

15. To study various syndromes and their karyotypes in human beings.

Date : / /

Aim :- To study the various syndromes and their karyotypes.

Principle :-

Karyotype is the name given to the whole group of characteristics that allow the identification of particular chromosome set. It includes the number and size of chromosomes that are constant for a species. Chromosomes are arranged in a series of their decreasing lengths representing what is called **idiogram**. The characteristics of chromosome in making idiogram are - the number, length, position of centromere, secondary constrictions, length of chromosome arms, arm ratio, banding pattern, etc.

The position of centromere is different in different chromosomes but it is always constant for a particular chromosome. You have already studied four different shapes of chromosomes based on the position of centromere.

The idiogram is arrangement of chromosomes according to their decreasing size, keeping position of centromeres aligned. The sex chromosomes are always shown at the end.

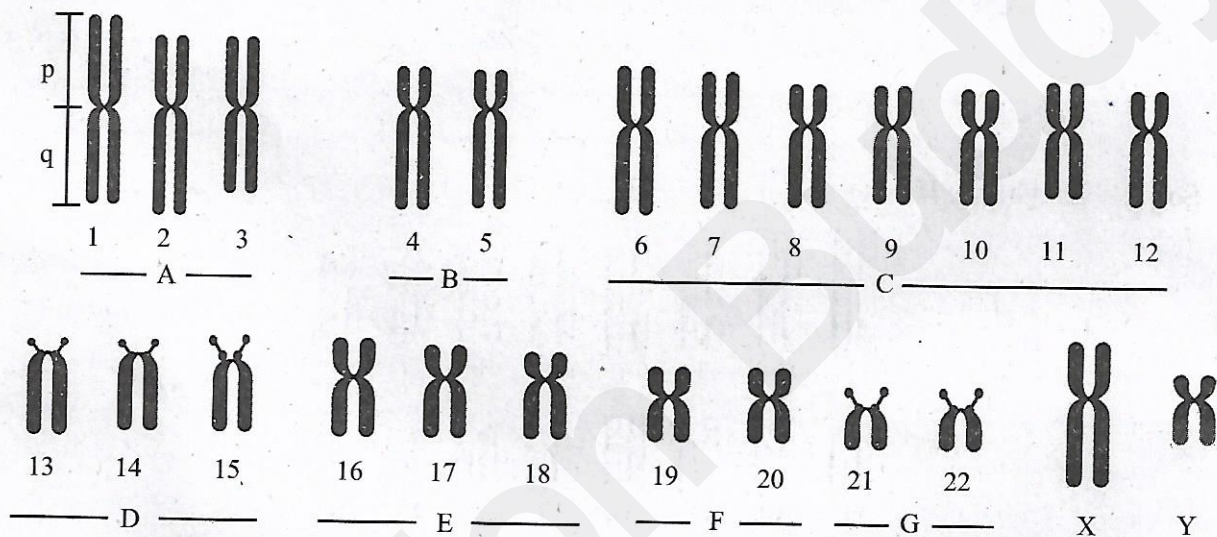


Figure :-

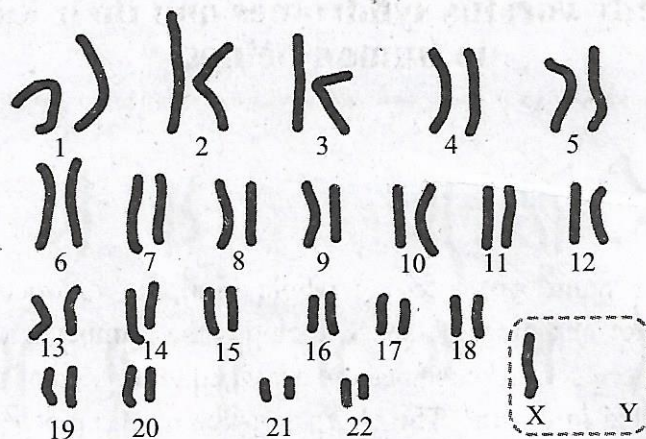
Procedure :-

A normal human being shows 46 chromosomes (23 pairs) out of which first 44 chromosomes i.e. 22 pairs are of autosomes and the last 2 chromosomes i.e. 23rd pair represents sex chromosomes. The first 22 pairs are arranged according to their size in descending order. In humans, chromosomes are classified into 7 groups viz. A, B, C, D, E, F and G. Sex chromosomes are placed at the end on the right side of group 'G'. Human karyotype is represented in the above diagram.

The karyotype is advantageous to study and diagnose different types of syndromes. Syndromes are structural or numerical abnormalities of human chromosomes

Observe the given idiograms and comment on syndrome.

