

Chromosomal disorders
'No. chromosome' vary (Gene ass-)

Aneuploidy

Trisomy
($2n+1$)

Monosomy
($2n-1$)

disease due to imbalance of chromosome

(I)

Autosomal

Down syndrome / Mongolian / Mongolism

* * 21th chromosome - Extra
(Trisomy) ($2n+1$)

↓ IQ low, etc.

(3) ~~pentasomy~~ (3) = 13th (Trisomy)

→ cardiovascular defective

(2) Edward syndrome

18th ($2n+1$)

Defective nervous system

⇒ ear less develop

lip ⇒ 6 months का
($c-1$ year)

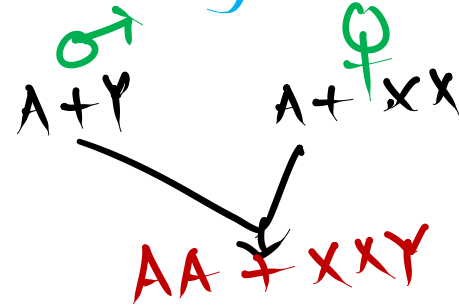
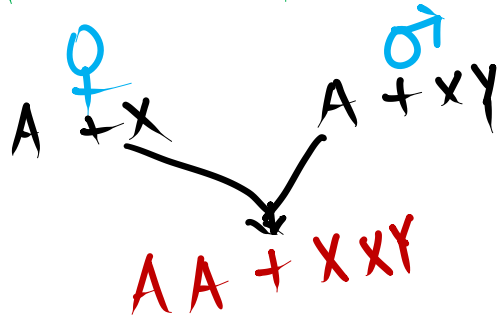
III Cri du chat syndrome
(cat-cry) 5th → small arm
Deletion

eg → eye (less dev)
small head (microcephaly)
moon face

Sex chromosomal Disorders

'Klinefelter's syndrome' (AA + XXY)

AA + XXY (47) (Trisomy of sex chromosomal)



- Mentally retarded
low IQ
- Hormonal imbalance

II Turner's Syndrome 41
(AA + XO)

(Monosomy) $\Rightarrow (2n-1) = AA + X\underline{O}$ (sex chromosome monosomy)

♀ female (sterile female)
 \uparrow ovaries - egg phenotypically ♀

\Rightarrow estrogen absent

\Rightarrow secondary sexual charact absent, low IQ sterile

Super female (AA + XXX)
(2 Barr body) (X-chromosome Trisomy)
AA + XXX

\Rightarrow phenotypically ♀
IQ less, sterile
sex hormone imbalance

Jacob (Jacob's syndrome)
Super male

AA + XYY
(Y chromosome \rightarrow Endoduplication)
 \rightarrow with excessive \rightarrow hormones
(Testosterone)
AA + XYY normal (initially "fearless")

Alkaptonuria (Black urine disease)

Autosomal recessive, A. Garrod

↳ Enzyme (Homogentisic Acid oxidase) due to absence of 1

⇒ Urine → Acid → Black colour pigment
↓
Urine (Black)

store in tissue organs

⇒ Nose - cartilage ⇒ Black

Acid → store ⇒ "Renal stone"

Sunburn ⇒ sunlight ⇒ skin burn.

photophobia ⇒ light to SR

eye is damage

due to absence of enzyme

- 1)
- 2)
- 3)

Albinism

Autosomal Recessive

Enzyme - Tyrosinase

DOPA (dihydroxy phenylalanine) → Melanine
↓
pigment

Main pigment of Human body

Melanine

↓
Skin of human, eye's hair
high ↑↑, ↑↑
uv. light ↑↑

Cystic fibrosis - Autosomal recessive.

CFTR (protein)

"cystic fibrosis transmembrane conductance Regulator"

Cell membrane
pH

(CFTR)
Cl⁻ ⇒ Transport →

Jordan's criteria
Hemolysis ↑↑

① G-6-P D Syndrome (Favism) disease
 X-linked Recessive disease

[Glucose-6-phosphate Dehydrogenase]

→ Absence

→ RBC and Hb fast → Break/damage

↳ Anemia → Jandev -

Sulpha, Drugs, chloroform, legu, etc.

② (Duchenne's Muscular Dystrophy)
 (X-linked Recessive dis.)

due to → Dystrophin absence.

↳ Muscle → Sarcolemma → Internal surface

↳ membrane → Tensile strength

↳ (Muscle contractile) Actin and Myosin

↳ paralysis → Body organ in repair