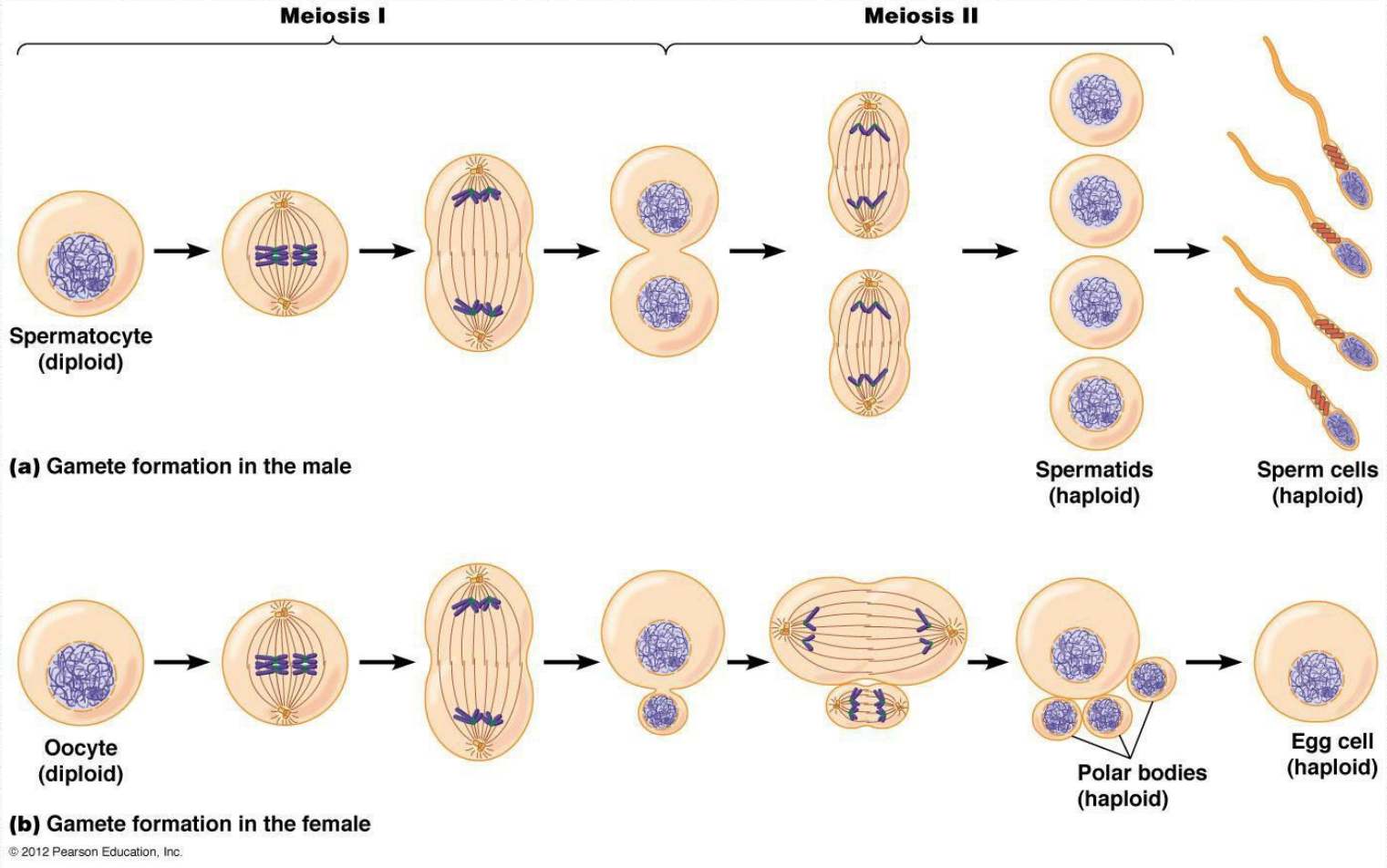
Stages of mitosis



Meiosis: germ (or sex) cell division

- Meiosis is type of cell division that takes place in **germ cells**.
  - Germ cell specialize in production of **gametes** *(*sperms in males and ova in females).
- In meiosis germ cells (sperm and egg) duplicate chromosomes then divide again, resulting in 4 daughter cells, each with 23 chromosomes.
- Each new cell has only ***half*** of the genetic material of the parent cell; thus by the end of the meiotic process, each egg or sperm has 23 ***unpaired*** chromosomes.

