Down's Syndrome (Mongolism)	Turner's syndrome
(i) John Langon Down 1st described the clinical symptoms of this disease in 1866. In this honour the syndrome has been named as Down syndrome.	(i) It was 1st described by H.H. Turner in 1938. In this honour the syndome has been as Turner's syndrome.
(ii) It is a condition in which an additional copy of chromosome (trisomy of 21st chromosome) is present in humans.	(ii) It is a condition in which a female is missing one of the two X - chromosomes.
(iii) Zygote having 2n = 47 chromosome.	(iii) Here chromosome no.'s 44+XO or 2n = 45.
(iv) It is caused due to trisomy.	(iv) It is caused due to monosomy.
(v) Symptoms are broad head with round face, wide nostril, open mouth, large tongue, hyperflexible joints etc.	(v) Symptoms are webbed neck, virtually no ovaries, limited secondary sexual characters.
(vi) Diagnosis by ultra sonography and amniocentesis.	(vi) Diagnosis is done by Physical examination and genetic analysis.

CHROMOSOMAL DISORDERS IN HUMANS

1) Down's Syndrome (21-trisomy)
2) Turner's Syndrome (2n = 45 or 44A+X)
3) Klinfelter's Syndrome (2n = 47 or 44+XXY)

1) Down's Syndrome:-

→ It is also known as Mongolian Idiocy or Mongolism or 21-trisomy.

→ It is one of the most common chromosome abnormality of man.

→ It was first reported by a British Physician Langdon Down in 1866.

→ It is caused by the Presence of an extra chromosome 21.

→ It is caused by a chromosomal aberration, known as aneuploidy (trisomy).

→ The twenty first chromosomes is present in three doses, instead of two in normal persons. Thus the diploid chromosome number becomes 47, instead of normal 46.

→ Zygote having 2n = 47 chromosomes (45+XY in male, 45+XX in female) instead of 46.

→ It is the result of primary nondisjuction (failure of separation of homologous chromosomes).

→ Through the ~~and~~ investigation of J. Lejeune in 1959, Down syndrome was recognized as the first genetic disorder in human.

→ It is also identified as trisomy 21.

Clinical Symptoms :-

→ Broad head with round face.
→ Wide nostril, open mouth and large tongue with distinct furrows.
→ Hyperflexible joints.
→ Mental retardation.
→ Short stature with an epicanthal fold.

Diagnosis, Treatment and Prevention :-

→ Parental screening of the pregnant women is undertaken by ultrasonography and amniocentesis sampling to make sure about the contraction of this disorder.
→ Life expectancy is 50-60 years.
→ There is no treatment available as yet.
→ However, counseling through education support and creation of sheltered work environment works encouragingly.

2) Turner syndrome!-

→ Henry H. Turner first described this genetic disorder in 1938.

→ It is a condition in which a female is missing one of the two X chromosomes, such that the complement becomes 45 XO.

→ It occurs in about 1 Per 2500 live females births.

→ It is caused by a chromosomal abernation, known as aneuploidy (monosomy).

→ In the female, one out of two X chromosomes is missing. Thus, the chromosomal number is 45 instead of normal 46.

Clinical symptoms!-

→ Webbed neck.
→ Short stature with low-set ears.
→ Shield-like chest.
→ Virtually no ovaries.
→ Limited secondary sexual characters.
→ Swollen hands and feet.

Diagnosis, Treatment and Prevention!-

→ Diagnosis is done by Physical examination and genetic analysis. Turner syndrome affected subjects undergo hormonal therapy.

→ Growth hormone injection in early childhood may increase the height by few inches.

→ Estrogen and Progesteron are administered together, a little later to initiate the monthly cycle.

→ Estrogen replacement therapy is undertaken at puberty to start the breast development.

→ Turner syndrome affected persons have a shorter life expectancy.

3) Klinfelter syndrome :-

→ H.F. Klinfelter first described this genetic disorder in 1942.

→ It is an abnormal genetic condition, caused by the presence of an extra X chromosome in addition to the usual male sex chromosome complement of XY.

→ Thus the diploid chromosome number becomes 47 with XXY sex chromosome complement.

→ The condition is due to the presence of an extra X chromosome in the male.

