09-12-21

EXPERIMENT NO. 2

AIM OF THE EXPERIMENT - To study human karyotypes and identify the disease caused.

THEORY-

Karyotyping is the process of pairing and ordering all the chromosomes of an organism, thus providing a genome-wide snapshot of an individual's chromosomes.. Karyotypes are prepared using standardized staining procedures that reveal characteristics structural features for each chromosome. Clinical cytogeneticists analyzed human karyotypes to detect gross-genetic changes – anomalies involving several megabases or more of DNA. Karyotypes can reveal changed in chromosome number associated with aneuploid conditions, such as trisomy 21 (Down Syndrome). Careful analysis of karyotypes can also reveal more subtle structural changes, such as chromosomal deletion, duplications, translocations or inversions. In fact as medical genetics becomes increasingly integrated with clinical medicine. Karyotypes are becoming

a source of diagnostic information for specific birth defects, genetic disorders and even cancers.

Preparing Karyotypes from mitotic cells:

Karyotypes are prepared from mitotic cells that have been arrested in the metaphase or pro-metaphase portion of cell cycle when chromosomes assume their most condensed conformations. A variety of tissue types can be used as a source of these cells. For cancer diagnosis, typical specimens include tumor biopsis or bone marrow samples. For other diagnosis, Karyotypes are often generated from peripheral blood specimen or a skin biopsy. For prenatal diagnosis amniotic fluid or chorionic villi's specimens are used as the source of cells.

The process of generating a Karyotype begins with the short-term culture of cells derived from a specimen. After a period of cell growth and multiplication, dividing cells are arrested in metaphase by addition of Colchicine, which poisons the mitotic spindle. The cells are next treated with hypotonic solution that

that causes their nuclei to swell and the cells to burst. The nuclei are then treated with a chemical fixative, dropped on a glass slide, and treated with various stains that reveal structural features of the chromosomes.

Group	Size and Centromere Position	Ideogram number	Number in Diploid Cell.
A or I	Large; Metacentric/ Sub-metacentric	1-3	6
B or II	Large; Submetacentric	4,5	4
C or III	Medium; Submetacentric	6-12 and X	15 (male) 16 (female).
D or IV	Medium; Acrocentric	13-15	6
E or V	Small; Metacentric/ Submetacentric	16-18	6
F or VI	Small; Metacentric	19-20	4
G or VII	Smallest; Acrocentric	21,22 and Y	5 (male) 4 (female).

Table showing types or classification of human chromosome.