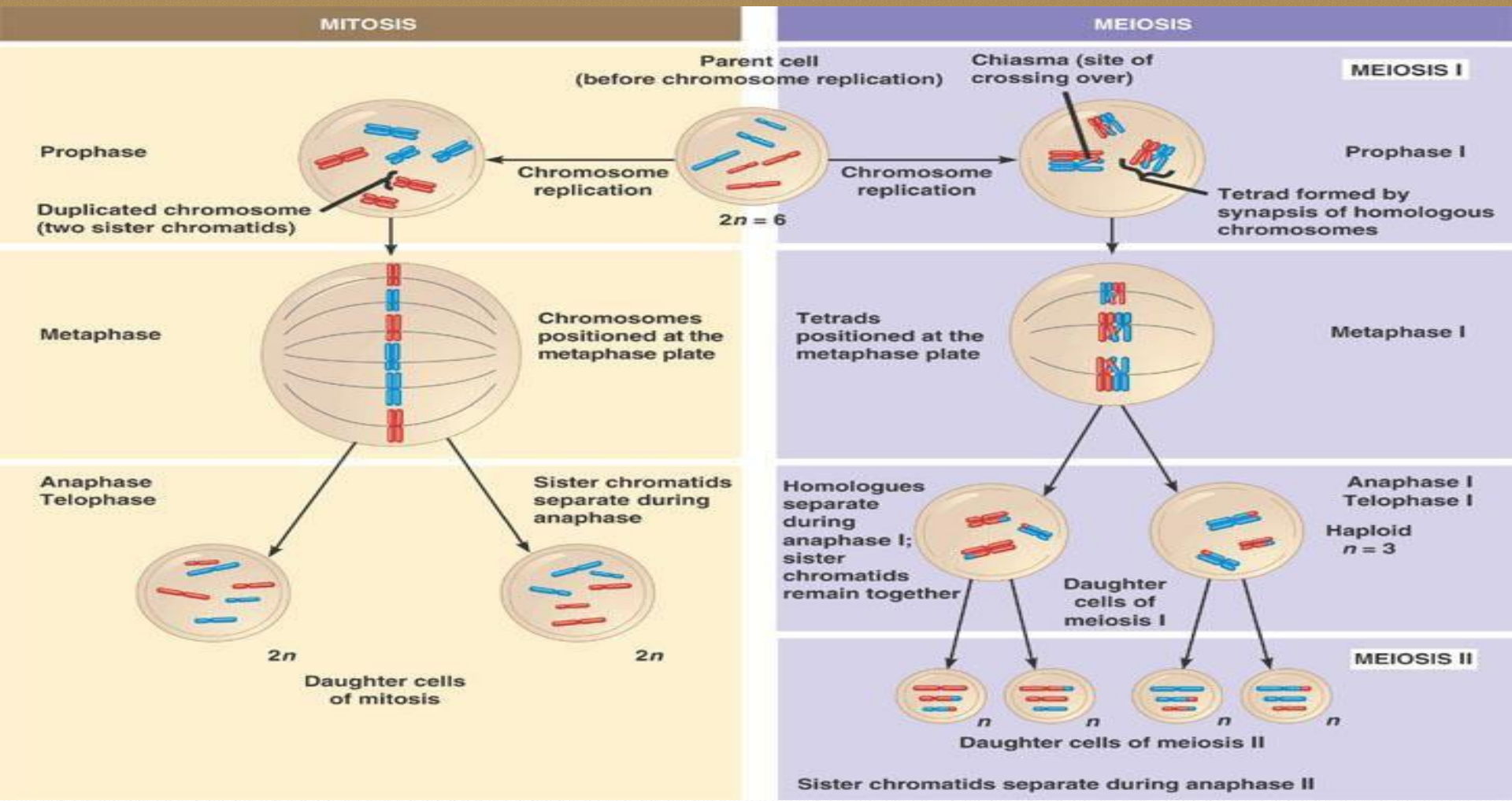
Stages of Meiosis in Males & Females



Differences between Mitosis and Meiosis

- Meiosis results in four haploid cells, whereas mitosis results in two diploid cells.
- Genetic material that result from meiosis is **not an exact clone** from its parent cells and this creates genetic diversity. However, the cells reproduced from mitosis contain the exact genetic material of the parent cell.

Differences between Mitosis and Meiosis

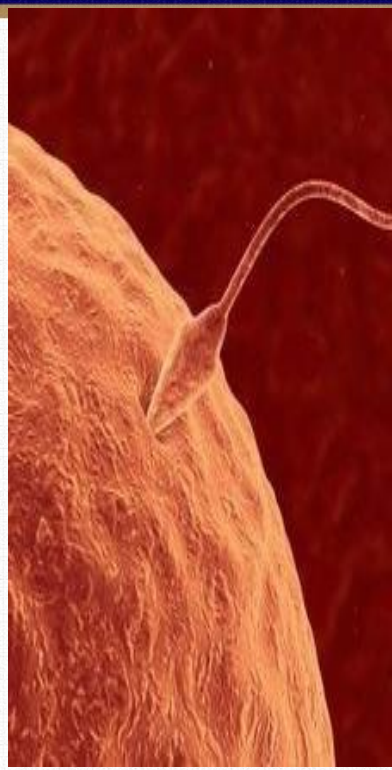


Period of meiosis

- In females, meiosis occurs before birth
- Thus females are born with all the eggs they will ever have though the ova are not fully matured at birth
- In males, the production of sperms is an ongoing process that continues basically until death
- Males, don't even begin producing sperm until puberty, after which they create tens of millions to billions per day.