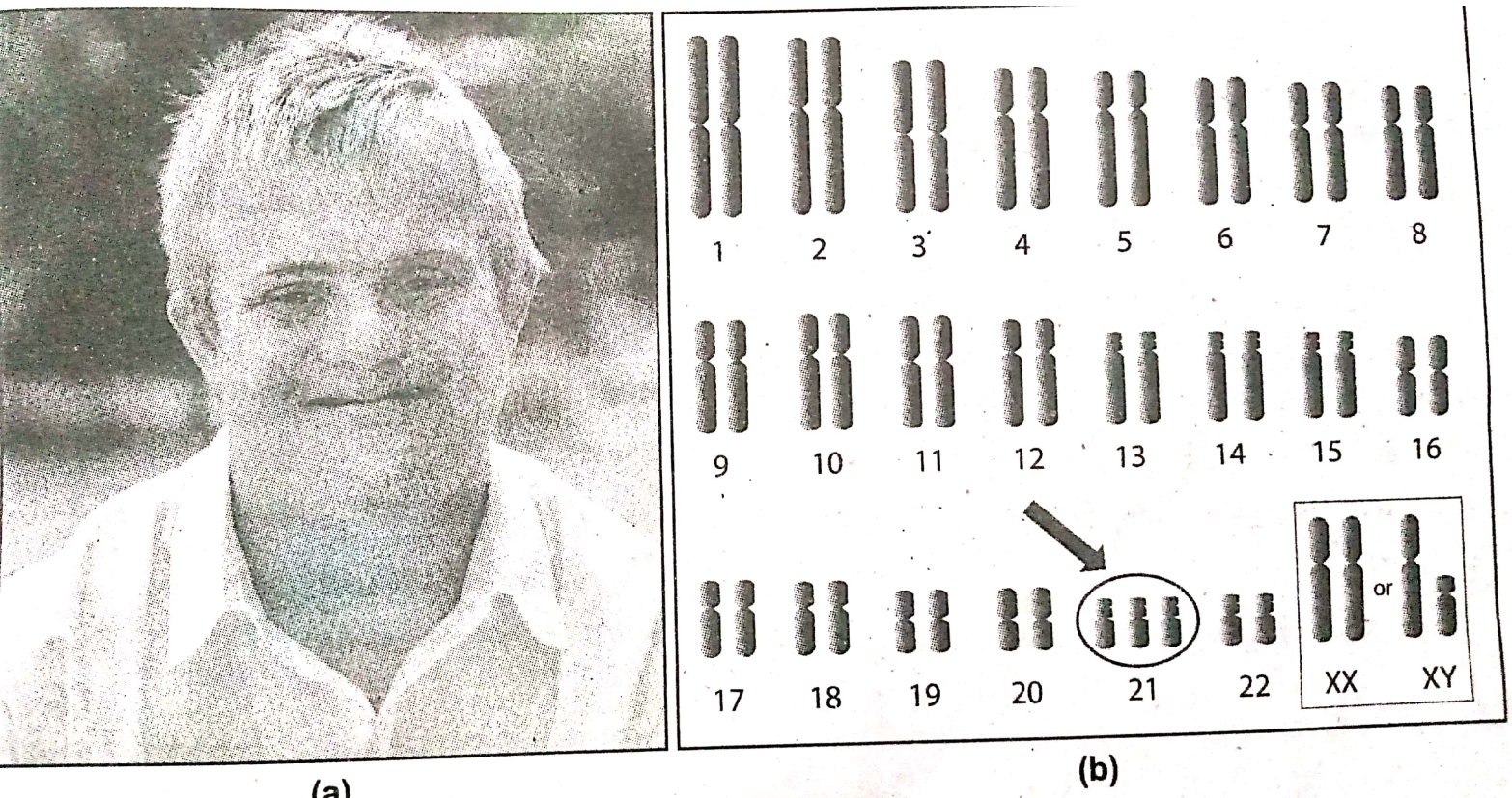
→ Klinfelter's syndrome is generally seen in one out of every 500 male births.

→ It arises by the non-disjunction of sex chromosomes during meiosis.

→ The syndrome is diagnised by a chromosome complement examination.