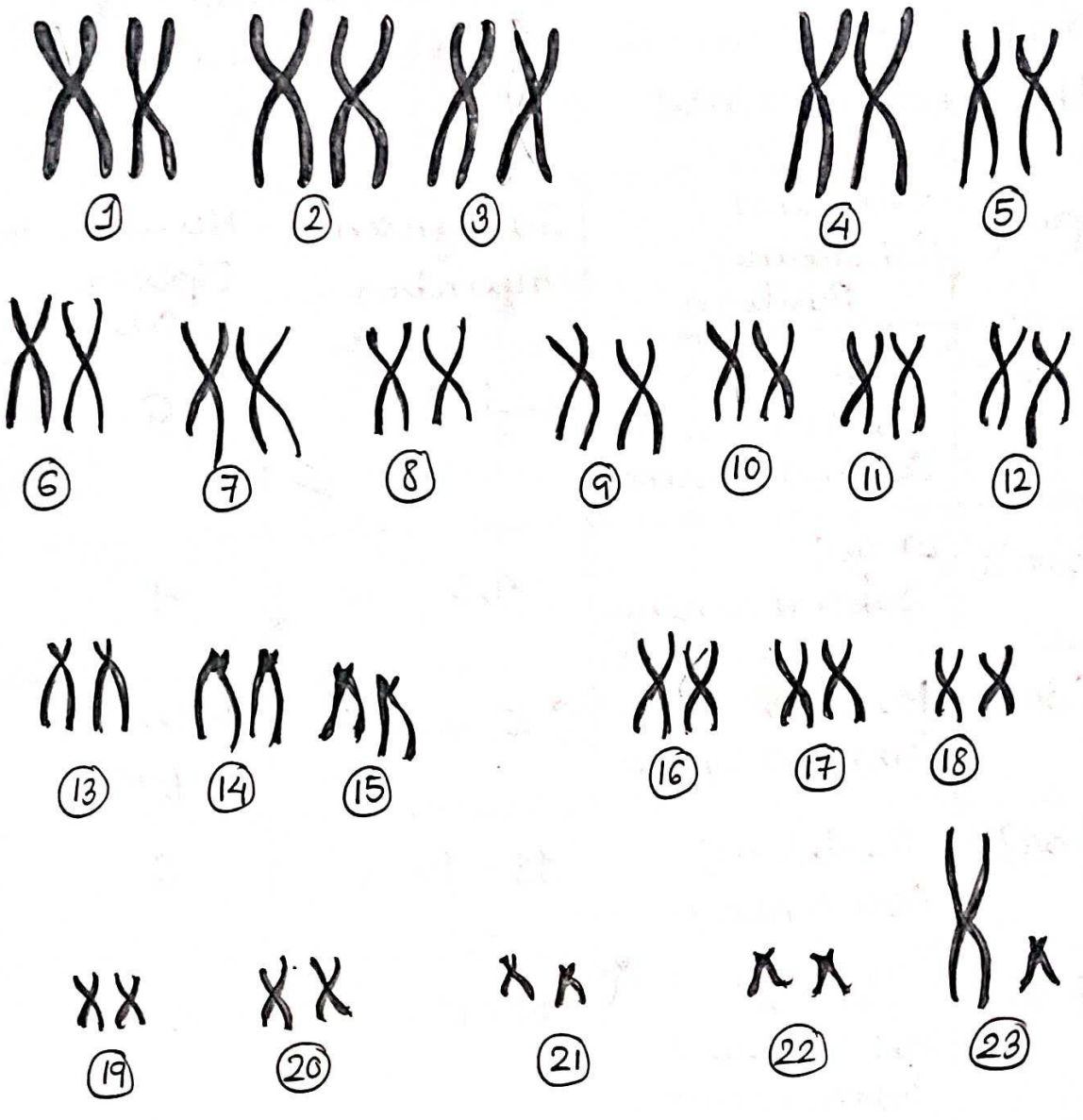

fig. Normal human Karyotype.

Materials - Karyogram, Idiogram of human karyotypes, pencil, pen, notebook, etc.

Procedure -

The supplied idiogram was observed for chromosome type and numbers.

Observation I -

(a) 22 pairs of autosomal chromosomes and one pair of sex chromosomes.

(b) The karyotypes for female contains two 'X' chromosomes and are denoted as 'XX'.

(c) The karyotypes for male contains both 'X' and 'Y' chromosomes and are denoted as 'XY'.