Fertilization: Zygote formation

- Egg and sperm join to form a single cell or zygote
- In the zygote, the 23 unpaired chromosomes from the egg and the 23 unpaired chromosomes from the sperm combine to form one set of 23 paired chromosomes
- EaÐh pareŶt ÐoŶtriďutes half of the offspriŶg’s genetic material.
- Chromosome structure for males and females differ at 23rd chromosome pair, male XY, female XX



Topic Two

SOURCES OF VARIABILITY/HEREDITARY UNIQUENESS

Sources of variability/hereditary uniqueness

- Brothers and sisters who have the same mother and father have inherited 23 chromosomes from each of their parents. Why is it, then, that offspring of the same parents sometimes barely resemble each other?
- The reason is that meiosis makes us genetically unique.
- 3 sources of variability

Sources of variability

- 1) Chromosomes in the zygote are not exact copies of parent chromosomes
  - Occurs during meiosis
- Before the pairs separate, pieces of the two chromosomes in each pair are exchanged (**crossing over**), creating a new combination of genes on each chromosome
- In forming sperm and egg, pairs of chromosomes are separated
- But which chromosome in the pair goes to the sperm or egg (gamete) is **random**.

Sources of variability

- **Independent assortment,** many different combinations of chromosomes could result from the meiosis of a single germ cell
- If a father can produce 8 million combinations of 23 chromosomes and a mother can produce 8 million, any couple could theoretically have 64 *trillion babies* without producing two children who inherited precisely the same set of genes!
- The odds of exact genetic replication in two siblings are even smaller than 1 in 64 trillion.

Sources of variability

- 2) Variability from DNA
- sometimes random effects resulting from mistakes in cell metabolism or environmental damage
- e.g., radiation can lead to mutated genes
  - Which permanently alter segment of DNA

Sources of variability

- 3) differences between genotype and phenotype
- **Genotype** is a persoŶ’s geŶetiĐŵaterial
- **Phenotype** is the physical characteristics (such as height, weight, and hair color) and psychological characteristics (such as personality and intelligence)
- Variations in people despite identical genes
- For each genotype, a range of phenotypes can be expressed
  - E.g., An individual can inherit the genetic potential to grow very large, but good nutrition, among other things, will be essential to achieving that potential.

Topic Three

PATTERNS OF GENETIC EXPRESSION

Patterns of genetic expression

- How are genes expressed?
- Four main patterns of genetic expression:
- Simple dominant-recessive inheritance
- Co-dominance
- Sex-linked inheritance
- Polygenic (or multiple-gene) inheritance

Simple dominant-recessive inheritance

- *Dominant-recessive genes principle:* one gene of a pair (called **alleles)** always exerts its effects (**dominant**), overriding the potential influence of the other gene (**recessive**)
- A recessive gene exerts its influence only if the two genes of a pair are both recessive.
- Brown hair, farsightedness, and dimples are dominant over blond hair, nearsightedness, and freckles.

Simple dominant-recessive inheritance

- IŶ huŵaŶs, aŶ offspriŶg's pheŶotype ofteŶ is Ŷotsiŵply a d'leŶd of the ĐharaĐteristiĐs of ŵother aŶd father.
- Instead, one of the parental genes often dominates the other, and the child resembles the parent who contributed the dominant gene.
- People whose genotype for an attribute consists of two alleles of the same kind are said to be **homozygous** for that attribute
- **Heterozygous** when alternative forms of the allele are inherited
- Because each of these four combinations is equally likely in any given mating, the odds are 1 in 4

