Long questions

1. **Write a short note on Genetic counseling-Definition, Objective, Legal and Ethical issues.**

Definition
Genetic counselling is a communication process which deals with human problems associated with the occurrence, or the risk of occurrence of a genetic disorder in a family.

Objective
This process involves an attempt by one or more appropriately trained persons to help the individual or family to
(1) comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management;
(2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
(3) understand the alternatives for dealing with the risk of occurrence;
4) choose the course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, to act in accordance with that decision; and
(5) to make the best possible adjustment to the disorder in an affected family member and/or the risk of recurrence of that disorder.

Ethical guidelines
1) Parental autonomy in abortion choices
(2) Non-directive counselling
(3) PND that must be provided when parents need the information to prepare themselves for the birth of a possibly affected child
(4) Practitioners need to disclose to the consultee the risks and benefits of each procedure in PND;
5) Information of XY females and XX males with great care that casts no ambiguity on the patient's social and phenotypic sexual identity;
(6) In case putative father is not the biological father of the foetus, the mother to be informed first to avoid social problems and she may be left to take final decision
(7) Medical geneticists to decide which of the disorders warrants the options of prenatal diagnosis and termination of pregnancy, and
(8) Consequences from the above to be evaluated in terms of basic ethical principles, and critical tests of what is best for the individuals, groups and society.

2) Define Prenatal Diagnosis and list the risk factors for qualifying a woman for prenatal diagnosis.

Prenatal diagnosis is the diagnosis of a disease or condition in a fetus or embryo before it is born. The aim is to detect birth defects such as neural tube defects, chromosome abnormalities, genetic diseases and other conditions.
Diagnostic prenatal testing can be by invasive methods or non-invasive methods
Aim of prenatal diagnosis:

1.Determines the outcome of pregnancy
2.Counsel and support the parents after diagnosis, for reproductive decisions i.e. Helps couple to decide whether to continue pregnancy

3.Indicates the possible complications during birth process, therefore to prevent or prepare for them:i.e.

a. Prepare the couple psychologically and financially for the birth of a child with an abnormality
b. May be of help in improving the outcome by using fetal treatment.

Risk factors qualifying a pregnant woman for prenatal testing

- Women over the age of 35
- Women who have previously had premature babies or babies with a birth defect, especially heart or genetic problems
- Women who have high blood pressure, lupus, diabetes, asthma, or epilepsy
- Women who have family history or ethnic background prone to genetic disorders, or whose partners have these
- Women who are pregnant with multiples (twins or more)
- Exposure to teratogenic agents

Short questions

Q1)Define Mutation and list types of mutation

In biology, mutations are changes to the base pair sequence of genetic material (either DNA or RNA).
When a mutation alters a protein that plays a critical role in the body, a medical condition can result. A condition caused by mutations in one or more genes is called a genetic disorder.
If a mutation is present in a germ cell, it can give rise to offspring that carries the mutation in all of its cells. This is the case in hereditary diseases.
On the other hand, a mutation can occur in a somatic cell of an organism. Such mutations will be present in all descendants of this cell, and certain mutations can cause the cell to become malignant, and thus cause cancer.

Types of mutations

- Single base substitutions
- Insertions and deletions
- Chromosomal Mutations

1) **Single base substitutions**

By structure

- Point mutations, often caused by chemicals or malfunction of DNA replication, exchange a single nucleotide for another.

- Most common is the transition that exchanges a purine for a purine (A ↔ G) or a pyrimidine for a pyrimidine, (C ↔ T).

- Less common is a transversion, which exchanges a purine for a pyrimidine or a pyrimidine for a purine (C/T ↔ A/G).

By function

- Silent mutations: which code for the same amino acid.
- Missense mutations: which code for a different amino acid.
- Nonsense mutations: which code for a stop and can truncate the protein.

2) **Insertions and deletions**

- Insertions add one or more extra nucleotides into the DNA. They are usually caused by transposable elements, or errors during replication of repeating elements (e.g. AT repeats).
- Insertions in the coding region of a gene may alter splicing of the mRNA (splice site mutation), or cause a shift in the reading frame (frameshift), both of which can significantly alter the gene product..
- Deletions remove one or more nucleotides from the DNA. Like insertions, these mutations can alter the reading frame of the gene. They are irreversible.

3) **Chromosomal Mutations**

A chromosome mutation is any change in the structure or arrangement of the chromosomes.

- Translocations - Translocations are the transfer of a piece of one chromosome to a non-homologous chromosome.

- Inversion - A region of DNA on the chromosome can flip its orientation with respect to the rest of the chromosome.

- Deletions - A large section of a chromosome can be deleted resulting in the loss of a number of genes.

- Duplications - In this mutation, some genes are duplicated and displayed twice on the same chromosome.

- Chromosome non-disjunction - During cell division, the chromosomes fail to successfully separate to opposite poles, resulting in one of the daughter cells having an extra chromosome and the other daughter cell lacking one.

