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Sub. Biology

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- Phenylketonuria
  - Autosomal recessive disease
  - Phenylalanine $\xrightarrow{\text{Enzyme}}$ Tyrosine

    The enzyme responsible for this conversion gets mutated.
  - Phenylalanine accumulates. Then,

    Phenylalanine → Phenylpyruvic acid → Accumulates in brain → Mental retardation
  - Phenylpyruvic acid also gets excreted through urine since kidneys poorly reabsorb it.

Chromosomal Disorders

- Total number of chromosomes in humans = 46 (23 pairs)
- Total 23 pairs = Autosomes (22 pairs) + Sex chromosomes (1 pair)
- Monosomy − Lack of any one pair of chromosomes
- Trisomy − Inclusion of an additional copy of chromosome
- Aneuploidy − Loss or gain of chromosomes due to the failure of segregation of chromatids during cell division
- Down's Syndrome
  - Cause: Presence of an additional copy of chromosome 21 (Trisomy of 21)
  - Affected individual has short stature, small, round head, furrowed tongue, partially opened mouth, palm crease, congenital heart disease and mental retardation.
- Klinefelter Syndrome
  - Cause: Additional copy of X chromosome, i.e., 47 chromosomes (XXY)
  - Affected individual has an overall masculine development with gynaecomastia; individual is sterile

- Turner’s Syndrome
    - Cause: Absence of one X chromosome, i.e., 45 chromosomes (XO).
    - Affected females are sterile; have rudimentary ovaries; secondary sexual characters are absent