Important hints:

- Name and indicate: We write a **direct answer.**
- Why ,how ,which ,explain ,discuss , justify: they depend on **acquired knowledge.** (We write everything we know about this idea.)
- State: logic **consecutive** answer. (or consecutive steps)
- Analysis: **Condition + Result.** (Before the result we can write :"**shows, provokes, or leads to**.)

_Between any two analysis there must be a connector: but, while, in contrast, on the contrary, moreover. (It is forbidden to use '**then'** in the analysis.)

_Here, if we have an increasing or decreasing graph, we have to say by how much it increases or decreases.

_If we are analysing a curve, the condition is x-axis, and the result is the y-axis.

_If there are steps in the experiment, we use: 'Before , upon and after the experiment'.

- Determine, interpret, deduce: **Analysis + Significance.** (Before the significance we can write: "hence, this means, thus, this shows.")

_Here, the **significance** should be from the document.

- Specify: **Significance+ Analysis.**

_Here, the **significance** is from the acquired knowledge.

- Comparison: Same as analysis.
- Conclude: We write a specific conclusion.
- Draw out: It is like conclusion but more general.
- Describe: We translate the scene as it is without adding or removing information. Connectors used in description are:" then, after that, finally, result is". (It is forbidden to use the connectors used for analysis in description.)
- Hypothesis: Is a concept that's true 100%.
- Pose a problem: We write a question asking about the idea in the form: (Is……? ; What……?)
- Pick out from the text: We write the information found in the text as it is.
- Verify: Confirm using argument and logical reasoning whether something is true or false.

_Here, if there is a biological expression or word which is not defined in the experiment, we have to define it before we start the verification.

➢ Identify: Same as verify.

Fluctuates:is a word used in analysis when the graph increases then decreases a little bit.

Part One: Reproduction &Genetics:

♣ Chapter One: Basic Mechanics of Sexual Reproduction:

- Reproduction: Is a process that insures continuity of species.(Production of new individuals).

_There are two types of reproduction:

1. Asexual reproduction: It's done by "amitotic division", where the descendants are copies of their parents: no polymorphism & no genetic diversity between them.
2. Sexual reproduction: Is a biological process that needs:

_ The presence of 2 individuals of same species & different sexes.

_ The presence of a functional reproductive systems in the two individuals.

♠ Document 1: Male and female reproductive systems:

➢ Anatomy of male reproductive system: It consists of:

1. Two gonads called testicles: They represent the primary sexual characteristics of the male (They are found **outside** his body).Their role is:

a) Production of male sex cells that are called spermatozoa or sperms.

b) Secretion of male sex hormone that's called 'testosterone', which is responsible for the appearance of the secondary sexual characteristics of male: Morphology, physiology, and behavior.

2. Epididymis: are fine clusters issued from the testes and located on the top of them. Their role is the storage and maturation of the sperms.
3. Spermiduct: Is a fine tubule issued from the epididymis. Its role is the conduction and maturation of sperms.
4. Accessory glands: Are exocrine glands that secrete seminal fluid in the spermiduct. They are: Cowper's glands, Secretory vesicles &Prostate.
5. Urethra: Is a prolongation of the spermiduct and it is located inside the penis. Its role is the discharge of urine & semin.
6. Penis: Is the organ of copulation between male and female.

* Hints:

_The semin consists of the sperms & the seminal fluid.

_Role of seminal fluid: It insures the survival of sperms since it contains nutrients , also it insures the motility of sperms since it contains fructose that provides the sperms energy in the form of ATP.

➢ Anatomy of female reproductive system: It consists of:

1. Two gonads called ovaries: they represent the primary sexual characteristics of the female.(they are found inside her body). Their role:
   a) Production of oocytes (female sex cells).
   b) Secretion of estrogen and progesterone(female sex hormones). **These hormones** are responsible for the appearance of the secondary sexual characteristics of the female: Morphology, physiology & behavior.
2. Fallopian duct or Oviduct: Its role:

   _Reception of oocyte.

   _It is the site of fertilization.

3. Uterus: It is a muscular sac. It is the site of:
   _Implantation of the embryo.
   _ Development of the embryo.
4. Crevix: It is a narrow neck region of uterus .Its role:

_Decreasing the penetration of the sperms to the uterus.

_Preventing partially the infection of the uterus.

5. Vagina: It is the organ of copulation between the male and the female.

♠ Document 2: Diploid and Haploid Cells:

➢ Types of cells concerning the number of chromosomes: Diploid &Haploid cells:

1. Diploid cells: are nucleated body cells that contain two identical sets of homologous chromosomes each pair of chromosomes is homologous (1 paternal and the other is maternal). The number of chromosomes is **2n**. Example: **Somatic cells & germ stem cells.**
2. Haploid cells: they are nucleated body cells that contain set of different chromosomes, represented by **'n'**. Example**: germ cells**.

* Hints:

_The number of chromosomes in the same species is constant.

_If two different species have same number of chromosomes, then they differ in:

a) Quantity of DNA.

b) Number of genes.

c) Location of genes.

➢ Types of chromosomes:

1. Autosomes.
2. Gonosomes (sex chromosomes).

- Karyotyping: Is the arrangement of chromosomes in decreasing order depending on:
  - Position of centromere.
  - Size of chromosomes.
  - Pattern of dark & light bands.
    - By karyotyping we can determine:
      - Number of chromosomes in species.
      - The sex of the cell.
      - If there is abnormality in the number of chromosomes or their structure.
    - Steps of karyotyping:
      1. Take a sample of blood and separate white blood cells & red blood cells by centrifuging.
      2. Culture white blood cells in a medium for their mitosis.
      3. Add chlochicine to stop mitosis at metaphase.
      4. Put the lymphocytes in a hypotonic medium, cells are burst and group of 2n chromosomes are dispersed in water.
      5. Take a drop of water & observe by electromicroscope then stain (color) them and arrange them in descending order.

♠ Document 3: Meiosis:

➢ Cell cycle:

i. Interphase: (G1, S ,G2)

_During G1: The cell grows & develops its organelles.

_During S period: The DNA is replicated (the chromosome with one chromatid becomes with two chromatids)

_During G2: The cell continues growing and developing its organelles.