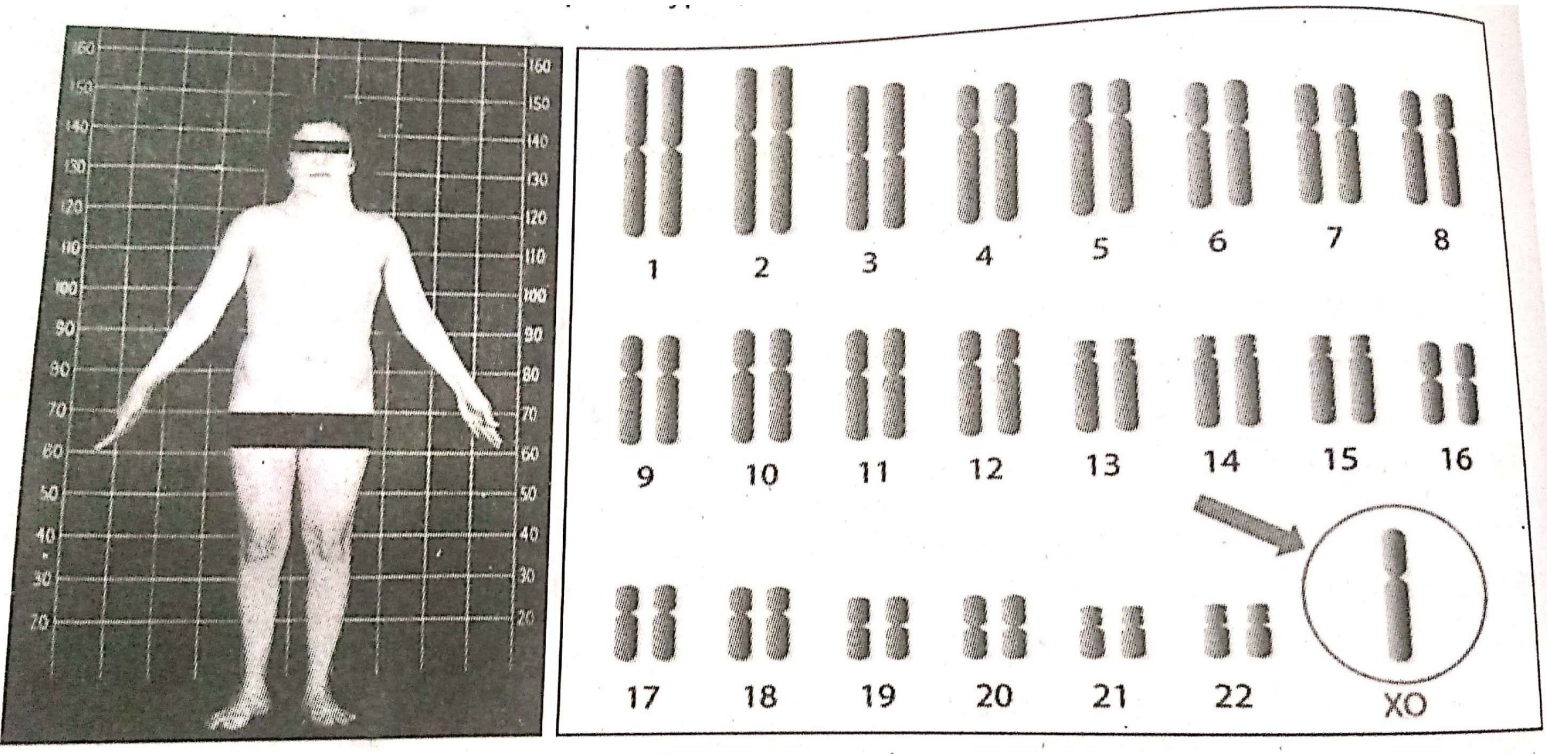
→ There is no treatment option.

→ DI can be prevented through education and genetic counselling by a trained clinical geneticist.



(a) (b)

Fig. 6.12 : (a) A person with Down syndrome phenotypes, (b) Down syndrome chromosome complement (the arrow shows three doses of 21st chromosome).



(a) (b)

Fig. 6.13 : (a) A female with Turner syndrome phenotypes, (b)Turner syndrome chromosome complement (the arrow shows one X chromosome, there being no Y chromosome).