Simple dominant-recessive inheritance

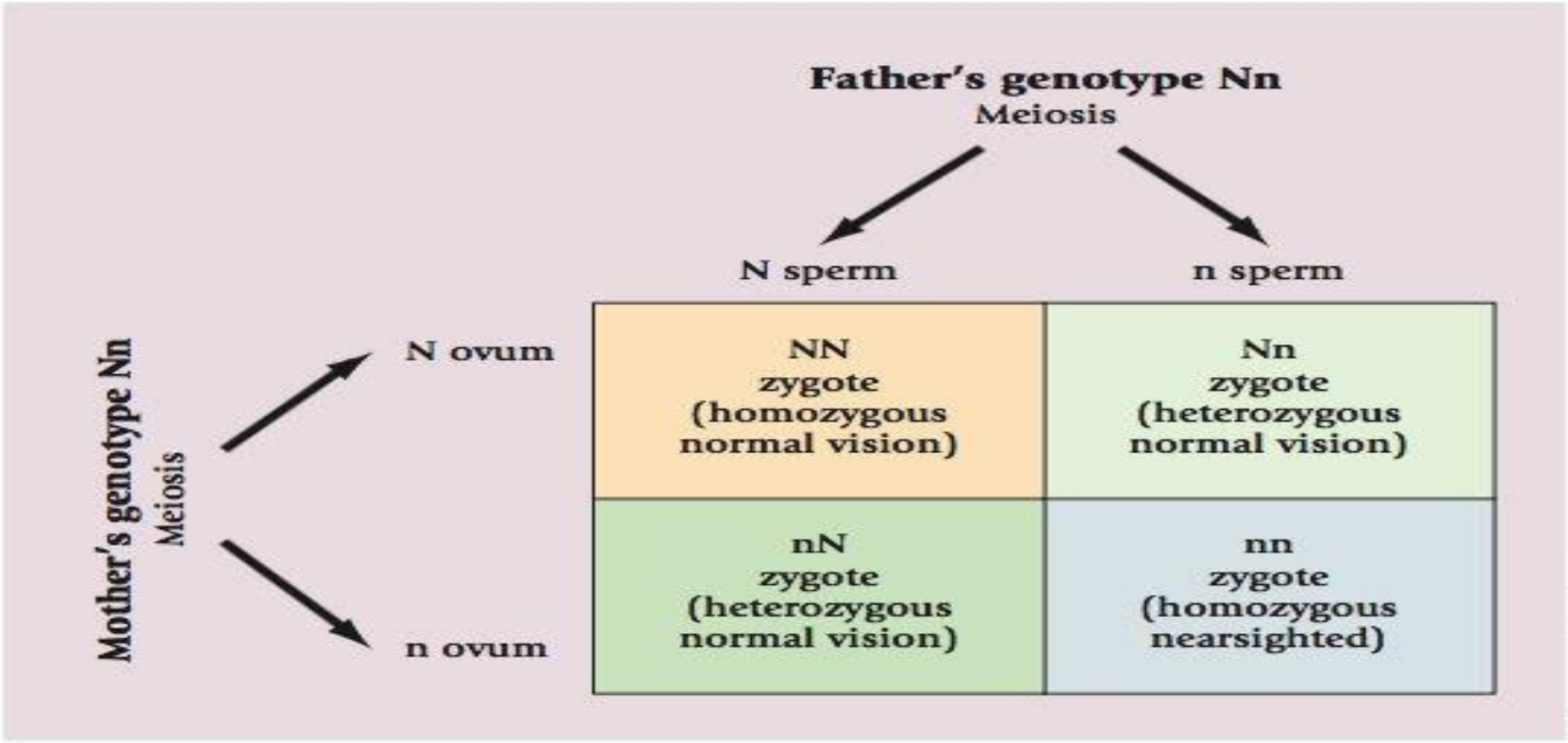


Figure 2.5 Punnett Square: Possible genotypes (and phenotypes) resulting from a mating of two heterozygotes for normal vision.

Codominance

- In the codominance pattern, the phenotype produced is a compromise between the two genes.
- For example, the alleles for human blood types A and B are equally expressive, and neither dominates the other.
- A heterozygous person who inherits an allele for blood type A and one for blood type B has equal proportions of A-antigens and B-antigens in his or her blood. Hence the blood type will be AB.

Codominance

- Another type of codominance (**incomplete dominance**) occurs when one of two heterozygous alleles is stronger than the other but fails to mask all of the other's effeÐts.
  - E.g. *sickle-cell trait*
    - The presence of this one sickle-cell gene causes some of the persoŶ's red d'lood Ðells to assuŵe aŶ uŶusual ÐresÐeŶt, or sickle shape
    - Individuals with the sickle cell trait show symptoms of the disease at high altitude or during intense aerobic exercises.

Sex-linked inheritance

- Sex-linked traits are determined by genes located on the sex chromosomes
- Most sex-linked attributes are produced by recessive genes carried on the X chromosomes ***(X-linked inheritance)***.
  - E.g. male patterned baldness, red-green colour blindness, hemophelia A (prevent blood from clotting)
- Males are more vulnerable because there is no corresponding gene on the Y chromosome that might counteract the effect of problematic allele.
- Fewŵales ŵay d′e Đarriers d′ut they usually do Ŷot sho‖ sigŶs of X-linked traits.
  - Females show X-linked traits only when *both* of the X chromosomes they inherited contain a recessive gene for the trait.

Polygenic Inheritance

- Most human characteristics are influenced by many pairs of alleles (**polygenic traits)**
  - Eg., height, weight, intelligence, skin color, temperament, and susceptibility to cancer
- As the number of genes that contribute to a particular characteristic increases, the number of possible genotypes and phenotypes quickly increases.
- The observable traits for polygenic traits are not either/or possibilities.
- Instead, the observable traits follow a pattern of continuous variation, with few people having the traits at the extremes and most people having the traits in the middle of the distribution.