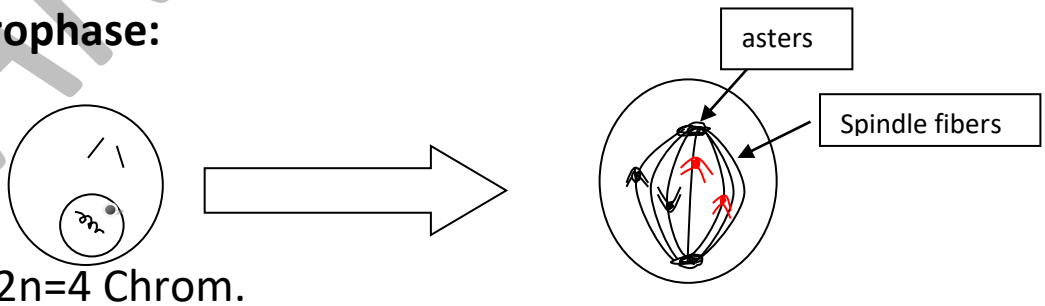
ii. Cell division → a)Mitosis
→ b) Meiosis

a) Mitosis: _ Is an equational division, since the number of chromosomes of the mother cell '2n' is equal to that of daughter cell.

_It is the multiplication of all diploid cells(somatic and germ stem cells).

_It passes in 4 phases:

i. Prophase:

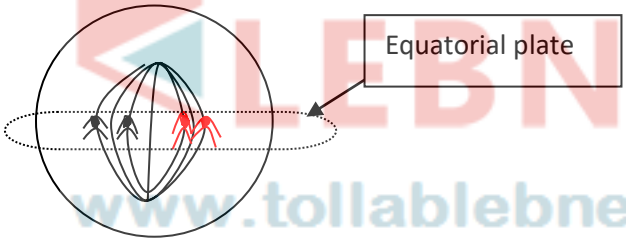


2n=4 Chrom.
QDNA=2 m.a.u.

2n=4chr
QDNA=2m.a.u.

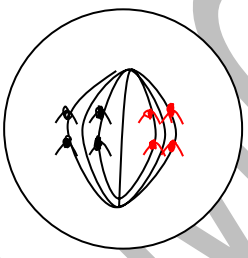
During prophase, chromosomes are dispersed randomly.

ii. **Metaphase:**



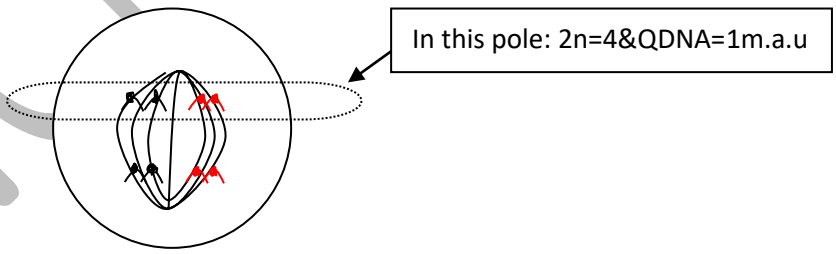
During metaphase, the chromosomes are aligned on equatorial (metaphase) plate.

iii. **Early anaphase:**



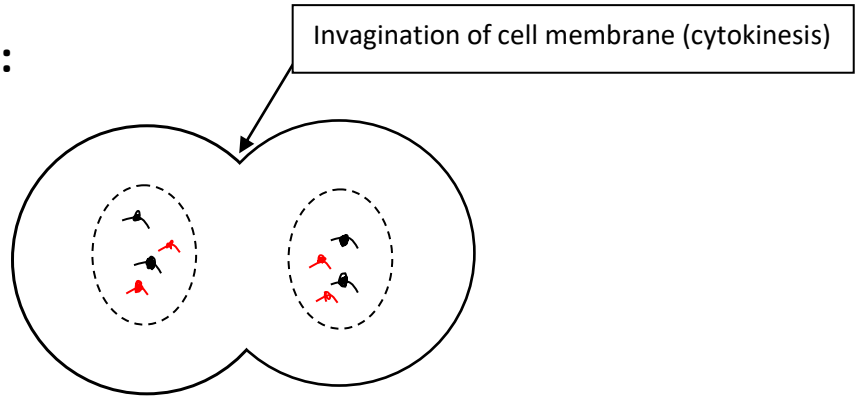
Here, we notice the cleavage of centromere to cut each chromosome into two chromatids.

iv. **Late Anaphase:**

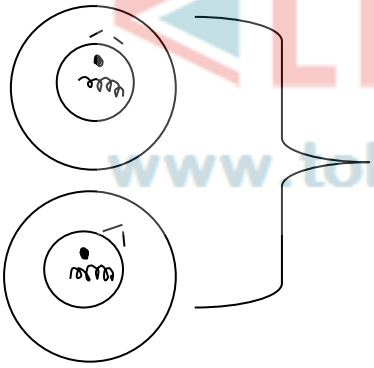


Here, chromosomes make polar ascention & polar formation.

v. **Early Telophase:**



vi. Late Telophase:



Each cell is 2n=4chr. (each with one chromatid),QDNA=1 m.a.u.

Continuity of cell cycle

Nb of divisions

_number of cells produced by mitotic division= 2^n

➢ Advantages of mitosis:

_ Growth of body in size.

_ Degeneration of dead & injured cells.

_ Conservation of DNA.

- Meosis: is a reductional division during which the number of chromosomes of daughter cell(n) is half of that of the mother cell (2n).

_Is the division of only germ stem cells.

_It takes place in testes in man& ovaries in woman.

_It is the production of gametes (oocytes in woman & sperms in man).

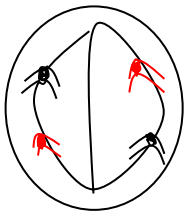
_It passes in 2 successive stages:

i. 1st meotic division: Reductional division.

ii. 2nd meiotic division: Equational division.

i. 1st meiotic division:

P1:

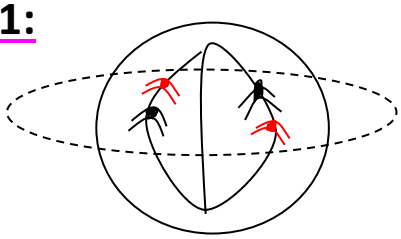


2n=4chr. (2 chromatids)

QDNA=2m.a.u.

