Introduction

Hereditary Hypophosphataemic Rickets (HPR) is a condition related to low levels of phosphate in the blood (hypophosphatemia). Phosphate is a mineral that is essential for the normal formation of bones and teeth in childhood and helps maintain bone strength in adults.

What causes HPR?

The condition is due to excessive loss of phosphate in the urine which leads to poorly formed bones (rickets), causing bone deformity in growing children, bone pain and sometimes tooth abscesses.

With recent advances in the understanding of the genetic basis of hypophosphataemic rickets the name of X-linked hypophosphataemic rickets has become more commonly used. This is due to changes in the phosphate regulating gene (PHEX) which is on the X chromosome. This is also the commonest type of HPR. The condition often runs in families through several generations, but in other families a newly diagnosed child may be the first and only affected person.

PHEX stimulates fibroblast growth factor 23 (FGF-23) which is a protein produced in bone cells. It is necessary for regulating the phosphate levels within the body. FGF 23 levels are sometimes monitored as part of the routine blood tests we undertake in patients with HPR.

The body’s phosphate levels are determined by the balance between:

- how much phosphate is taken in from food,
- how much is stored in the skeleton
- how much is passed out (excreted) in the urine.

Along with FGF23, a bone hormone called PTH (parathyroid hormone) plays an important role in controlling how much phosphate the kidneys excrete. PTH is also routinely measured in our blood tests.

Signs and symptoms of HPR

In most cases, the signs and symptoms of hereditary hypophosphataemic rickets begin in early childhood. The features of the disorder vary widely, even among affected members of the same family. Many children experience slow growth and are shorter that their peers. They develop bone abnormalities that can interfere with movement and cause bone pain.

The most noticeable of these abnormalities are bowed legs and knocked knees (a condition in which the lower legs are positioned at an outward angle). These abnormalities can become apparent with weight bearing activities such as walking. If untreated, they tend to worsen with time and a number of children with HPR will require the input of orthopaedic surgeons.
Fractures tend not to occur in HPR but mobility can become limited because of discomfort, deformity or both. Having said all this, if the condition is detected early and compliance with treatment is good, it is possible to have reasonably good growth and little in the way of bony deformities.

How is HPR treated?

The clinical management of hypophosphataemic rickets involves:

1. Medication with phosphate and a vitamin D preparation called alfacalcidol (or one alpha)
2. Physiotherapy/hydrotherapy as required
3. Orthopaedic surgery and the use of orthotics such as supportive shoes and insoles
4. Regular dental checks

Phosphate and alfacalcidol are used together and doses are adjusted as the child grows and dependent on blood results. Blood tests will be frequent at first but will lessen once the condition is more stable. It is important that once a year the child’s urine is checked for calcium and an ultrasound scan of the kidneys is done. This is because alphacalcidol can cause calcium to be excreted in the urine in higher amounts than usual, and sometimes some of this calcium can be deposited in the kidneys (a bit like chalk). This does not usually cause the child problems but would be monitored.

Are there any alternatives to this treatment? What will happen if HPR is not treated?

Although at present there is no cure for HPR, the best chance of a good outcome in terms of growth and mobility lies in early diagnosis and good compliance with medication. Children should ideally be seen in a specialist paediatric metabolic bone or endocrine clinic at least three times a year, or in a shared care arrangement with a local paediatrician.

Who to contact for further information

Lynne Hatchard, Endocrine Nurse Tel: 0151 252 5534
             Monday- Friday  8.30 am - 9.30 am
             or
             2.00 pm -3.00pm

Gill Murphy, Secretary Tel: 0151 252 5281

This fact sheet only gives general information. You must always discuss the individual treatment of your child with the appropriate member of staff. Do not rely on this leaflet alone for information about your child’s treatment.

This information can be made available in other languages and formats if requested.

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