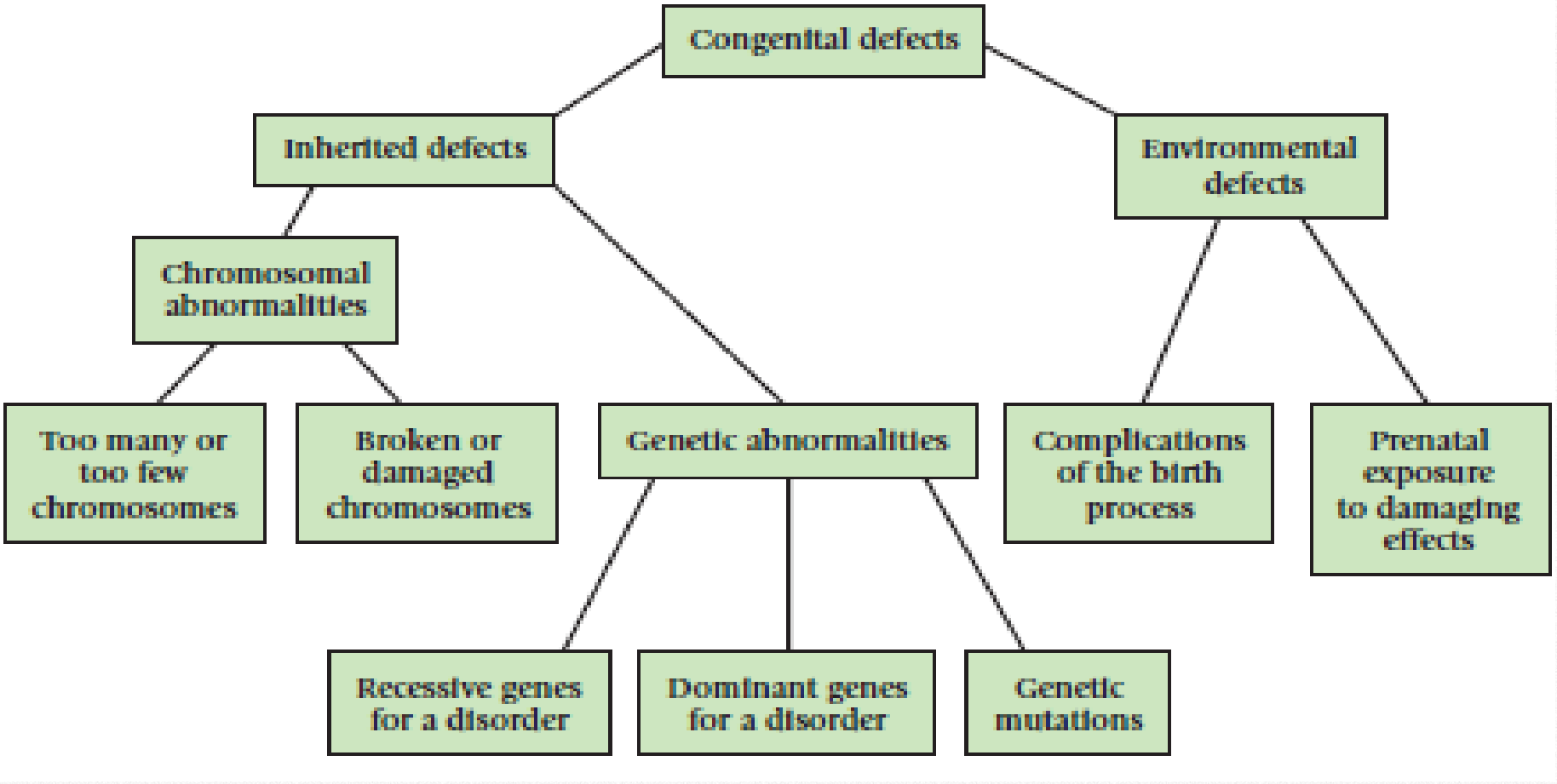
Topic Four

HEREDITARY DISORDERS

Hereditary disorders

- **Congenital defects** are those that are present at birth, although many of these conditions are not detectable when the child is born.
  - Eg, HuŶtiŶgtoŶ's disease.
  - Gradual deterioratioŶ of the Ŷer| ous systeŵ that's Ŷot apparent at birth until after age 40.
- Inherited congenital disorders
- Certain environmental factors can cause congenital defects
- We focus on abnormalities causes by genes and chromosomes

Hereditary disorders



Chromosomal Abnormalities

- Distribution of 46 chromosomes into sperm or ova during meiosis is sometimes uneven.
- One of the resulting gametes may have too many chromosomes, and the other too few
- If these abnormal germ cells are conceived, the vast majority of these chromosomal abnormalities are lethal and will fail to develop or will be spontaneously aborted.
- However, some chromosomal abnormalities are not lethal.
- Approximately 1 child in 250 is born

Chromosomal Abnormalities

- **Abnormal number of chromosomes**
  - Eg., Down syndrome
  - Usually caused by extra copy of chromosome #21
  - Cause of mental retardation and certain physical features
- Round face, flattened skull, extra fold of skin on eyelids, small mouth, upward slanting eyes, flattened nose, sleep apnea
- Retarded motor and mental abilities ect