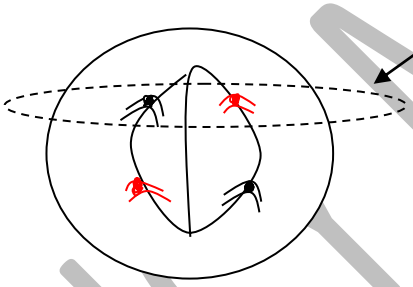
Formation of tetrads

M1:



Tetrads are aligned on metaphase plate

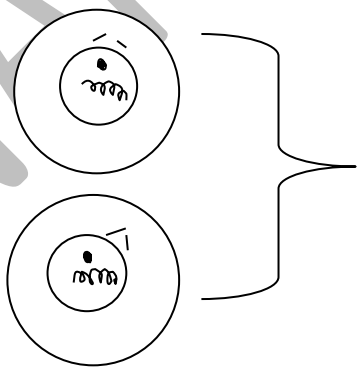
A1:



N=2 chromosomes. (with 2 chromatids).

QDNA=1m.a.u.

T1:

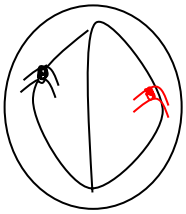


n=2 chromosomes (with two chromatids)

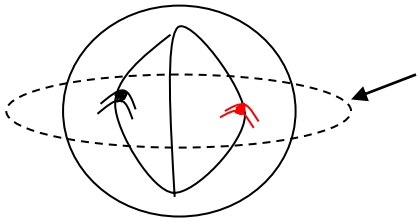
QDNA= 1m.a.u.

ii. 2nd meotic division:

P2:



M2:



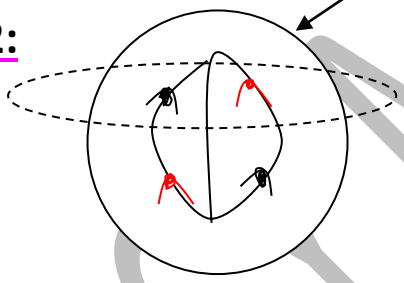
N=2 chr. (with two chromatids)

QDNA=1 m.a.u.

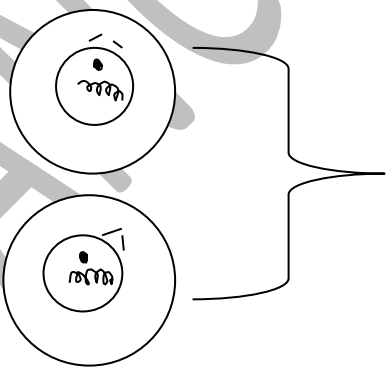
For one pole: n=2chr(with 1 chromatid).

QDNA=1/2m.a.u.

A2:



T2:



n=2 chr(with one chromatid)

QDNA=1/2 m.a.u.

* **Hints:**

This is a simplified example of the meiosis in the human's body, where we take 2n=4chr. & we

suppose an imaginary unit (arbitray unit).ex: 1m.a.u.:micro arbitrary unit.

- Importance of meiosis :

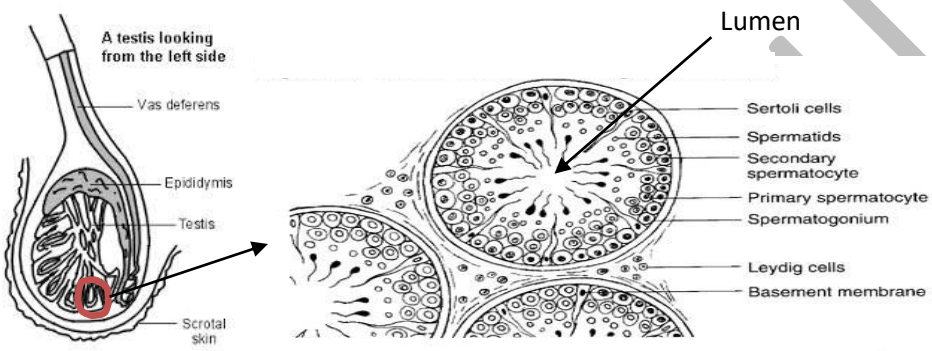
  _ Production of gametes.

  _ Continuity of species.

  * Hint: there is no DNA replication during mitosis or meiosis.

♠ Document 4: Spermatogenisis:

➢ Spermatogenisis: Is the production of male sex cell (sperm). It takes place in testicles in man mainly in fine tubules called seminiferous tubules. It begins at puberty & continues till death.



- Role of Sertoli cells: They support germ cells & conduct sperms to lumen.
- Role of Lydig cells: They secrete testosterone.
  - Spermatogenisis is centripetal: it begins at the level of basal lamina& ends on lumen.
- Stages of Spermatogenisis:

i. Multiplication: During which undifferentiated male germ stem cells "spermatogonium" , reproduce mitotically to produce spermatogonia(2n with 2 chromatids).

ii. Growth period: During which spermatogonia passes in Interphase stage & is transformed into spermatocyte I (2n with 2 chromatids).

iii. Maturation: Corresponds to meioses: where Spermatocyte I(2n with 2 chromatids) produces 2

spermatocyteII(n with 2 chromatids) , that in turn produce 4 spermatids(n with one chromatid).

