Meiosis

Describe the process of meiosis.

- This two-phase process divides the chromosomes of a diploid germ cell and generates 4 haploid gametes
- **Meiosis I**: Homologous chromosomes separate
    - Prophase I – nuclear envelope begins to break down, nuclear chromatin condenses into individual chromosomes made up of two sister chromatids
    - Metaphase I – pairs of homologous chromosomes move along their microtubule attachments so they are lined up along the metaphase plate
    - Anaphase I – attachments between the homologous chromosomes break down, kinetochores pull the homologous chromosomes towards opposite poles
    - Telophase and cytokinesis – cells split apart forming two daughter cells
- **Meiosis II**: Sister chromatids separate
    - Prophase II – nuclear envelope breaks down and the spindles reform
    - Metaphase II – chromosomes align along the metaphase plate
    - Anaphase II – sister chromatids (considered individual chromosomes when they separate) move towards opposite poles of the meiotic spindle
    - Telophase II - nuclear envelope forms around each set of chromosomes and cytokinesis occurs, producing four daughter cells, each with a haploid set of chromosomes

Differentiate between meiosis and mitosis.

Type of Reproduction	Sexual	Asexual
Occurs in	Humans, animals, plants, fungi.	All organisms.
Genetically	Different	Identical
Crossing Over	Yes, mixing of chromosomes can occur.	No, crossing over cannot occur.
Definition	A type of cellular reproduction in which the number of chromosomes are reduced by half through the separation of homologous chromosomes, producing two haploid cells.	A process of asexual reproduction in which the cell divides in two producing a replica, with an equal number of chromosomes in each resulting diploid cell.
Pairing of Homologs	Yes	No
Function	Genetic diversity through sexual reproduction.	Cellular reproduction and general growth and repair of the body.
Number of Divisions	2	1
Number of Daughter Cells produced	4 haploid cells	2 diploid cells
Chromosome Number	Reduced by half.	Remains the same.
Steps	(Meiosis 1) Prophase I, Metaphase I, Anaphase I, Telophase I; (Meiosis 2) Prophase II, Metaphase II, Anaphase II and Telophase II.	Prophase, Metaphase, Anaphase, Telophase.
Karyokinesis (division of nucleus)	Occurs in Interphase I.	Occurs in Interphase.
Cytokinesis	Occurs in Telophase I and in Telophase II.	Occurs in Telophase.

Centromeres Split	The centromeres do not separate during anaphase I, but during anaphase II.	The centromeres split during anaphase.
Creates	Sex cells only: female egg cells or male sperm cells.	Makes everything other than sex cells.

Recognise the that karyotype of a standard human cell.

Karyotype: test to identify and evaluate the size, shape, and number of chromosomes in a sample of body cells
The normal human karyotypes contain 22 pairs of autosomal chromosomes and one pair of sex chromosomes (allosomes).
Normal karyotypes for females contain two X chromosomes and are denoted 46,XX; males have both an X and a Y chromosome denoted 46,XY.
Any variation from the standard karyotype may lead to developmental abnormalities.

Recognize the different sexual cycles used by different organisms.

Describe the sources of genetic variation in organisms.

- **Independent Assortment**: Each pair of chromosomes sorts maternal and paternal homologues into daughter cells independently of the other pairs. The daughter cells will be genetically different and can get a different mixture of gametes occurring. The number of combinations possible when chromosomes assort independently into gametes is 2n, where n is the haploid number (humans: n=23).
- **Crossing over**: produces recombinant chromosomes; it combines genes from each parent
    - Prophase I: synapsis and crossing over occur, then homologs move apart slightly
      Chiasmata and attachments between sister chromatids hold homologs together, moves to metaphase I plate
    - Breakdown of proteins holding sister chromatid arms together allows homologs with recombinant chromatids to separate
- **Random fertilisation**: any sperm can fuse with any ovum (unfertilised egg)
    - In humans, each male and each female gamete represents one of ≈8.4 million (2^23) chromosome combinations because of independent assortment.
    - So, fusion with any sperm with any egg will make a zygote with any of 2^23 x 2^23 different combinations!
    - On top of that, there is the variation due to crossing over.

Briefly outline how anomalies in chromosomes number can occur.