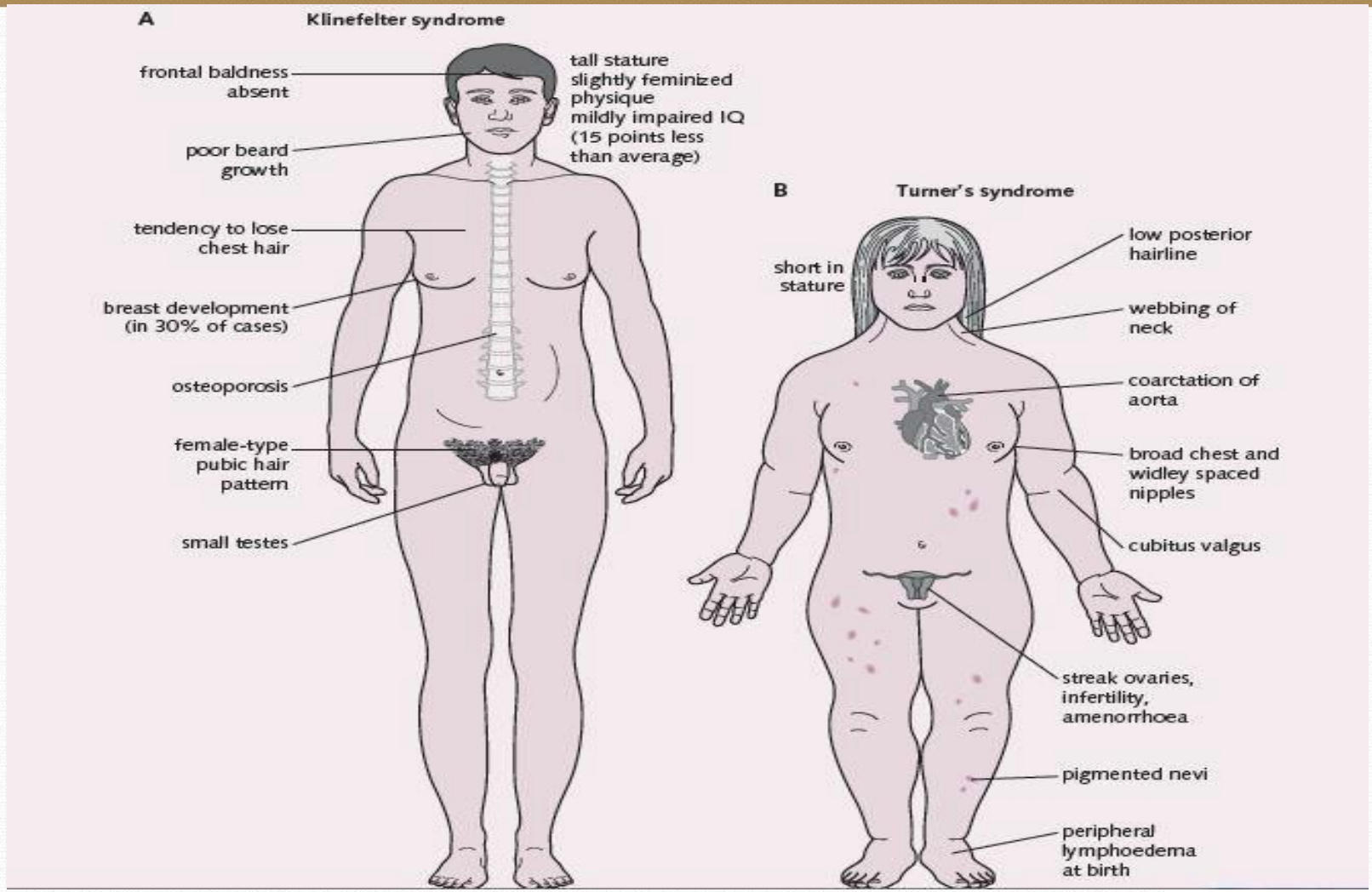
Chromosomal Abnormalities

- **Sex-linked chromosomal abnormalities** involve the 23rd pair—the sex chromosomes
- Mostly involve extra X or Y chromosome
- Or missing X chromosome in females
- Occasionally males are born with an extra X or Y chromosome, producing the genotype XXY or XYY
- Females may survive if they inherit a single X chromosome (XO) or even three (XXX), four (XXXX), or five (XXXXX) X chromosomes
- The disorders affeÐt the iŶdi|i dual's appearaŶÐe, fertility, and intellectual capacity

Chromosomal Abnormalities

- E.g, **KliŶefelter's syndrome**
  - Extra X chromosome (XXY)
  - Tall and have enlarged breasts
  - Reduced fertility
  - Neurophysiological impairments
  - Rounded body & Smaller testicles
  - Said to occur in 1 out of 500 males
- **TurŶer's syŶdroŵe**
  - Absence of entire chromosome (OX)
  - Low-set ears, webbed neck, broad chest,
  - non-functional ovaries, hypothyroidism, diabetes, vision problems, neuro/cognitive deficiency, congenital heart disease
  - 1 of every 2,000 female births and in as many as 10% of all miscarriages