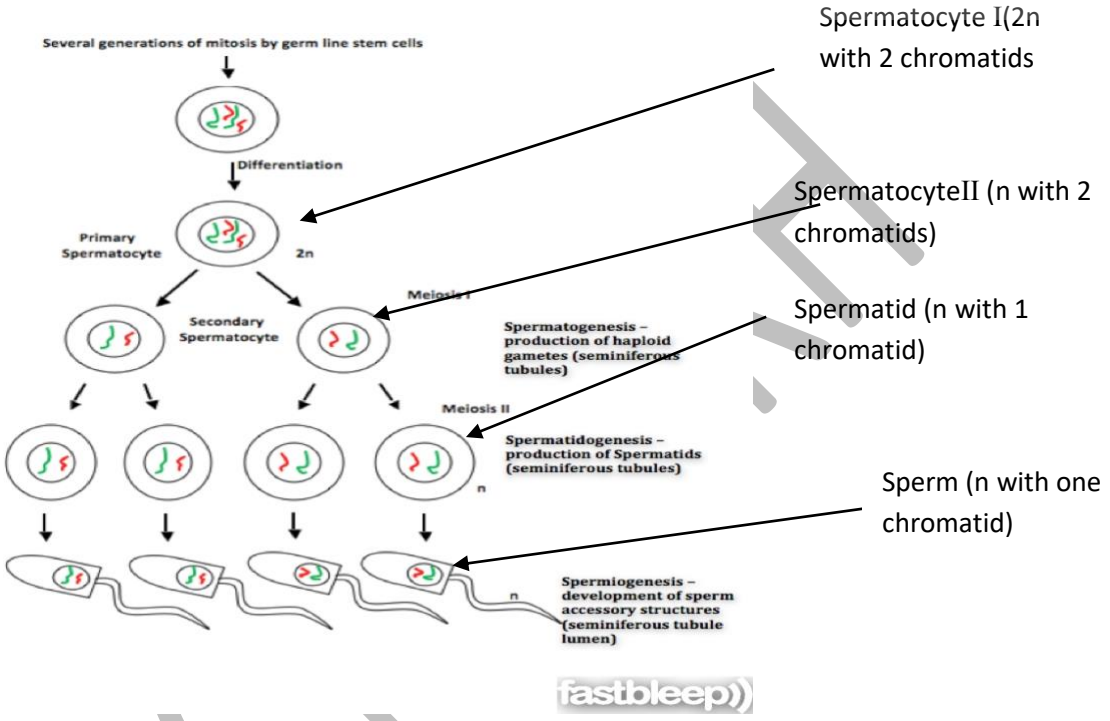
- Process of spermiogenisis: The spherical immobile spermatid is transformed into an elongated and mobile sperm. During which, Golgibody derives Achrosome that occupies the head of the nucleus, and distal centriole derives flagellum. Cytoplasm is restricted to the opposite sides of spiral filaments where mitochondria are found. The residual cytoplasm detaches & degenerates. The result is: an elongated mobile sperm that consists of 3 parts: head, middle part & flagellum.

iv. Differentiation(spermiogenisis): Where spermatids are differentiated into sperms.

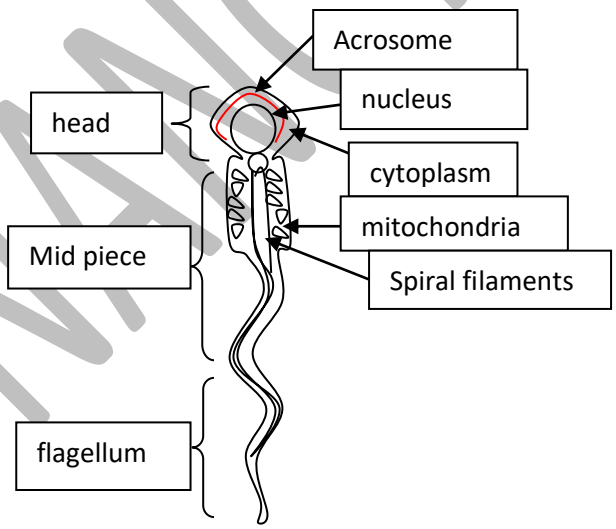
- Process of spermiogenisis: The spherical immobile spermatid is transformed into an elongated and mobile sperm. During which, Golgibody derives Achrosome that occupies the head of the nucleus, and distal centriole derives flagellum. Cytoplasm is restricted to the opposite sides of spiral filaments where mitochondria are found. The residual cytoplasm detaches & degenerates. The result is: an

elongated mobile sperm that consists of 3 parts: head, middle part & flagellum.

Simplified diagram on Spermatogenisis:



Sperm:



Roles of:

1. Flagellum: It orients the sperm towards oocyte during fertilization.
2. Mitochondria: Supplies sperm with energy in the form of ATP for its motility during fertilization.
3. Achrosome: it contains digestive enzymes that digests the membrane of oocyte allowing the penetration of sperm to it during fertilization.

♠ Document 5: Oogenesis:

➢ Oogenesis:

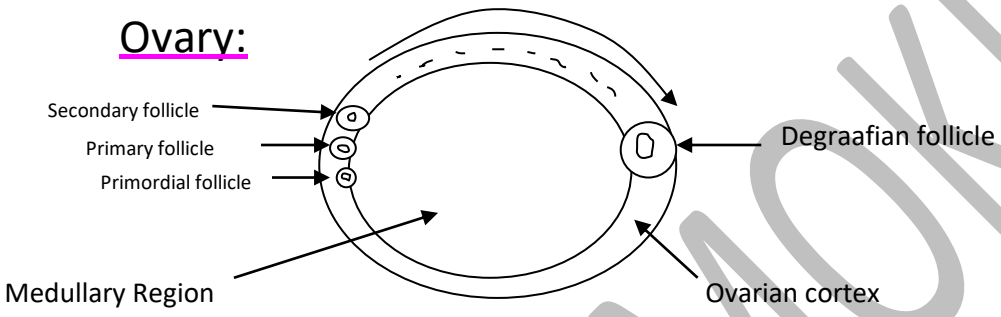
_Is the production of female gamete called oocyte.

_It is cyclic (discontinuous).

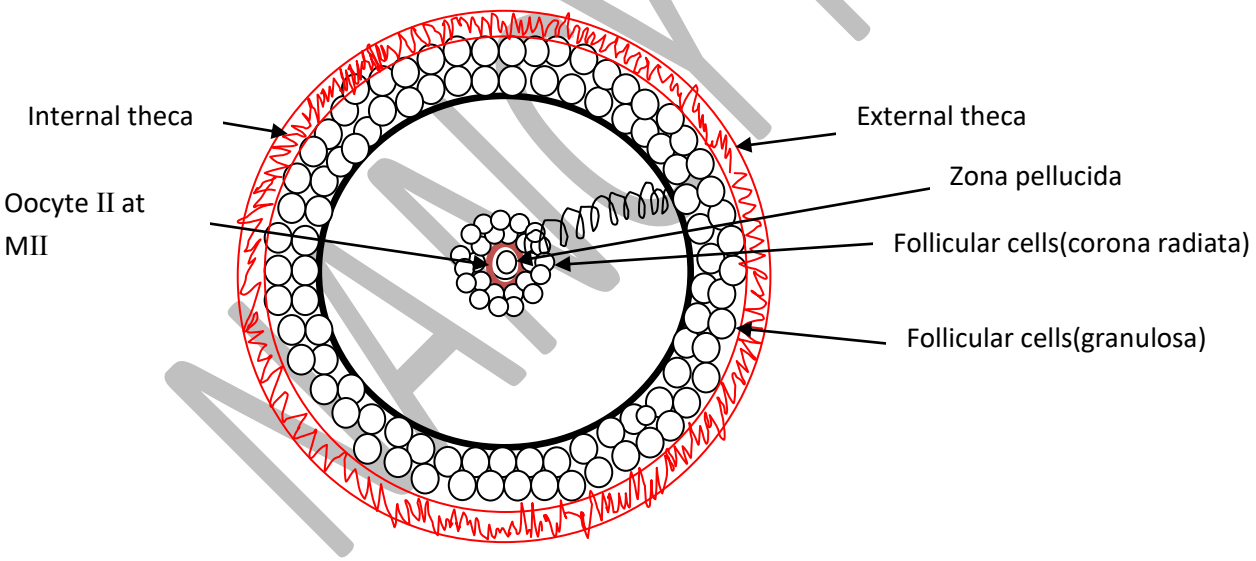
_It is interrupted by gestation & blocked at menopause.

