# Genetics: → Diagrams ★

Q.1 Barr body ★
Q.2 Lyons hypothesis
Q.3 Turners syndrome ★ (X-monosomy)

Q.4 Karyotyping ★
Q.5 Monosomy of sex chromosomes (XO) ⇒ Turner's syndrome
Q.6 Down's syndrome ★
Q.7 Structural chromosomal Abnormalities
Q.8 Autosomal Dominant Inheritance
Q.9 Structural & Numeral chromosome Abnormalities with Example.

- Trisomy → 21 → Downs syndrome
  → 18 → Edward syndrome
  → 13 → Patau syndrome

Q8 Turner's syndrome ? / X-monosomy.

Ans → It is a sex chromosome abnormality.
Genetic condition in which a female is partly or completely missing an X chromosome

→ Named bcoz it was described by Turner

→ Etiology →
* caused by partial or complete monosomy of X- chromosome
* XO karyotype with no barr body.

→ Clinicals →

① short- stature with a broad chest- with widely spread nipples
② cystic tumor of neck leading to webbed neck appearance
③ lymphedema of extremities
④ 1° Amenorrhea usually present
⑤ Infantile genitalia & breast-
⑥ Heart- defect- congenitally. [Eg → Coarctation of Aorta]
⑦ Increased risk for developing → DM
→ Hypertension
⑧ Juvenile external genitalias.

→ findings → Decreased estrogen production
→ Increased FSH & LH production

→ Treatment- → Estrogen replacement- → to develop 2° sexual characters.
→ Administration of GH to treat- short- stature

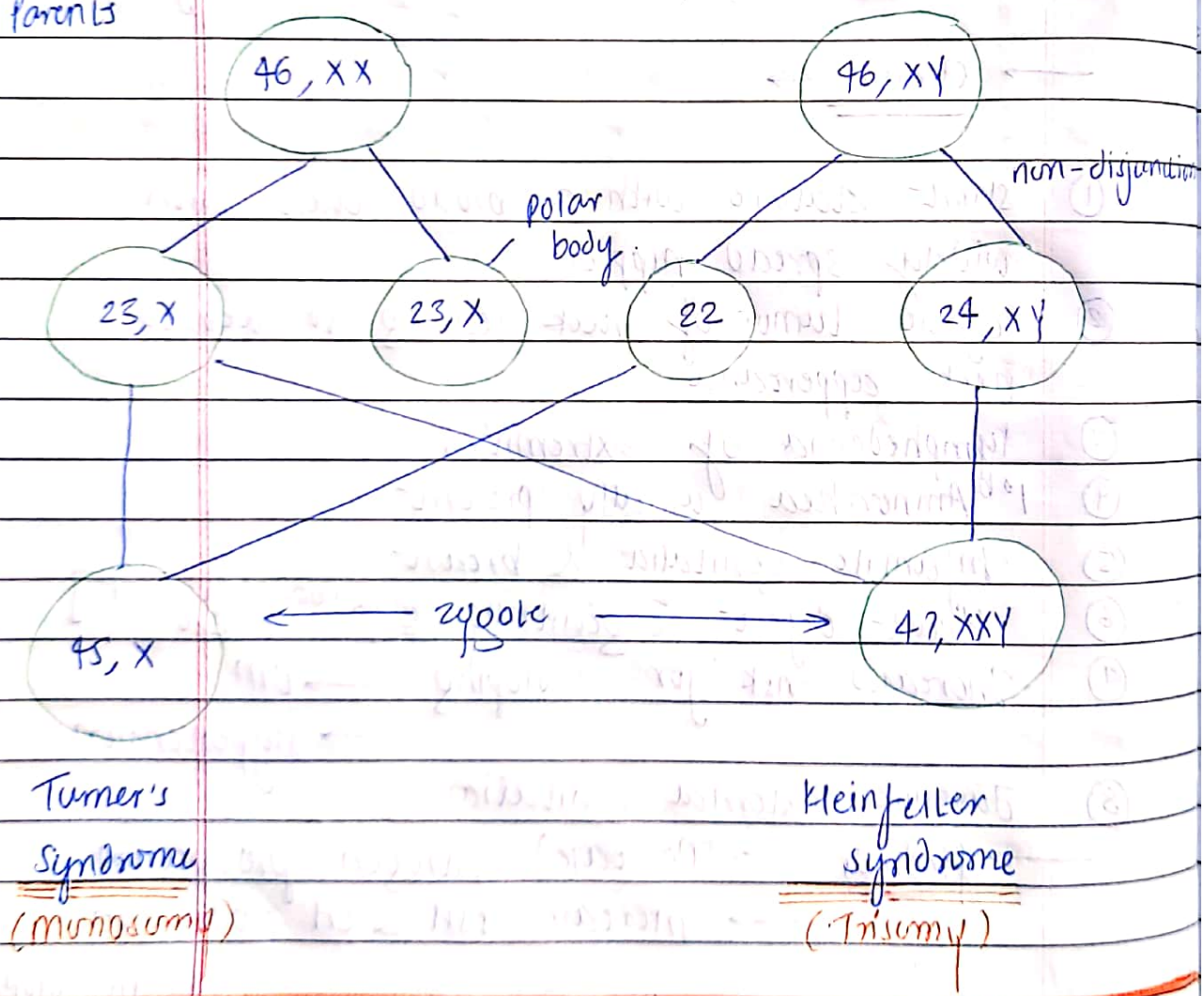
→ Cytogenetics →

* >60% patients → 45, X Karyotype

Others have alteration in their other X chromosome

X → Xq (most common)*

[ischiochromosome]



Q. Structural chromosomal Aberrations : →

Ans Results from breaks followed by reconstitution of chromosomes.