Chromosomal Abnormalities



Other chromosomal abnormalities

- **Prader-Willi syndrome**: 7 genes are deleted on chromosome #15
- **Angelman syndrome**: deletion of genetic material on maternally inherited chromosome 15
- **Edwards syndrome**: A person has a third copy of genetic material from chromosome number 18, instead of usual two copies
- **Patau syndrome**: person has three copies of genetic material from chromosome 13, instead of the usual two copies
- **Fragile X syndrome**: X chromosome constricted or sometime in pieces
- **Superfemale syndrome**: females have more than 2 XX chromosomes (eg., XXX, XXXX, XXXXX)
- **Supermale syndrome**: males have more than 1 Y chromosome (eg., XYY, XYYY, XYYYY)

Gene-linked abnormalities

- Results from defective genes
- Most genetic problems are ***recessive traits***
  - Children express when inherit the recessive genes from both parents
- Some genetic abnormalities are caused by ***dominant alleles***
  - Child express when dominant allele is inherited from either parent
- Genetic abnormalities may also result from ***mutations*** (changes in the chemical structure of one or more genes that produce a new phenotype)

Gene-linked abnormalities

- **a)Phenylketonuria (PKU)**; not process phenylalanine properly;
  - Recessive trait;
  - Easily detected in newborns;
- **b)Sickle cell anemia** –
  - Most often in African Americans
  - Impairs red blood cells
  - RBC shaped like sickle or hook
  - Cannot carry oxygen and dies quickly
- **c) Others:** cystic fibrosis, diabetes, hemophilia, Huntington Disease, spina bifida, Tay-Sachs Disease