_It starts at the embryonic life of the girl.

Ovary:



Degraafian follicle:



* Follicle: is a sac that surround & protects female gamete.

➢ Stages of oogenesis:

1. Multiplication: During which germ stem cell of female (oogonium) reproduces mitotically to produce oogonia 2n(with one chromatid) .It happens during embryonic life of the girl.(number of oogonia produced =700x10^6).
2. Growth period: It happens during the embryonic life during which DNA is replicated in oogonia to become oocyte I.Also, follicles are developed (folliculogenesis).Furthermore, follicular atresia is done which leads to degeneration of millions of follicles.(nb of rest follicles=6 million).
3. Maturation period: before birth oocyte I enters in 1st meiotic division& stops at PI.
   From the day of birth, till the 1st day of sexual cycle; there is another follicular atresia(nb of remained follicles=400,000).
   -During the sexual cycle, about 10 follicles try to develop but only one reaches maturation, synchronized with the continuity of meiosis inside oocyte I, that produces oocyteII & 1st polar body and it stops at MII.
4. Differentiation: There are 2 cases:

1st case:If there is fertilization,sperm stimulates oocyte II at MII to continue its 2nd meiotic division, and it produces 1 ootid and a 2nd polar body ,also the 1st polar body produces two 2nd polar bodies. The three 2nd polar bodies degenerate and the ootid is developed to become ovum which is fertilized to produce the 1st embryonic cells which is called zygote.

2nd case: If there is no fertilization, oocyteII at MII degenerates.

- Sexual or Menstrual Cycle:
  1. Follicular phase or preovulatory phase: (12-14 days):During which primordial follicles , only one reaches maturation , synchronized with the continuity of meiosis of oocyteI at PI that continues its 1st meiosis & produces oocyte II at MII AND 1st polar body at MII.
  2. Ovulation: It is momenterly at day 12(+or _1). During which degraafian follicle is raptured and oocyteII at MII is expulded to the oviduct ,where the granulose stays in the ovarian cortex.

3. Luteal phase or post-ovulatory phase(14-28 days): During which the cells of granulose develop & form a yellow body called corpus luteum, then, at day 21, corpus luteum is regressed to become white body called corpus albicans and then it degenerates, except if there is fertilization where corpus luteum stays few months and its name becomes 'pregnant yellow body', then it is regressed to form white body & then degenerates.
    * **The age of female gamete= age of female herself.**

♠ Document 6: Fertilization:

- Fertilization: Is the union between male gamete(sperm) and the female gamete (oocyte). The result is a zygote (1st embryonic cell).
  Sperm n (1 chromatid)+ oocyte n (1 chromatid) ⟶ Zygote 2n (2 chromatids).
  During fertilization, sperm replicates its DNA from n chromosome with 1 chromatid to n chromosome with 2 chromatids ,same for oocyte, which produces zygote with 2n chromosomes of 2 chromatids.
- Importance of fertilization:
  _ Reestablishment of diploid state from haploid one.
  _Production of new individuals. (continuity of species).
- During fertilization: sperm capacitation occurs , where the sperm acquires its fertilization capacity in the female genital tract.
- Chemotaxis: attraction of sperm to oocyte by a chemical substance released by oocyte.
  - Sperms acquire their motility in epididymis.

- After ejaculation, sperms are ready to fertilize within 48 hours.
- OocyteII at MII stays 24 hours in the oviduct & then degenerates.

♣ Chapter two: Transmission of Genes:

- Mode of inheritance: Recessive, dominant ,codominant.

  _ The genetic content of living organism is inside the nucleus.

  _ Allelic character: Is the general hereditary character of the individual.

  _ Allele or allelic trait: It represents a particular trait of the allelic character.

  _ Gene: is the expression of 2 alleles on a pair of homologous chromosomes that corresponds to a specific allelic character.

- _ Monohybridism(pure): Is the cross breeding between 2 pure races that differ in one allelic character. The descendants are called hybrids of F_1.

  _pure DD x pure rr ⟶ Dr 100%(first filial generation)

  _Dr (F_1) x Dr (F_1) ⟶ [D]:75% ; [r]: 25%

  _test cross: Dr (F_1)x rr ⟶ [D]:50% ; [r]:50%

  _Laws of Mendel:

1) Law of homoginity of F_1: All descendants of F_1 have same phenotype & same genotype.
2) Law of heteroginity of F_2: The descendants of F_2 have different phenotype & genotype.
3) Law of segregation(purity) of gametes: The gametes obtained during meiosis contain only 1 allele for each particular allelic character. (Any gamete cannot hold 2 alleles of same allelic character).

- Monohybridism(Codominant or intermediate): Is the cross breeding between 2 individuals of pure races that differ in one allelic character . F_1 obtained is hybrid of different phenotype that carries the alleles of the parents.

  _LL x SS ⟶ LS : F_1

  _LS x LS ⟶ [L]:25% ;[LS]:50% ;[S]:25%

- Lethal genes: Some genes are lethal , they lead to the death of the individual in their homozygous state.

  _If Fa x Fa ⟶ [F]: 100%, then allele 'a' is lethal when homozygous.

- Dihybridism: Is the cross breeding between 2 pure races that differ in 2 allelic characters. The descendants are dihybrids of F_1.

