

Unit-5.

Human Chromosome

Human - 23 pair - 46
total chromosome. Female - 2
copies of X and male one X and
one Y - 23rd - Sex chromosome

Karyotyping - collection of chromosomes
- test - help - to - identify genetic
disease - also known - chromosome
analysis.

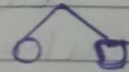
Test performed in - Amniotic
fluid - Blood - Bone marrow

Pedigree analysis.

The study of inherited
trait - group of individuals - phenotypic
- data - collected ^{from} several generation -
drawn - determine - whether - Recessive
or dominant .

Important tool - both - basic -
research & Genetic Counselling
□ - Male ○ - Female □○ - Mating

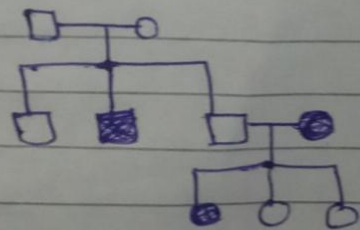
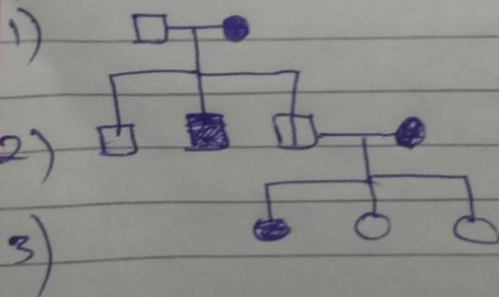
○-○ - Identical twins



Non identical twins

Dominant

Recessive



Klinefelter's Syndrome.

Genetic condition - male -
born with - extra copy - x chromosome.
Medical diagnosis - produce - less -
testosterone - development of small
testicles - cause - reduce muscle mass -
reduced body - facial hair - Enlarged
breast tissue.

Down Syndrome.

Genetic chromosome 21 -
Cause - developmental and intellectual
delays - abnormal cell division - trisomy 21
During development of sperm
or egg - small - round head - palm is
broad - furrowed tongue - open mouth.

Turner Syndrome.

Chromosomal disorder -
female - born with - one x chromosome -
Causes - developmental problems -
short height - failure of ovaries -
develop heart defects.

Inborn errors of metabolism.

Phenylketonuria.

Birth defect - Causes -
amino acid - phenylalanine - build up
- body - enzyme - not secret - phenylalanine
- accumulated - converted - pyruvic
phenyl pyruvic acid, excrete - through
- urine.

Alkaptonuria.

Rare genetic disorder of metabolism - Accumulation - of - homogentisic acid - Affected - who lack of enzyme breakdown - homogentisic acid - Black urine disease - dark spots in sclera - black earwax - Dark sweat - Kidney stones - Arthritis.

Genetic Counselling.

Process - advising - individuals - families - affected by at risk of disorders - help them - to understand - adapt - to - medical - psychological - familial implications.

Ethnics.

Study of improvement of human functioning and well being by improvement of living condition - Altering external factor - education - Controllable environment - Prevention - removable of Contagious disease - parasites - environmental^{ism} - education - regarding employment - home Economics - Sanitation - housing.

Eugenics.

Improving of human race through heredity law or using pedigree analysis.

Science of improving human species by selectively mating people

With specific desirable hereditary traits. Aim \rightarrow to reduce human suffering by "breeding out" disease.
1883 - Sir Francis Galton - term.

② types.

① positive \rightarrow increasing desirable trait

② Negative \rightarrow decreasing desirable trait