Conclusion -

From the above observation it is identified as normal human karyotype of male

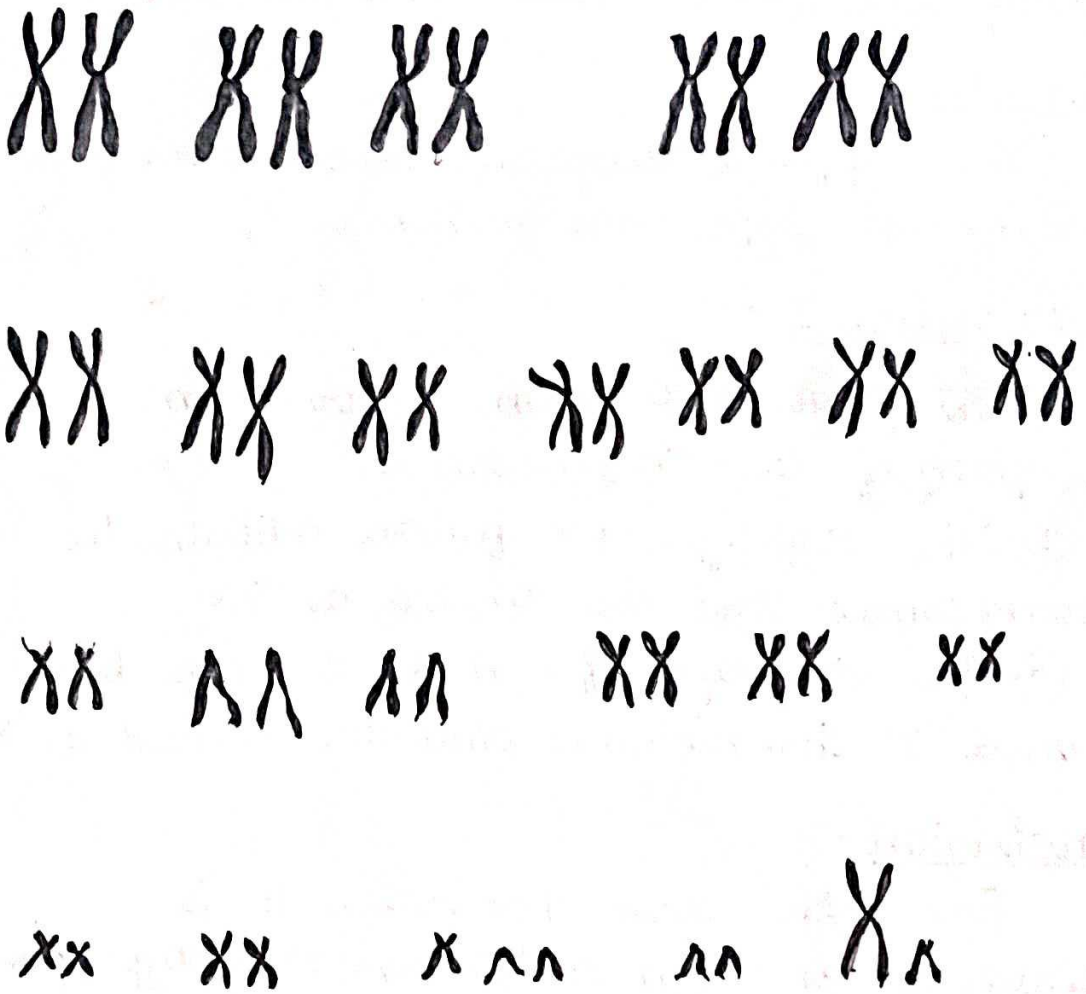

fig. Down's syndrome Karyotype in human

Observation II:

An extra copy of chromosome 21 (trisomy 2n+1) down's syndrome an aneuploid condition.

Symptoms of the disease:

(a) A broad flat face, eyes with an epicanthic fold.
(b) Short stature.
(c) Short hands with a crease across the middle.
(d) Large wrinkled tongue.
(e) Flat back of head and abnormal ears.
(f) Congenital heart disease.
(g) Big toes widely space.

Conclusion -

From the above observations it is identified as Down's syndrome caused due to trisomy of the 21st chromosome.