_It is consisted of 2 cases:

a. Interchromosomic assortment: If the two genes are autosomal (independent).
b. Intrachromosomic assortment: If the two genes are linked on a pair of homologous chromosomes.

a. Interchromosomic assortment: Where the genes are linked on two different chromosomes(Bidominant-birecessive inheritance):3 cases:

1) Bidominant x birecessive $\longrightarrow$ F_1 :hybrid
2) F_1 x F_1 $\longrightarrow$ $\frac{9}{16}$Bidominant;$\frac{3}{16}$; $\frac{3}{16}$; $\frac{1}{16}$birecessive.
3) Test cross: F_1 x birecessive $\longrightarrow$ 4 equal phenotypes.

b. Intrachromosomic assortment: where the two genes are linked on a pair of homologous chromosomes.

_Linkage can be:

1) Absolute linkage: If F_1 x birecessive ⟶ 2 equal phenotypes:It is of 2 forms:

_Cis-form : if the phenotypes of the descendants are like their parents.

_Trans-form: if the phenotypes of the descendants are new recombined.

2) Partial Linkage: F_1 x birecessive ⟶ 4 different phenotypes.It has 2 forms:

_Cis-form: If the sum of parental phenotypes>sum of new recombined ones.

_Trans-form:If the sum of parental phenotypes< sum of new recombined ones.

- To discuss Intrachromosomic assortment:

i. Test cross of birecessive.

ii. Result is not 4 equal phenotypes: alleles are not independent.

iii. Result is 4 different phenotypes: partial linkage followed by crossing over in meiosis of cis-or of trans -form.

* In female F_1 , the linkage can be absolute or partial one.

* In male F_1 , the linkage is always absolute.

_% of C.O.R=$\frac{sum\ of\ 2=phenotypes(smallest\ ones)}{sum\ of\ all\ phenotypes}$x100

_1% of C.O.R ⟶ 1 CM (scale)

- How to bring phenotypic proportions:

i. We divide all the values by the smallest value. The obtained number is called 'ratio'.

ii. We sum the ratios, then we divide each ratio by the sum of ratios, the number obtained is called phenotypic proportion.

♣ Chapter 3:Genetic variation & polymorphism:

- Usually, genetic variation in sexually reproducing species has 3 causes:
  1. Behavior of the chromosomes during meiosis.
  2. Fertilization.
  3. Mutation.

♠ Document 1: Mutations in the environment :

- ➢ Population: living species that are adapted with the conditions of the area living in.
- ➢ Mutation: Is a sudden change in the nucleotide sequence of DNA. It is spontaneous and capable of producing new phenotypes from already existing ones. It can be induced experimentally.
- ➢ Causes of mutations:

a. Errors in DNA replication during cell cycle.
b. Stressful environmental factors (pollution,…)

* Mutations are inherited only if they occur in germ stem cells since these cells are responsible for the production of gametes by meiosis, so, gametes produced will be mutated, so the body cells of the embryo will be mutated especially germ cells and so on……..
* Environment affects phenotype.
* Hemoglobin is a protein which consists of 2 chains: α-hemoglobin &β-hemoglobin.

♠ Document 2:Mutations and multiple alleles:

- Gene: Is a coded fragment of double strand of DNA.
- DNA(Deoxy ribo-Nucleic Acid): Is a double strand of nucleotide sequence.
- Nucleotide: is the chemical building unit of DNA , it is composed of 3 chemical combined elements:
  1. Phosphoric acid.
  2. Deoxy-ribose sugar.
  3. One of the four nitrogeneous bases (Adenine, Guanine, Cytosine, Thymine).

- Kinds of DNA concerning their function:
  1. Coded DNA: It is a functional DNA that represents the gene.
  2. Uncoded DNA: It is not functional DNA.
- RNA(Ribo-nucliec acid): Is a monostrand of nucleotide sequence.
- Nucleotide: Is the chemical building unit of RNA, it is composed of 3 combined elements:
  1. Phosphoric acid.

2. Ribose sugar.
3. One of the 4 nitrogeneous bases (Adenine, Guanine, Cytosine, Uracil).

➢ Table showing the differences between DNA &RNA:

DNA	RNA
1. Double-stranded	1. Mono-stranded
2. Stable	2. Unstable
3. Spiral	3. Linear or folded
4. Thymine	4. Uracil
5. Deoxy ribo-nucleic acid	5. Ribo-nucleic acid

➢ Every gene in DNA is responsible of the synthesis of particular protein, that is responsible for the appearance of specific phenotype “allelic trait”.

➢ The synthesis of protein passes in 2 stages:

1) Transcription
2) Translation

1) Transcription: It is done in the nucleus of the cell. It is the synthesis of mRNA from a copy of gene (DNA)

where RNA polymerase is involved to unwind 2 complementary strands.

2) Translation: it passes in 3 stages "Initiation, Elongation, Termination".During which a protein is synthesized from mRNA using tRNA, ribosome, RER.

➢ RNA is of 3 types:

i) mRNA: messenger RNA, it is linear and found inside the nucleus .It carries a nuclear message in the form of nucleotides.

ii) tRNA: it is folded and found in the cytoplasm. It transforms nuclear message into sequence of amino acids. It has two sites : (Amino-acid attachment site, Anti-codon site).

iii) rRNA: ribosomal RNA found in the ribosome.

➢ We have 2 types of mutations:

1) Genetic mutation:
   - Substitution or Replacement.
   - Insertion or Addition
   - Deletion or Elimination

2) Chromosomal mutation.

1) Genetic mutations(point mutations):

a) Substitution or Replacement: During which a nucleotide of base … is replaced by another one of base….

_Consequences of this mutation:

i) Change in the nucleotide sequence of DNA.
ii) Change in the genetic codon sequence of mRNA.
iii) Change in the amino acid sequence.
iv) Production of abnormal protein that leads to the appearance of abnormal phenotype.

➢ Silent Mutation: is a substitution mutation that occurs without change in the phenotype.

_Its consequences:

i) Change in the nucleotide sequence of DNA.
ii) Change in the genetic codon sequence of mRNA.
iii) No change in the sequence of amino acids.

iv) Production of a functional normal protein, which leads to the appearance of normal phenotype.

b) Deletion or Elimination: During which a nucleotide of base…. Is eliminated.
It has the same consequences of Substitution mutation (not silent one).

c) Insertion or Addition: During which a new nucleotide of base… is added between the 1st nucleotide of base… and the 2nd one of base ….(nucleotide's number can be any number not obligatory to be 1st & 2nd).