1. Sample karyotype I :-



Chromosomal condition or syndrome (if any) : **Turner's syndrome in male.**

Symptoms : **Short stature (Height) and webbed neck,**

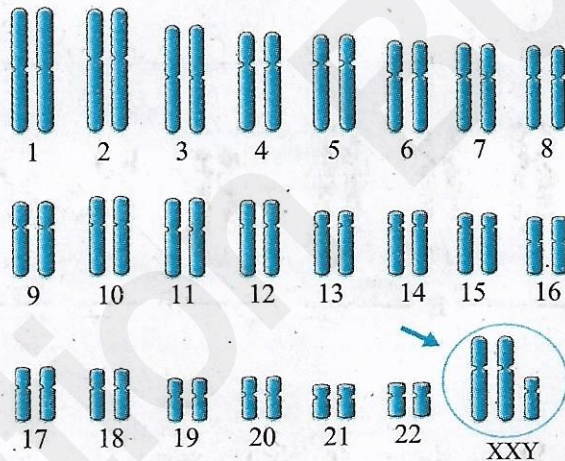
lower posterior Hair line,

Broad shield shaped chest

poorly developed ovaries and breast

low intelligence.

2. Sample karyotype II :-



Chromosomal condition or syndrome (if any) : **'Klinefelter's syndrome'**

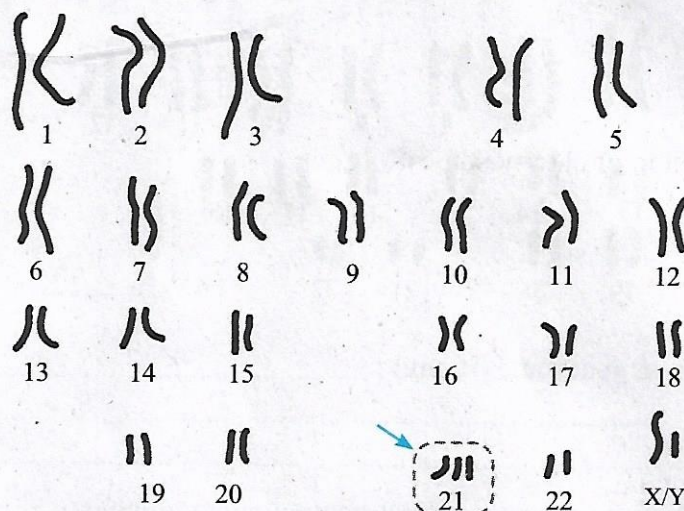
Symptoms : **Harsh voice,**
under developed testis

Tall with long arms,

development of breast(Gynaecomastia)

Sterile.

Sample karyotype III :-



Chromosomal condition or syndrome (if any) : **21 Trisomy (Down's syndrome)**

Symptoms :

- Flattened face, Protruding tongue
- Small head, Short neck, Short height
- Upward slanting eye lids (palpebral fissures)
- Unusually shaped or small ears
- Tiny white spots on the colored part (iris) of the eye called Brushfield's spots
- Poor muscle tone
- Broad, short hands with a single crease in the palm
- Relatively short fingers and small hands and feet

Questions

1. What is trisomy?

A condition in which an extra copy of chromosome is present in the nucleus of cell causing developmental abnormalities.

e.g. 21st Trisomy (Down's syndrome)

2. What is autosome?

The chromosomes which control the somatic characters are known as autosomes.

3. Define monosomy and mention how does it differ from trisomy.

Monosomy- Absence of one chromosome from pair of chromosome.

In trisomy condition extra chromosome is present in diploid chromosomal condition by which three chromosomes are present instead of two chromosomes.

4. What is non-disjunction of chromosomes?

Non-disjunction is the failure of homologous chromosomes to separate properly during cell division.

5. Explain - the karyotype.

Karyotyping is the process of pairing and ordering all the chromosomes of an organism.

Multiple Choice Questions

- Down's syndrome is due to non-disjunction of chromosomes
 - crossing over
 - linkage
 - sex linked inheritance
 - non-disjunction of chromosomes
- Ovum producing Klinefelter's syndrome shall have chromosome number 24
 - 21
 - 22
 - 23
 - 24
- Chromosome number of Down's syndrome or Mongolism is 47
 - 46
 - 44
 - 48
 - 47
- Chromosomes were first seen by Waldeyer
 - Strassburger
 - Hofmeister
 - Flemming
 - Waldeyer
- In human beings, single X / XO abnormality causes Turner's syndrome
 - Down's syndrome
 - Klinefelter's syndrome
 - Turner's syndrome
 - Patau's syndrome

Remark and Signature of Teacher