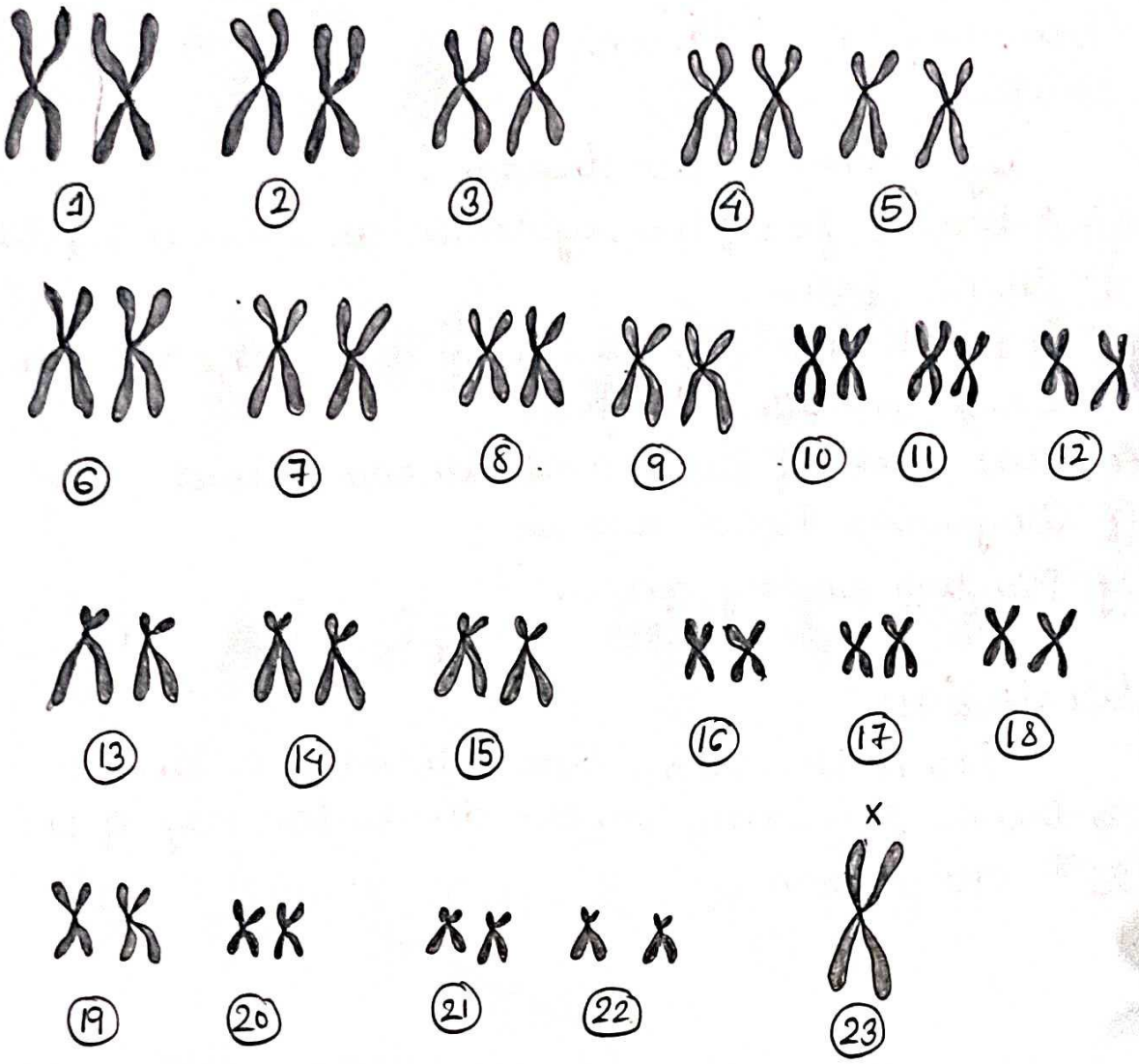

fig. Turner's syndrome karyotype in humans.

Observation III :

A single copy of X-chromosome (monosomy 2n-1), Turner syndrome.

Symptoms of the disease :

ⓐ Sterile female short in stature.
ⓑ Poor breast development.
ⓒ Elbow shows deformity
ⓓ Brown spots
ⓔ Often has a web of skin extending between the neck and the shoulder.

Conclusion -

From the above observation it is identified as Turner syndrome caused due to monosomy of X-chromosome.