Responsible factors : → ① Ionizing radiations
② Chemical agents
③ Viruses.

Classification : →

① Stable → • Deletions
• Inversions
• Translocations
• Ischiochromosomes.

② Unstable → • Dicentric
• Ring chromosomes

→ Among above mentioned ★★★ * Inversion & * Translocation can be transferred from parent to child.

★★★ Deletion → involves loss of a part of chromosome
✓ of two types

① Terminal deletion

② Interstitial deletion → कोई भी हो सकता है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**Terminal Deletion →**

- Involves single break
- Terminal part of chromosome is lost

Example → ★★★

Cri-du-chat-syndrome

— Cri-du-chat results from deletion of the short arm of chromosome-5
— Named as cry of baby mimics mewing of cat

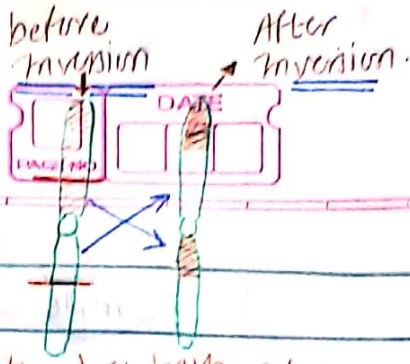
Features → ① Typical facial appearance
② Micro-cephaly → (small head)
③ Hyper-Telorism → (Abnormal large distance b/w eyes)
④ // // Anti-mongoloid slant- of eyes मतलब Nasal side से Temporal से
⑤ Micro-gnathia (small jaw)

**Interstitial Deletion →**

- Involves two breaks
- Intervening portion of chromosome lost

Example →

✓ Prader-willi-syndrome (PLOS)
✓ wilms tumor (most common kidney cancer in babies)
+
Aniridia
↳ (Partial absence of colour of iris)

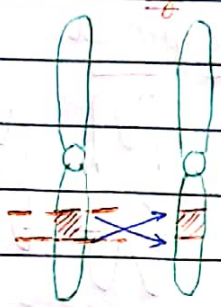



★★★ Inversions →

✓ of two types

① Pericentric inversion → involves both p & q arm of chromosome (centromere is involved)

② Paracentric Inversion → involves either p or either q.



— No Abnormal phenotype produces
— However Abnormal gametes can be produced during meiosis

↓

producing Abnormal progeny.

★★★ Translocation →

✓ of two types

① Robertson translocation → involves acrocentric chromosome

② Reciprocal translocation → involves non-homologous chromosomes.

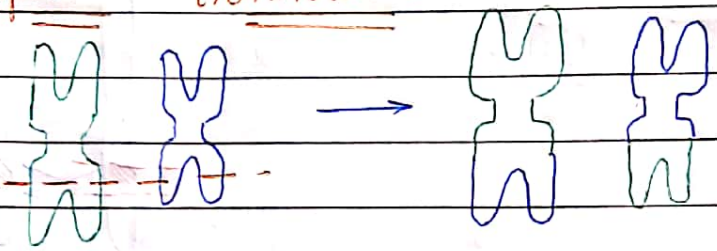
Robertsonian translocation → (centre fusion)

Example → D/G Translocation.

→ D group = (13-15) chromosome

→ G group = 21 chromosome

Reciprocal translocation →



★★★ Isichiochromosome →

Involves abnormal split along centromere leading to seperation of Arms.

Example → Xq in Turner's syndrome

★★★ Ring chromosome →

→ Involves two breaks at terminal portion of chromosome followed by fusion of cut ends.

→ found in $\frac{1}{5}$th cases of Turner syndrome

Mosaicism

① → one cell line → with 1 X-chromosome active

① → one cell line → with other active X-chromosome

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Down's Syndrome ?

Ans

* Also called Mongolism

→ Genetic disorder caused by presence of all or part of third copy of chromosome 21

→ Etiology →

* meiotic non-disjunction which
* Increases with maternal age
[found in 1 in 25 births in mothers over 45]

* Robertsonian translocation →
  - 4% cases
  - D/G Translocation

* Mosaicism →
  - 1% cases
  - mitotic non-disjunction of chromosome 21 during embryogenesis.

