Mapping the lived healthcare experiences of adults with X-linked hypophosphataemia (XLH)

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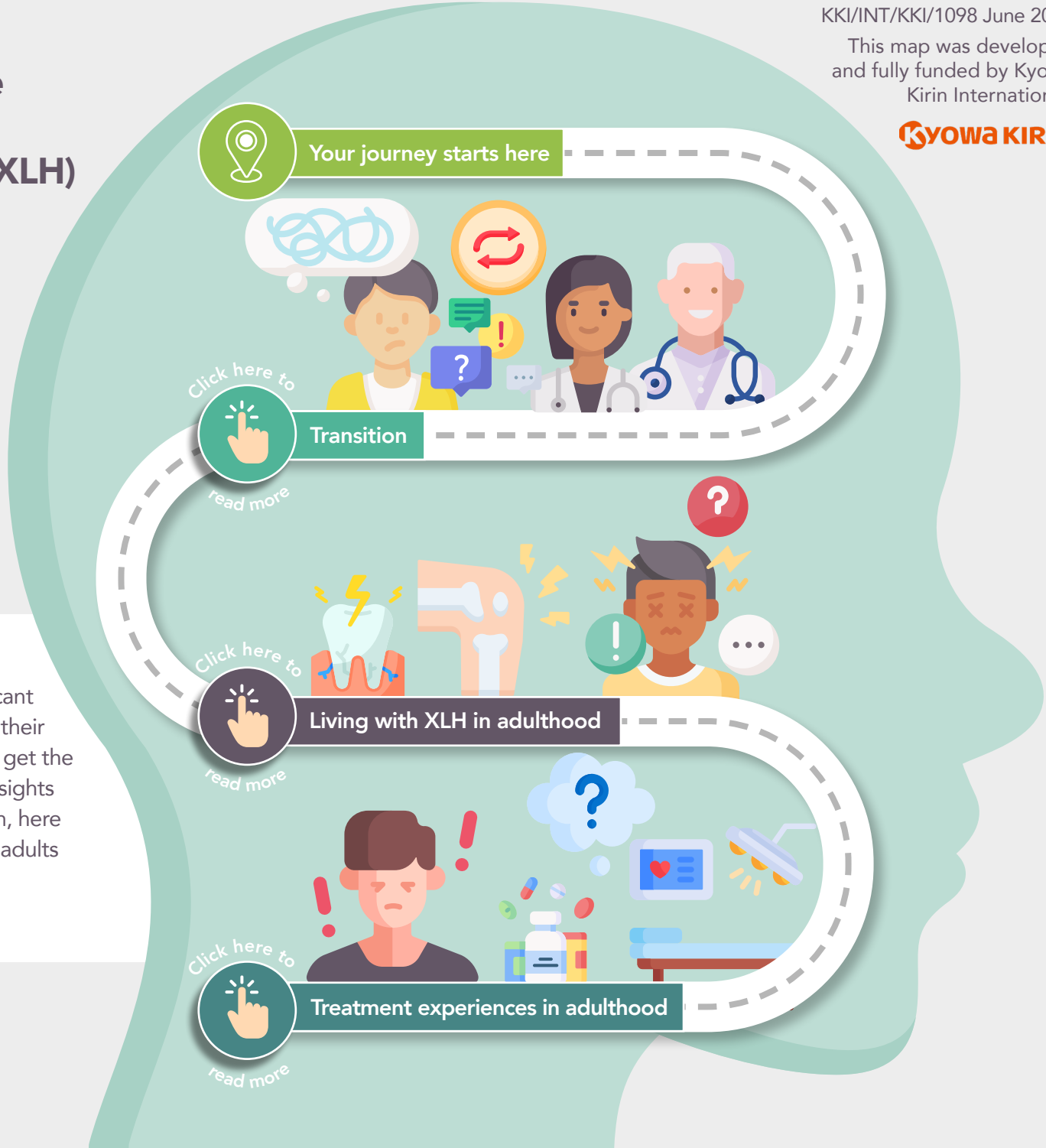

Adults with XLH have often experienced a very challenging childhood, but still experience significant practical and emotional problems. The burden of their condition can be severe, but they may not always get the support they need to live independently. Using insights from patient interviews, surveys and desk research, here we explore how the feelings and unmet needs of adults with XLH are affected by their condition.