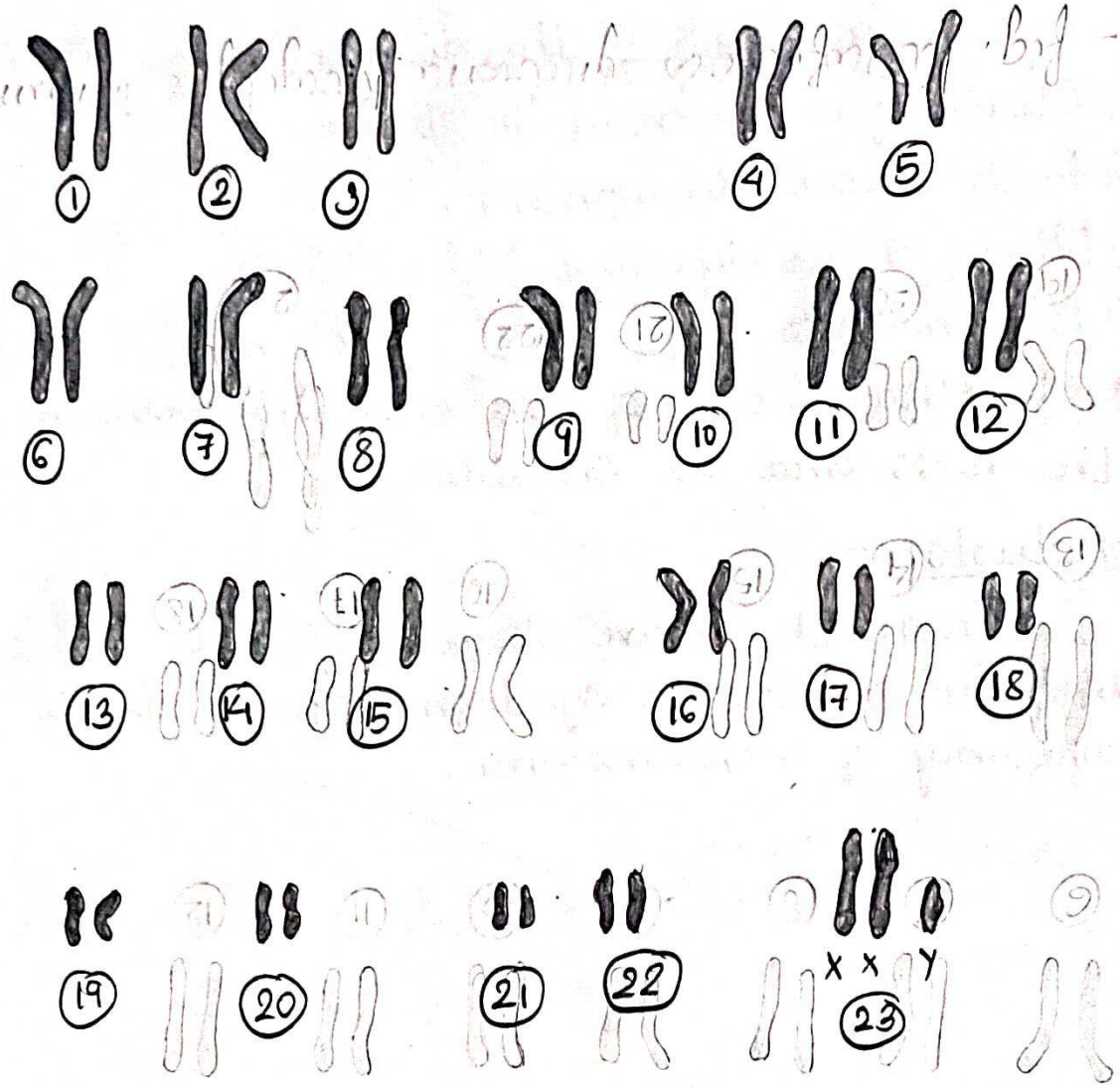

fig. Klinefelter syndrome karyotype in human.

Observation IV :

An extra copy of chromosome X (Trisomy 2n+1) Klinefelter syndrome.

Symptoms of the disease -

(a) Male with small testes and developed breasts.
(b) Long limbs.
(c) Knock knees that is a condition in which the legs curve inward so that the feet are apart when the knees are touching.
(d) Underdeveloped body hair.

Conclusion :

From the above observations it is identified as Klinefelter's syndrome caused due to trisomy of X-chromosome.

14/12/21