- Gene: is a version of 2 alleles in human and of multiple alleles in population.
- Blood group:
  - Allele A is responsible for the synthesis of transferase E_A .
  - N-galactose Amine+H $\xrightarrow{E_A}$ Antigen A on RBC.
  - Allele B is responsible for the synthesis of transferase E_B.
  - Galactose sugar +H $\xrightarrow{E_B}$ Antigen B on RBC.

- Allele O is responsible for the synthesis of un functional enzyme called E_O.
- H $\xrightarrow{E_O}$ substance H on RBC.

➢ The blood group of an individual is represented by the presence or absence of the antigens A, B, or AB on the RBCs.

➢ Ancestral DNA ⟶ Ancestral gene of blood group ⟶ it gives:

by mutation by substitution ⟶ Allele A

by mutation by substitution ⟶ Allele B

by mutation by deletion ⟶ Allele C

♠ Documents 3:Polymorphic genes in population:

➢ Genetic Polymorphism: is a version of many functional alleles of the same gene.

- Example1: Gene of blood group in population is of 3 alleles : A, B&O.
- Example 2: genes of MHC & HLA.
  - MHC: Major Histocompatibility complex.
  - HLA: Human Leukocyte antigen.
  - MHC genes are co-dominant.
  - There are 6 genes for MHC on pair of chromosomes number 6 .These 6 genes have 6 loci: A, C, B, Dp, DQ, DR.
  - These 6 genes are involved in graft rejection between the donor and the recipient.

- Grafting: It is the implantation of an organ or a tissue between 2 individuals of same species.
- There are 3 types of grafting:

1) Autograft :it is done between the person and himself(it is accepted).

2) Isograft: It is done between 2 individuals having same strain "same MHC genes" usually 2 identical twins. (It is accepted).
3) Allograft: it is done between 2 individuals having different strains. (it is rejected).

- Proteins: They are as: hormones, enzymes, antigens, antibodies, neurotransmitters, co-enzymes & some vitamins.
- Locus: Is the site of the gene on the chromosome.
- Polymorphic gene: is a gene that has many alleles in population.
- Wild type allele: is the allele that corresponds to most common phenotype in population.
    * Note that: If the mutation happened at the level of uncoded DNA, it has no effect.

♠ Document 4:Detection of Genetic Polymorphism:

- We can detect or distinguish the genetic polymorphism by:
  i) Analysis of phenotypic variations.
  ii) RFLP: recognition fragment length polymorphism.

➢ Gel electrophoresis: Is the separation of coded and uncoded fragment of DNA molecule electrically in a gel.

➢ Restriction enzyme: Is a biological enzyme produced by bacteria. There are around 100 different restriction enzymes in nature. The restriction enzyme cuts DNA in to fragments of different sizes.

➢ Recognition site: it is composed of different number of nucleotide sequence with particular order, it ranges between 4 & 8.

➢ How to determine genetic polymorphism by gel electrophoresis: (by RFLP analysis):

1) Take DNA of 2 individuals X &Y.
2) Add same restriction enzyme to both nuclei, so that it will cut their DNA into different sizes that are charged negatively.

3) Put them on gel electrophoresis that is connected to electric current, thus, fragments of DNA will repel each other to the gel in decreasing order of strength of repulsion(negative charge), so we obtain 2 restriction maps on gel of the 2 persons X &Y.
4) Add radioactive substance that corresponds to ethyl bromide.
5) Observation by radioactivity of the different number & sizes of DNA which proves the genetic polymorphism.
    - Restriction map: is the arrangement of the DNA fragments of one person on Gel elecrophoresis in decreasing order. It is independent of gene function since it is the arrangement of coded and uncoded DNA.
    - Upon mutation:
        1) If the mutation occurs in the recognition site of the restriction enzyme, then the restriction enzyme will not recognize this site,

leading to the decrease in the amount of fragments obtained.

2) If the mutation occurs in a place rather than the recognition site, them there will be no change in the restriction map.

♠ Document5:Genetic Identity of individuals:

➢ Localization of a gene on a chromosome:

We determine it by a technique called FISH (fluorescent in sito hybridization): It is done by:

1) Taking DNA from the individual.
2) Denaturation: separation of chromatids of chromosome that is separation partially of the 2 chromatids of each chromosome.
3) Hybridzation: add DNA probe, that is fluorescent & complement to the natural DNA of gene which binds into its natural strand of DNA, and then observe by autoradiography.

➢ DNA finger print: It is detected by a technique called **Southern Blotting Technique** (Gel electrophoresis +FISH).Its steps:

1) Add restriction enzyme to DNA, it cuts it into fragments of different sizes that are charged negatively.
2) Place DNA on Gel electrophoresis that is connected to an electric current, leading to the repulsion of

DNA fragments depending on their strength of repulsion.

3) We obtain a restriction map of the individual.
4) Put a filter paper on the gel of Gel electrophoresis where DNA fragments are found. (Blotting :transfer of restriction map from Gel electrophoresis to the filter paper).
5) Denaturation.
6) Hybridzation.
7) Wash the filter paper so that uncoded DNA fragments are eliminated.
8) By autoradiography, we observe the alleles of the gene that is called DNA finger print.

- DNA probe: is a fluorescent synthetic DNA mono-strand that is complement to a natural one.

    * For the same family, at least one band of the child should be the same with his father or with mother's DNA finger print.

♣ Chapter 5:Human Genetics:

It is the study of inheritance as it occurs in human beings.

This chapter is important, it will teach you how to treat with the pedigree, about mode of inheritance, and many other things.

♠ Document 1: Inheritance of Genetic Traits:

➢ The aim of constructing a pedigree:

1) To determine the mode of inheritance of a specific allelic trait.
2) To determine the probable or the accurate genotype of the individuals in the pedigree.
3) To estimate the probability for a couple to have child with a certain hereditary trait(genetic risk).

* Consanguineous mating: is the union between two individuals who are related as second cousins or closer.

♠ Documents 2&3: Autosomal diseases &Sex-linked diseases:

- The appearance of any hereditary trait (phenotype) needs 2 copies of the gene that is 2 alleles as a genotype.

Example: AA phenotype: [A]
Aa phenotype: [A]
aa phenotype: [a]

_These information before, is in the case when the gene is autosomal.

Exception: If the gene is sex-linked: There are 3 cases:

i. On non-homologous segment of X chromosome:

_The female needs 2 copies of the gene for the appearance of the phenotype.