- Non-disjunction: failure of chromosome to separate during meiosis, leads to significant abnormalities in genotype
- Members of chromosome pair fail to separate, hence move to the same pole during Anaphase I
- Chromatids fail to move to different poles during Anaphase II
- If non-disjunction occurs; fetus will be unviable, may have miscarriages
- Non-disjunction gives rise to anomalies in chromosome number.
    - Eg. Trisomy 21 (Down syndrome): extra chromosome 21, but the individual with down syndrome is still healthy and normal

Use your knowledge of the above points to make predictions of possible gametes that could be produced by different cells.

Different Modes of Inheritance

Describe the unique patterns of inheritance associated with X-linked or (sex-linked) genes

- For a recessive X-linked trait to be expressed:
    - A female needs two copies of the allele, male needs only one copy of the allele
    - Therefore, males are *hemizygous* for the X chromosome

Explain how X inactivation can lead to mosaic phenotypes using the example of the tortoiseshell cat

- In mammalian females, one of the two X chromosomes in **each** cell is randomly inactivated during embryonic development.
- The inactive X condenses into a **Barr body**
    - **Barr body**: The inactive X in each cell of a female and condenses into a compact object which lies along the inside of the nuclear envelope. Most of the genes of the X-chromosome that forms the Barr body are not expressed. In the ovaries, however, Barr-body chromosomes are reactivated in the cells that give rise to eggs, such that following meiosis every female gamete (egg) has an active X.
- If a female is heterozygous for a gene located on the X chromosome, she will be a mosaic for that character.
- **Tortoiseshell cat:** the tortoiseshell gene is on the X chromosome, and the tortoiseshell phenotype requires the presence of two different alleles – one orange, one black.
    - Normally only females can have both alleles because they have two X chromosomes.
    - If a female cat is heterozygous for the tortoiseshell gene, she is tortoiseshell.
        - Orange patches are formed by populations of cells where the X chromosome with the orange allele is active.
        - Black patches have cells where the X chromosome with the black allele is active.

Distinguish between incomplete dominance and codominance

- **Incomplete dominance**: when neither allele is completely dominant
    - F1 hybrids have a phenotype somewhere between those two parental varieties
    - E.g. When red snapdragons are crossed with white snapdragons; all the F1 hybrids have pink flowers.
- **Codominance**: the alleles of a gene pair in a heterozygote are fully expressed
    - Results in offspring with a phenotype that is neither dominant nor recessive
    - E.g. ABO blood group system

Distinguish between dominance and epistasis

- **Dominance:** Where one allele of a gene is expressed over another allele of the same locus
- **Epistasis:** A gene at one locus alters the phenotypic expression of a gene at a second locus
    - E.g. in Labrador retrievers, the coat colour depends on two genes:
        - One gene determines pigment colour
        - Other gene determines whether the pigment will be deposited in the hair

Use a pedigree chart to determine the mode of inheritance

Distinguish between the terms monogenic, polygenic and multifactorial

- Monogenic: controlled by a single gene
- Polygenic: controlled by two or more genes; broad norm of reaction
    - Quantitative variation usually indicates polygenic inheritance
    - E.g. skin colour in humans
    - recent study using genomic methods identified at least 180 genes that affected height
- Multifactorial: when genetic and environmental factors collectively influence phenotype

Broadly speaking, explain how phenotype can be influenced by the environment i.e. that certain types of phenotypes (e.g. polygenic traits) are more heavily influenced by environmental factors than others.

- A genotype is generally not associated with a rigidly defined phenotype, but rather with a range of phenotypic possibilities due to environmental influences.

- The **norm of reaction** is the phenotypic range of a genotype influenced by the environment; generally broadest for polygenic characters.
    - E.g. ABO blood groups have no range variation that is dependent on environment, but the number of red blood cells does vary around the norm.
    - E.g. hydrangea flowers of the same genetic variety range in colour from blue-violet to pink, with the shade and intensity of colour depending on the acidity and aluminium content of the soil

Give examples of traits that are heavily affected by environmental factors and traits that are essentially unaffected by the environment.

- Heavily affected by environment
    - Weight – lifestyle choices, diet
    - Height
    - These are both environmental and inherited factors
- Unaffected by environment
    - Blood type
    - Eye colour