Q2) **Role of a nurse in Genetics**

Genetics nurses play a vital role in the facilitation of information and the fault of a lagging education system lies in the hands of an ill prepared society.

- A genetics nurse identifies genetic risk factors, holds nursing interventions, and relays data to the individual or family seeking genetics information.
- An advanced practice genetics nurse further fulfills these responsibilities through the provision of genetic counseling and case management for those with, or at risk, for a disease resulting from a genetic susceptibility.

Nurses in genetics work with patients and their families in many settings such as:

- Specialty clinics where gene-based diagnoses and therapies are offered
- Prenatal and reproductive technology centers
- Cancer centers
- Primary healthcare settings
- Pediatric clinics
- Industrial health
- School health
- Research centers, Biotech and insurance industries

Q3) **Define any two birth defects with examples**

- Malformation
- Deformation
- Disruption
- Dysplasia

Malformation

A malformation is a primary structural defect of an organ,or part of an organ,which results from an inherent abnormality in development.
E.g. congenital heart abnormalities,neural tube defects,isolated defects show multifactorial inheritance.
Multiple malformations are due to chromosomal abnormalities

Disruption
Is an abnormal structure of organ as a result of external factors disturbing the normal developmental process.
E.g. disruption of limb development when a band of amnion becomes entwined around baby's forearm or digits

Deformation
Is a defect which results from an abnormal mechanical force which distorts an otherwise normal structure
e,.g. dislocation of the hip and positional talipes which can be caused due to lack of amniotic fluid or intra-uterine crowding.
Dysplasia
Is an abnormal organisation of cells into tissue.
E.g. in skeletal dysplasia such as thanatropic dysplasia.

1.In DNA adenine normally pairs with:

a) cytosine.

b) guanine.

c) thymine.

d) uracil.

2.In meiosis, recombination occurs in:

a) Metaphase I.

b) Prophase I.

c) Metaphase II.

d) Prophase II.

3.Which of the following is an example of monosomy?

a) 46,XX

b) 47,XXX

c) 69,XYY

d) 45,X

4.Consanguinity shows a strong association with which pattern of inheritance?

a) Autosomal dominant

b) Autosomal recessive

c) X-linked dominant

d) X-linked recessive

5.Which of the following disorder does not show X-linked inheritance?

a) Duchenne muscular dystrophy

b) Tay-Sachs disease

c) Haemophilia A

d) Haemophilia B

6.DNA replication during the cell cycle takes place in the

a)G1 phase

b)G2 phase

c)S phase

d)M phase

7. The process of conversion of an RNA into a functional protein is called

a)Transcription

b)Translation

c) Mutation

d)Polymerisation

8.The codon is a set of

a)RNA

b)Nucleotides

c) Amino acids

d) 3 Nitrogen bases

9.An autosomal dominant disease follows ALL EXCEPT

a) Vertical Inheritance pattern

b) A structural protein defect

c) Recurrence in every pregnancy is 50%

d) The parents are carriers

10.A couple may have a child affected with a genetic disorder

a)Advanced maternal age

b)Consanguinity

c)History of Teratogen

d)All of these

Ans-1-c,2-b,3-d,4-b,5-b,6-c,7-b,8-d,9-d,10-d

1.In RNA adenine normally pairs with:

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d) uracil.

2.In meiosis, recombination occurs in:

a) Metaphase I.

b) Prophase I.

c) Metaphase II.

d) Prophase II.

3.Which of the following condition exhibits trisomy?

a) Down syndrome

b) Potter syndrome

c) Turner syndrome

d) Marfan syndrome

4. Affected males and carrier females are seen in which inheritance pattern?

a) Autosomal dominant

b) Autosomal recessive

c) Mitochondrial

d) X-linked recessive

5. Which of the following disorders shows Autosomal recessive inheritance?

a) Duchenne muscular dystrophy

b) Sickle cell anaemia

c) Haemophilia A

d) Huntingtons disease

6.Which essential vitamin supplement is recommended to prevent neural tube defects

a) Folic acid

b) Vit C

c) Biotin

d) Vit A

7. The process of conversion of DNA into a messenger RNA is called

a)Transcription

b)Translation

c) Mutation

d)Polymerisation

8.The following methods are used for pre- natal diagnosis

a)Amniocentesis

b)Chorion villus biopsy

c) Maternal serum screening

d) All of the above

9.An autosomal recessive disease follows ALL EXCEPT

a) Horizontal Inheritance pattern

b) An enzymatic protein defect

c) Recurrence in every pregnancy is 25%

d) One parent is affected

10Chromosome Y is a ---------- chromosome

a)Metacentric

b)Sub metacentric

c)Acrocentric

d)Telocentric

Ans-1-d,2-b,3-a,4-d,5-b,6-a,7-a,8-d,9-d,10-c