_The male needs only 1 copy of the gene for the appearance of the phenotype.

ii. On the non homologous segments of X&Y chromosomes:

Both couples (man & woman) need 2 copies of the gene for the appearance of the phenotype, it is like autosomal genes.

iii. On non homologous segment of Y chromosome:

It is exclusive for males, the male needs only 1 copy of the gene for the appearance of the phenotype.

➢ Transmission of the alleles from the couples to their children:

Here, take "d" is the gene of the disease studied.

* If the gene is sex-linked:

1) If the gene of the disease "d" is located on the non-homologous segment of X chromosome, (X^d), then:

_ The father inherits X^d only to his girls.

_ The mother inherits X^d to both children girls & boys.

2) If the gene "d" is located on the homologous segments of X and Y :

_ The father inherits X^d to his girls & Y^d to his boys.

_ The mother inherits X^d to both children girls & boys.

3) If the gene "d" is located on the non homologous segment of Y chromosome: (Y^d):

_In this case, only fathers inherit Y^d to their boys.

- Mode of inheritance of the gene of disease:

Wherever we have couple that are normal which have diseased child, then the gene of disease is recessive masked by the gene of normal.

- Localization of the gene of disease:

It is studied corresponding to the case of the pedigree. (Examples will be given later in another pdfs).

_Note that if by crossing all the male children are like their mother and the female children are like their father then the allele studied is located on the non-homologous segment of X-chromosome.(which is exception found directly without discussion).

_If the gene of the disease is dominant , and we want to discuss its localization, then we take the gene of normal and discuss it 'since treating with

recessive allele is easier ‘ (knowing that gene of normal and of disease studied have the same locus).

_If the disease affects only boys or only girls the it is sex-linked (on non-homologous segment of X or Y), not on homologous segments.

- Genetic probability of a couple to have an affected child:(genetic risk):

Probability of the fetus to be affected= P(mother to be hybrid)x number of gametes produced +P (father to be hybrid)x number of gametes produced.

_Probability of the parent to be hybrid is:

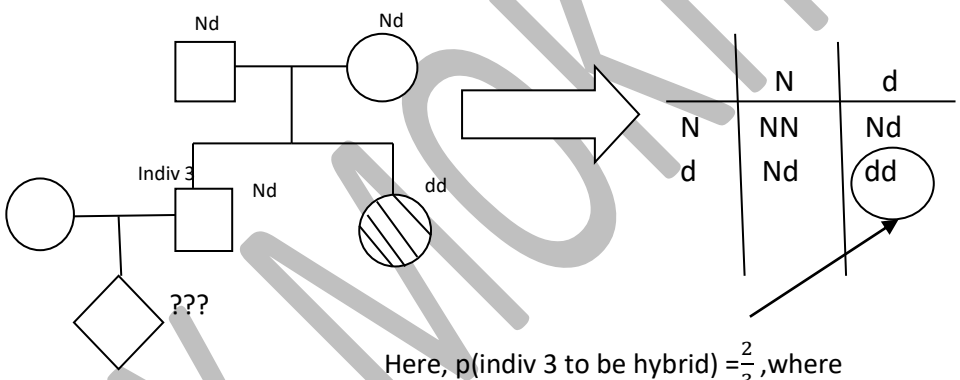
1) If we are sure 100% that s/he is hybrid , we put P(..)=1.
2) If we are sure 100% he is pure, we put P(…) =0.
3) Otherwise, if we don’t know if s/he is hybrid or pure, then:

   _we look on its phenotype.

   _we see his parents by table of cross the proportion of that phenotype, then we t5ake the proportionality of hybridism from

the genotypes corresponding to that genotype.

Example:



Here, p(indiv 3 to be hybrid) $=\frac{2}{3}$,where the normal phenotype is eliminated.

* If the gene is located on the non-homologous segment of X chromosome, and we want to discuss the probability of the fetus, then we say:

_If he is a boy, then he takes…… so P(…)=a

_ If she is a girl, then she takes….. so P(…)=b

But, P(fetus to be a girl)=P(fetus to be a boy)=1/2

So P(fetus to be affected)=$\frac{1}{2}$ x a +$\frac{1}{2}$ x b.

* The consanguineous marriage is the cause of the increase of the disease in a family or an area.
* Chromosomal analysis:

i. If the gene is autosomal, we draw the chromosomes.
ii. If the gene is sex-linked, then we draw X &Y chromosomes.

* Note that: If we have the P(heterozygous)=a in a certain area, and we have the pedigree of this area, so, when we want to take P(hybrid or heterozygous) of an individual of a family from this area, then we look on the family not on the probability given , except if the individual is not from the family studied.

➢ Color blind or Daltonism:

The gene of this disease is recessive and located on the non-homologous segment of X chromosome,X^d.
_partial color blind : the diseased person can't distinguish between colors.

➢ Hemophilia : (A &B);

Its cause is the absence of a protein that causes blood coagulation.

The gene of the disease is recessive and located on the non-homologous segments of X &Y. The allele of the disease is lethal when homozygous.

♠ Document 4: Chromosomal mutations:

➢ Chromosomal mutations are done in:

1) Number of chromosomes(numerical mutation).
2) Structure of chromosomes (structural mutation).

1) Mutation in number:Examples

i. trisomy 21(autosomal abnormality): its cause is the presence of extra copy of chromosome 21 . 2n=47chromosomes=45A+2G.
ii. Turner syndrome(gonosomal abnormality): Its cause is the presence of 1 X in gonosomes. 2n=45chr=44A+X.
iii. Klein-filter syndrome: its cause is the presence of extra X in gonosomes:2n=47chr.=44A+XXY.

2) Mutation in structure: ex:

Cri-du-Chat: its cause is the deletion of a fragment of chromosome number 15.(deficiency in QDNA).

♠ Document 5:Prenatal Diagnosis:

There are 3 methods for prenatal diagnosis:

1) Amniocentesis.
2) Chorionic villus biopsy.
3) Sampling of fetal blood.

Their aim is to obtain a nucleated fetal cell that contains the DNA of the fetus.

There are 3 tests that can be made on this fetal cell that's obtained by 1 of the 3 methods written above:

i. Karyotyping: We can determine by it if there is abnormality in the number and the structure of chromosomes. We can also determine the sex of the embryo.
ii. DNA analysis (DNA finger print): We can determine by it the presence of thee abnormal (mutant) gene that causes the disease.
iii. Biochemical analysis: we can detect by it the presence of the abnormal protein or the deficiency of a certain functional protein.

Best Wishes