Climb™

Supporting those affected by Inherited Metabolic Disorders

X-Linked Hypophosphataemic Rickets

Calcium and phosphate are minerals that are essential for the integrity of bones. XLH is a genetic condition which lowers the amount of phosphate in the body by affecting how the kidneys work. The resulting low phosphate levels particularly affect the growth and strength of the skeleton. The kidneys normally either lose phosphate when there is too much, or retain phosphate when the body needs more. The important FGF23 protein tells the kidneys what to do and this also helps in the formation and maintenance of bones. In XLH, defects in the PHEX gene lead to an increase in FGF23 and contributes to short stature, deformity and fragility of the bones and a variety of abnormalities of the teeth, including recurrent dental abscesses.

Synonyms

Alternative names for this condition are:

- XLH
- X-Linked Hypophosphatemia
- Familial Hypophosphatemia or Familial Hypophosphatemic Rickets
- Vitamin D-Resistant Rickets (VDRR)
- Genetic Rickets
- Vitamin D-Dependent Rickets (VDDRI and VDDR II)

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

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Telephone: **0845 241 2173** Freephone: **0800 652 3181**

Email: contact@climb.org.uk Website: www.climb.org.uk