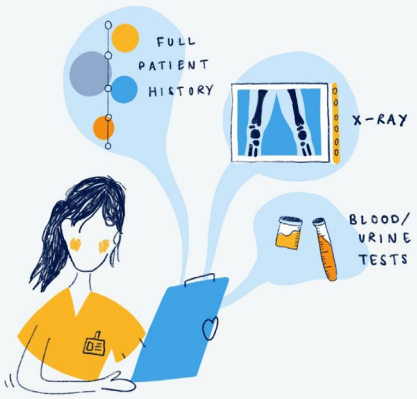
International XLH Alliance

Our 12 Recommendations

X-linked hypophosphataemia (XLH) is a rare chronic disease that substantially impacts the quality-of-life of affected people throughout their life. It is a multi-system disease that evolves over time, and there is considerable variation in the characteristics and severity of XLH between patients. Due to its rarity, the diagnosis and disease-specific treatment of XLH are frequently delayed, which can have significant negative effect on patient outcomes. These graphics present the 12 key recommendations that you can use to advocate for your better care. They are illustrated here in an easy-to-understand format and complement the current clinical practice recommendations for the diagnosis and management of XLH. The full recommendations are published by Dr Dieter Haffner[1] and other clinical and patient experts.

1



Clinical Evaluation

The following diagnostic measures should be undertaken to establish if an individual has XLH: a detailed clinical evaluation, a radiological evaluation to diagnose and grade rickets and osteomalacic lesions as well as biochemical tests.

2

Confirm Diagnosis

The clinical diagnosis of XLH should be confirmed by genetic analysis of the PHEX gene with a further work-up aimed at diagnosing the presence and severity of disease complications. First-generation family members of a patient with XLH should also be investigated for XLH.

3

Transitional Care

Genetic counselling should be offered to patients with XLH, especially at the transition from child to adult care and to families planning pregnancies.

4

Specialist Teams

Patient care should be provided by multidisciplinary teams organised by an expert in metabolic bone diseases. Persisting deformity and/or the presence of symptoms interfering with mobility should be considered for surgical treatment by a surgeon with expertise in metabolic bone diseases.

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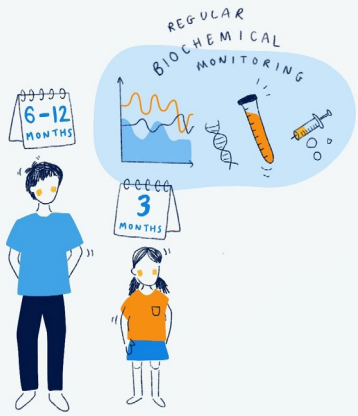

Regular Check-ups

Children with XLH should be seen at least every 3 months during phases of rapid growth (infancy and puberty) as well as after beginning any treatment. Adult patients should be seen every 6 months if receiving treatment, or once a year if not being treated with medication.

6

Hearing Tests

Patients and families should be informed that hearing problems might occur and that any suspicion of hearing impairment should be investigated thoroughly.

Illustrations and design by: @cwhitworthdesign

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ACCESS your BEST Care

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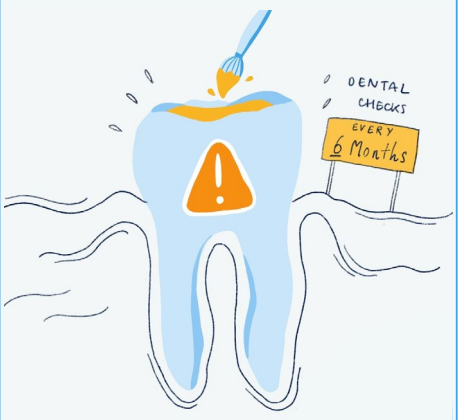

Dental Care

Adults with XLH should have a dental check-up at least every 6 months if they do not already need dental work. Some children with XLH may also need regular dental treatment.

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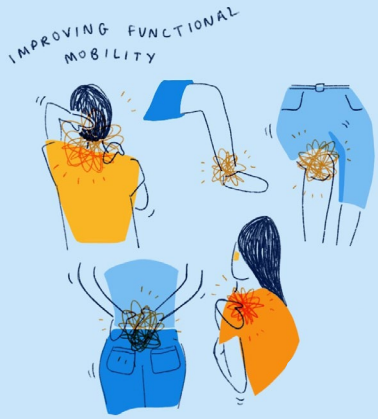

Musculoskeletal Treatment

Interventions aimed at improving functional mobility as well as attempting to reduce bone and joint pain, deformity, stiffness and muscular weakness are recommended.

9

Neurological Assessments

Alongside regular check-ups, we suggest a yearly basic neurological assessment. Where patients do not have any neurological symptoms, we do not recommend further investigations. Patients and families with any concerns about central nervous system function should report them to their consultant so that they can be addressed promptly.

10

Medication

Children and adults should be assessed by their consultant to understand the most suitable treatment available for them. One option for children and symptomatic adults may be a combination of oral phosphate and active vitamin D.

11

New Treatments

Children and adults should be assessed by their consultant to understand the most suitable treatment available for them.

12

Patient Organisations

We highly recommend providing the contact details of patient organisations to XLH patients. These resources can provide invaluable support and up-to-date information for patients and their families.

Citation:

[1] Haffner, Dieter et al. "Clinical practice recommendations for the diagnosis and management of X-linked hypophosphataemia." Nature reviews. Nephrology vol. 15,7 (2019): 435-455. doi:10.1038/s41581-019-0152-5

Disclaimer:

This infographic is not a validated clinical decision aid. For the clinical recommendations on which it is based, please refer to: Haffner D, et al. Nat Rev Nephrol 2019;15(7):435-455.

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