Infantile Hypophosphatasia

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About Infantile Hypophosphatasia

- It is a rare genetic disease in which the patient’s bones and teeth demineralise, making her fragile and prone to fractures.
- Symptom
    - It may have no noticeable abnormalities at birth, but complications become apparent within the first six months of life.
    - The initial problem may be the baby’s failure to gain weight and grow as expected, referred to as “failure to thrive.”
    - Sometimes the skull bones fuse, called craniosynostosis, which can lead to a deformed head (brachycephaly).
    - Affected infants have softened, weakened and deformed bones consistent with rickets.
    - Rickets is a general term for the complications due to defective skeletal mineralization during growth with softening of bone and characteristic bowing deformities.
- Cause
    - It is caused by mutations in the ALPL gene.
    - This is the only gene that causes HPP. Genes provide instructions for making proteins that have an important function in the body.
    - When a mutation occurs, the protein may be faulty, inefficient, or absent, as in HPP.
- There is no known cure for this disease.

Q1) What is Rickets?

Rickets is a medical condition that primarily affects children and is characterized by the weakening and softening of bones. It is primarily caused by a deficiency of vitamin D, calcium, or phosphate in the diet, which are essential for proper bone development and maintenance.

Source: Families of India’s first cases of two rare disease patients struggle for inclusion under national policy