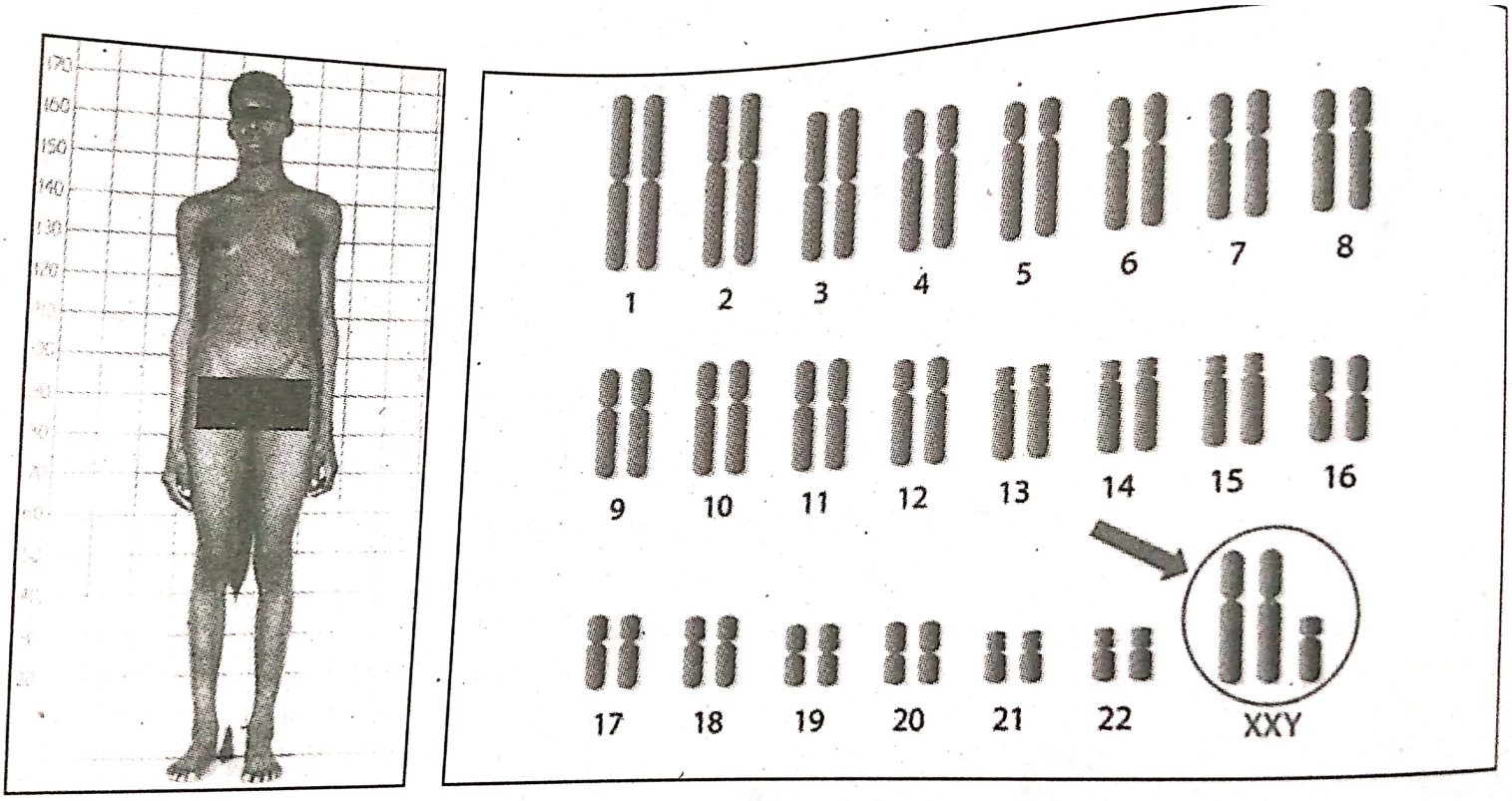


Fig. 6.13 : (a) A male showing Klinefelter syndrome phenotypes and (b) Chromosome complement (the arrow shows the presence of an additional X chromosome).

SUMAN STUDY ACADEMY
Keep Learning...