Mapping the lived healthcare

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Transition

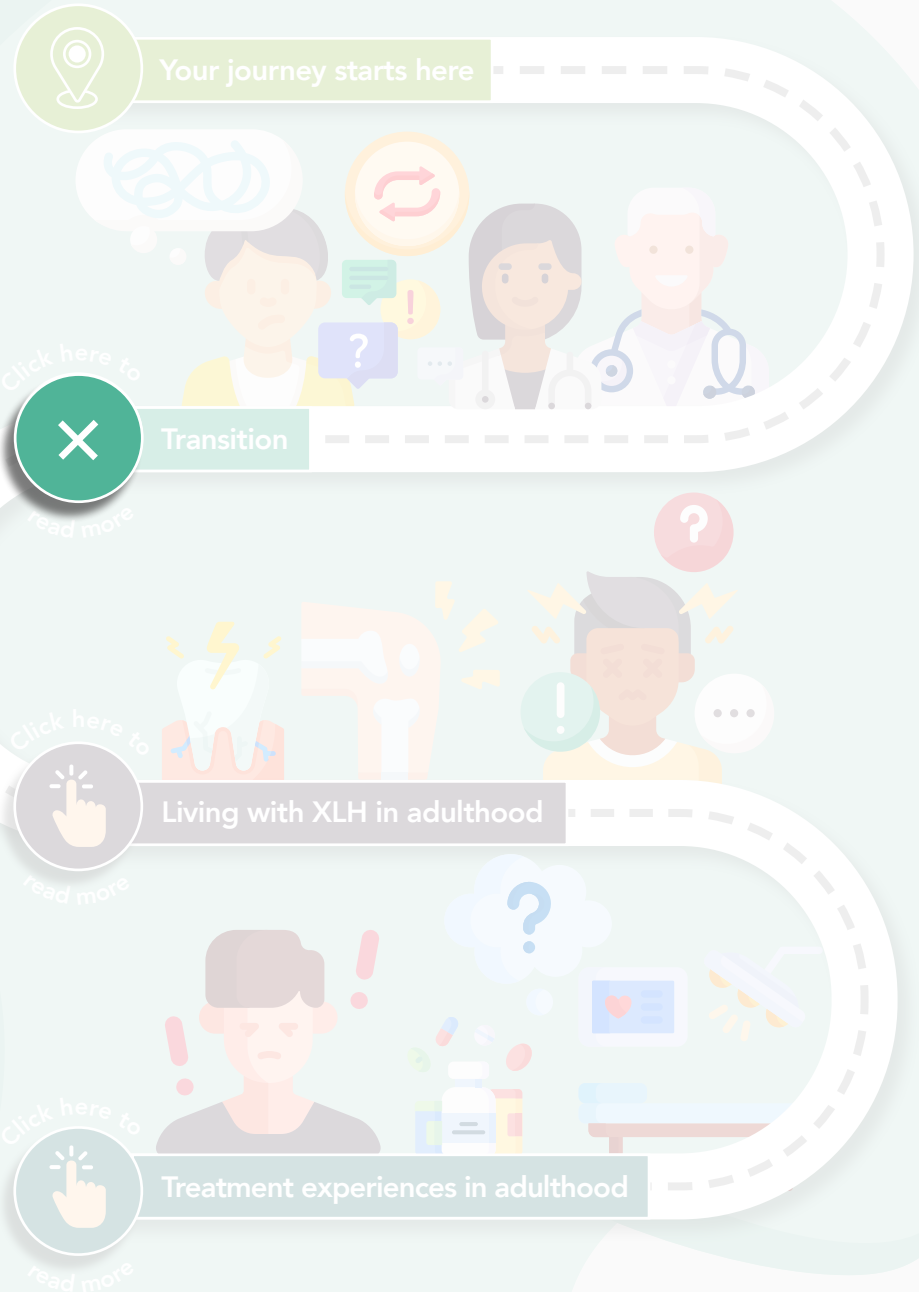
The transition to adult care can be informal, with limited continuity of care. Adult care is often managed by a community-based general practitioner rather than a specialist, which leads to patients repeatedly having to explain their condition and treatment needs.

"The biggest problem is I think, "Where should I go?" Who has expertise when it comes to this disease? I wouldn't see my GP about it. Time has shown that this doesn't make sense" – XLH patient

While the clinical manifestations of XLH in children are fairly well understood, new debilitating symptoms can emerge in adulthood that patients feel unprepared to manage. The lack of information and limited awareness among physicians can lead patients to rely on the internet for education, or to discontinuing care entirely.

"Paediatric patients are attended by specialised centres for a number of years. Then after having built up friendships, bonds and trust with the doctors, when they have to move on to adults, they abandon everything…" – Rheumatologist

"Changing to adult care wasn't easy and we're still struggling" – XLH care partner



Living with XLH in adulthood

Pain can be a constant presence in the life of adults with XLH. It can affect different parts of the body, affecting movement, sleep and almost every aspect of daily life. Patients are prone to dental abscesses, which cause significant discomfort – the loss or discolouration of teeth can also have a big impact on wellbeing. Dentists may not be aware of the specific needs of people with XLH, limiting access to appropriate treatment.

"I can't move freely. I find it difficult to move. I'm in constant pain. I can't keep up with anybody else my age." – Adult with XLH

Other signs of XLH like bowing of the legs and delayed development of bones are even more visible. Patients report being stared at, which can seriously damage their self-esteem and self-confidence. Patients also worry that their children might go through similar experiences if their XLH is passed on genetically. Difficult decisions around whether to have children have to be made, taking into consideration the physical burden on an already struggling body, impact on ongoing treatment and emotional impact of passing on a genetic disease.

"After years of deliberation, my husband and I considered that the physical demands of pregnancy, delivery, and raising children, would just be too hard on my body. For me, also, running the risk of passing XLH on to my children was just too great." – Adult with XLH

Adults with XLH can find it difficult to be independent when they face so many challenges with mobility. This has a knock-on effect on care partners who may have to take time off work to support with tasks like taking patients to medical appointments. The direct and indirect costs of living with XLH can have a huge impact on financial stability, causing stress and anxiety.

"My daily life is limited to surviving at the moment." – Adult with XLH

Adults with XLH are sometimes still treated in paediatric clinics, which can feel inappropriate, but there are very few places that specialise in adult XLH care. Other unmet needs include lack of awareness of XLH among healthcare professionals, lack of coordination between healthcare teams looking after people with XLH and poor access to physiotherapy.

"The nurse came back into the room and she looked at me and asked if I had been in a car accident and I said 'No I'm fine'; she had never seen XLH before." – Adult with XLH

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Treatment experiences in adulthood

Each person's XLH treatment experience may differ, not only in their access to doctors with the expertise to provide effective care but in the availability of psychological support to help them cope with the emotional impact of their condition.

Surgery is an option for some people with XLH, but it's not suitable for everyone.

Adults with XLH manage their pain and stiffness with anti-inflammatory drugs or painkillers, but also report worrying that their high doses of painkillers will stop them noticing if a new problem emerges.

"I understand that there is not much to do for adult patients with this disease." – Adult with XLH