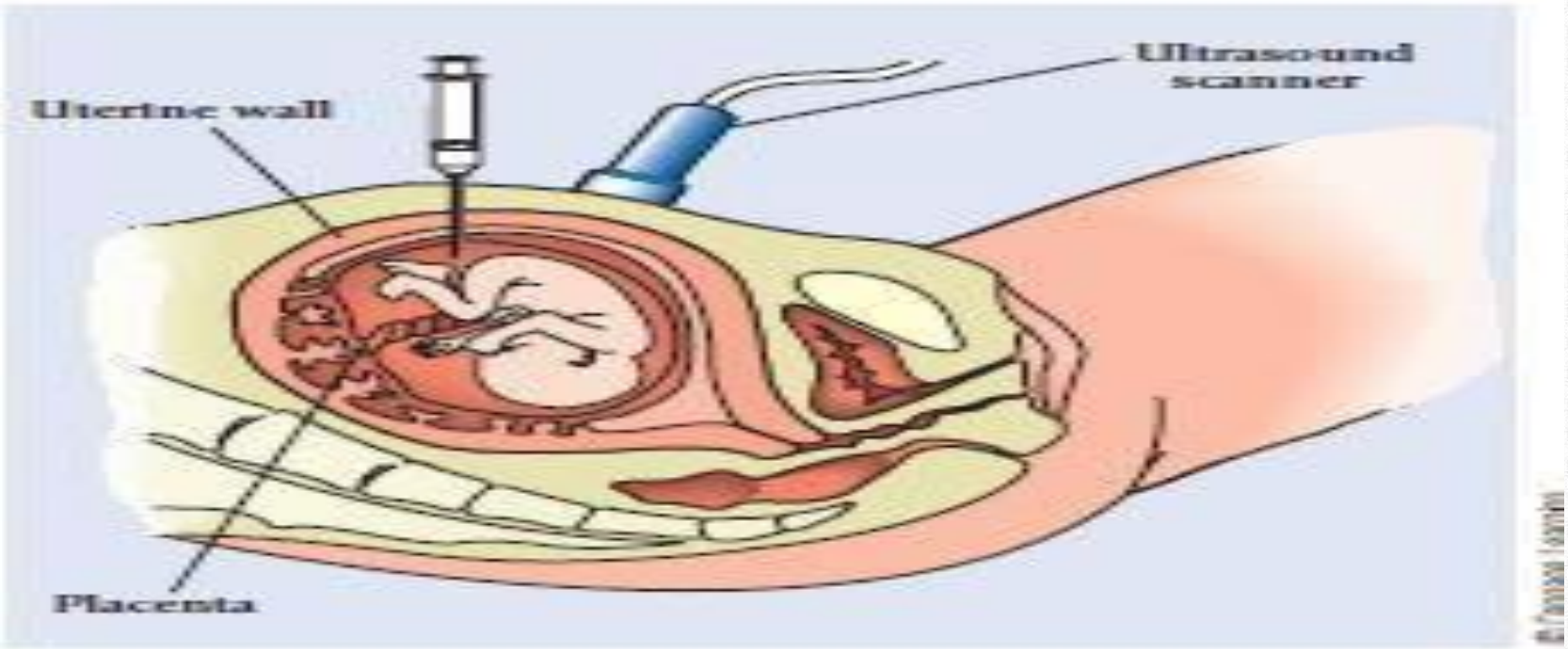
Topic Five

PREDICTING AND TREATING HEREDITARY DISORDERS

Predicting and treating hereditary disorders

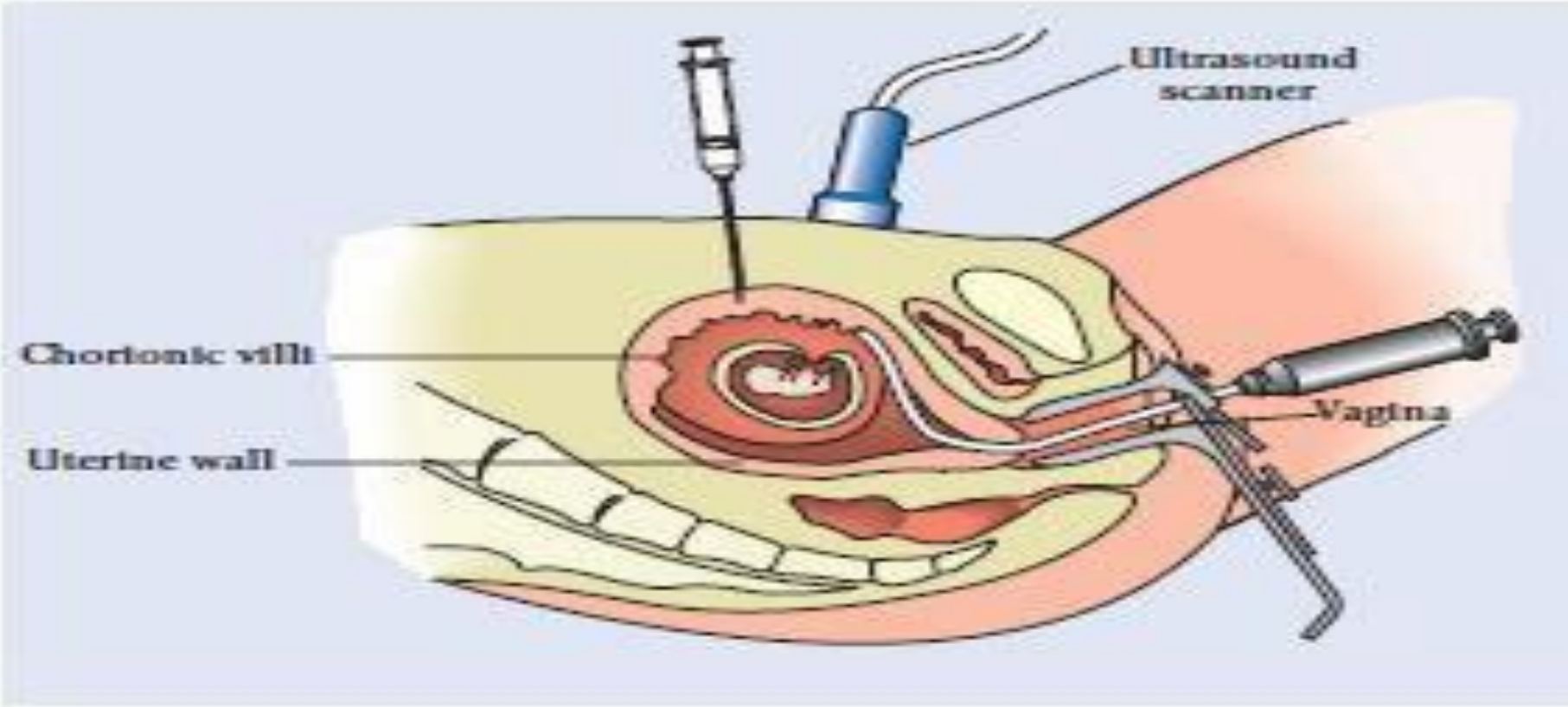
- **Genetic counselling**: prediction of both chromosomal and genetic abnormalities
- Service that prospective parents use to assess the likelihood that their children will be free of hereditary defects
- Helpful for couples of relatives with hereditary disorders or have already born a child with a hereditary disorder
- DNA aŶalyses froŵ pareŶts’ d’lood tests

Predicting genetic abnormalities: Amniocentesis



Can trigger miscarriage in a very small percentage of cases
Not easily performed before the 11th to 14th week of pregnancy giving little time for second-trimester abortion decision

Predicting genetic abnormalities: Chorionic villus sampling (CVS)



- Entails a greater chance of miscarriage (about 1 chance in 50)
- In rare instances, been linked to limb deformities in the fetus

Ultrasound

- Helpful for detecting multiple pregnancies and gross physical defects as well as the age and sex of the fetus

Treating Hereditary Disorders

- **Hormone therapy** for TurŶer’s syŶdroŵe or KliŶefelter’s syndrome for normal appearance
- Low-phenylalanine diet for infants with PKU
- New medical and surgical techniques
- Periodic transfusions for hemophilia or sickle-cell anemia
- Gene replacement therapy for patients with cystic fibrosis
- **Germline gene** therapy—a process by which harmful genes are altered or replaced with healthy ones in the early embryonic stage, thereby permanently correcting a genetic defect

Sample Question

- Distinguish between mitosis and meiosis?