→ Clinical features →

① Severe mental retardation
② Duodenal & oesophageal Atresia [narrowing of passage] *
③ short-hands with simian crease [single palmar crease]
④ specific facial features → (flat face, white set-eyes)
⑤ Congenital heart defects → valve malformation

→ Complication →

* acute leukemias
* Infections
* Degenerative changes in brain (Alzheimer disease) in middle age

→ Treatment →

* Surgical treatment for
  ✓ Atresia & ✓ Heart defects

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~~8.~~ Barr body ?.

Ans also called "sex chromatin"

→ In 1949 Barr & Bertram

↓

while studying cat neurons found that some of these cells show a chromatin mass in their nuclei

*** (Observed only in females not in males)

Subsequently labelled as | Sex chromatin / Barr body. |

V. Imp → cells having barr body ⟹ Chromatin +ve

cells not having barr body ⟹ chromatin -ve

→ Barr body can be found in many cells but can be clearly examined in "buccal mucosa"

Procedure → ① scrapping from inner cheek taken on slide

② fixed in Alcohol, stained with Thionin

③ mounted on neutral medium & watched under microscope

→ Previously, Barr body → was diagnostic tool for disorder of sexual Development.

Now, karyotyping is used

V.Imp → No. of barr body in cell depends on no. of X- chromosome

Barr bodies ⇒ no. of X-chromosome − 1.

Eg → ① Individual with 47, XXX have ③ X-chromosome

Barr body = 3 − 1 = 2;

② → Individual with Turner 45, XO

Barr body = 1 − 1 = 0 ; ⇒ so chromatin -ve

→ Barr body represents one of two X-chromosome of female cell.

→ This remains condensed & inactive throughout Interphase + Replicates slowly.

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Q. Lyon's Hypothesis?

Ans Dr. Mary Lyon in 1962 stated in her hypothesis about inactivation of X-chromosome

→ it was observed at Prophase

↓

1 X-chromosome is - late replicating &
- Heteropyknotic

i.e this X-chromosome differs from the other in respect to state of condensation and staining.

Females → Of both only 1 ⇒ active in cellular metabolism
other ⇒ inactive forms sex chromatin

Males → have only 1 X-chromosome so they don't have Barr body.

Hypothesis →

① In female somatic cell, only one of both X-chromosome is active.
2nd = inactive, condensed, appearin form of sex-chromatin in interphase.

② Inactivation occurs in embryonic life.

③ Inactivation is random but fixed.
The inactive X can be maternal or paternal ($X^m$ or $X^p$) in diff. cells of same individuals.
- However, once decision is made that which X-chromosome will be inactivated
↓
Then all clonal descendent will have to follow the decision.

Sex chromatin is detected in blastocyst at 9-12 days.
1st in → syncytio-trophoblast
Then → chorionic mesoderm
followed by → Yolk sac.

Q. Karyotyping ?

Ans Process by which karyotype is obtained in this process metaphase chromosomes are obtained for Analysis & photo-micrograph

→ Photograph of individual chromosomes are cut & arranged according to standard classification.

→ This is called Ideogram or karyotype

Chromosomal Preparation →

→ can be obtained from somatic cells by culturing them.

→ It can be a- short-term-culture or
- Long-term-culture
depending upon the cell used

★ → cells of fibroblast & Amniotic fluid are of long-term culture.

→ In some cells chromosomes can be observed without micro-scope

eg → ① Bone marrow
② chorion villous sample.

Steps → = ① collection of blood

= ② Planting

= ③ Incubation

= ④ Harvesting and

= ⑤ Staining

→ and then observing under microscope.

Applications → ① Clinical Diagnosis →

✓ karyotyping helps in reaching a clinical diagnosis.

✓ especially indicated in patients with congenital malformations involving

= Multiple systems

= Mental retardation

= Ambigious genitalia.

(जिसके बारे में साफ तौर पर निर्णय नहीं ले सकते)

② Gene mapping →

Helps in knowing proper location of human genes on chromosomes

③ Role in cancer →

Detection of Philadelphia chromosome

in patients with CML (chronic myelogenous Leukaemia) alters prognosis.

→ philadelphia chromosome is formed by translocation b/w chromosome → 22 & 9

④ Repeated foetal loss

⑤ Prenatal Diagnosis → knowing defects
↓
so if MTP required it can be performed

Q. Types of chromosomal Abnormality?

Ans

Numerical

→ Aneuploidy → Autosomal
→ sex-chromosomal

= Mono-somy (Loss of 1 chromosome)
= Tri-somy (gain of 1 chromosome)
= Tetra-somy (gain of 2 chromosome)

→ Polyploidy → Triploidy
→ Tetraploidy.

Structural → Stable
→ Unstable

* Discussed earlier.

Q. Autosomal dominant- Inheritance ?

Ans 1 of 4 types of mendelian Inheritance.
An autosomal dominant trait- expresses in heterozygous state.

— Homozygous are seriously affected due to double dose of Abnormal gene.

→ Traid through various generations

Example → In south Africa numerous cases of "Porphyria variegata"

have been traced from couple of 17th century.

① → it is a disorder of metabolism, presenting skin blisters owing to increased sensitivity to sunlight.

② → Urine becomes = "port wine"
Reason → Numerous porphyrins.