ABLES® KOTA

Chromosomal disorder's

'No. chromosome' vary (Genc als-

disease due to inbalance of chromosome

Aneuploidy

- Trisomy ($2n+1$)
- Monosomy ($2n-1$)

Ⓘ Autosomal

Down syndrome / Mangolian / Mangolism

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

1) IQ Low, etc.

② Edward syndrome

18th ($2n+1$)

Defective Nervous system

⇒ ear less develop

③ Patau syndrome ⇒ 13th (Trisomy)

→ cardiovascular defective

lip ⇒ कटा फटा

(← 1 year)

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

eg → Syn (less dev.
small head (microcephaly)
moon face

Sex chromosomal Disorder

'Kleinfelter's syndrome' (AA + XXY)

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

- Mentally retarted
  low (IQ)
- Hormonal imbal.

♀ A + X × ♂ A + XY → AA + XXY

♂ A + Y × ♀ A + XX → AA + XXY

41

II Turner's syndrome (AA + XO) 45

(Monosomy ⇒ (2n-1) = AA + XO

(sex chromosome monosomy)

♀ female (sterile female) ← ovaries - egg

phenotypically ♀

⇒ estrogen absent

⇒ secondary sexual charact absent, low IQ sterile

Jacob ('Jacob's syndrome')

Super female (AA + XXX)

(2 Barr body) AA+XXX

(X-chromosome Trisomy)

⇒ phenotypically ♀

IQ less, sterile

sex horm imbalance

⇒

Super male

AA + XYY

(Y chromosom → Endoduplication

♂ with excessive ♂ Hormones (Testosteron's)

AA + XYY Normal (mini)

"Fearless"

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Alkeptonuria (Black urine disease)

Autosomal recessive, A. Garrod

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

⇒ Urine ⟶ Air ⟶ Black colour pigment con-
⇓
Urine (Black)

stored ← tissue ← organs ←

⇒ Nose - cartilage ⇒ Black

Air ⟶

Kidney ⟶ stone ⇒ "Renal stone"

due to absence of melanin

1) Sunburn ⇒ sunlight ⇒ Skin burn.

2) photophobia ⇒ light से डर

3) eye में damage

Albinism

Autosomal Recessive

Enzyme. Tyrosinase

DHPA (dihydroxy phenylalanine) ⟶ Melanine ↓ pigments

Main pigment of Human body
↓
Melanine
↓
Skin of Human, eye's Hair
high ↑↑, ↑↑ u.v. light

Cystic fibrosis - Autosomal recessive.

CFTR (Protein)

"cystic fibrosis transmembrane conductance Regulator"

(CFTR)

⇓

cell membrane

ent

Cl^- ⇒ Transport

Jondis पीलिया

Hemolysis ↑↑

(I) G-6-P D Syndrome (Favism) disease

X-linked Recessive disease

[Glucose-6-phosphate Dehydrogenase] → Absence)

⇒ RBC and Hb fast → Break/damage)

↳ Anemia → Jondice

Sulpha, Drugs, chloroquine, legumes, etc.

(II) (Duchhen's Muscular Dystrophy)

(X-linked Recessive dis)

due to ⇒ Dystrophin absence.

↳ Muscle → Sarcolemma → Internal surface

↳ mem → Tensile strength

↳ (Muscle (contractile) Actin and Myosin

↳ paralysis ⇒ Body organ mein