MAPPING LIVED EXPERIENCES OF CHILDREN WITH XLH

Growing up with X-linked hypophosphataemia (XLH) is difficult. In addition to the emotional ups and downs of childhood, patients with this rare condition can face an uphill battle securing a diagnosis and finding a specialist able to manage their significant healthcare needs. Using insights from patient interviews, surveys and desk research, here we explore how the feelings and unmet needs of people with XLH can change over the course of their childhood.

Follow this treasure map to learn more about the experiences of children with XLH.

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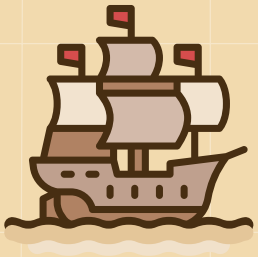

YOUR JOURNEY STARTS HERE

PRE-DIAGNOSIS

DIAGNOSIS

TREATMENT INITIATION

LIVING WITH XLH IN CHILDHOOD

TRANSITION TO ADULT CARE

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KYOWA KIRIN

MAPPING LIVED EXPERIENCES OF CHILDREN WITH XLH

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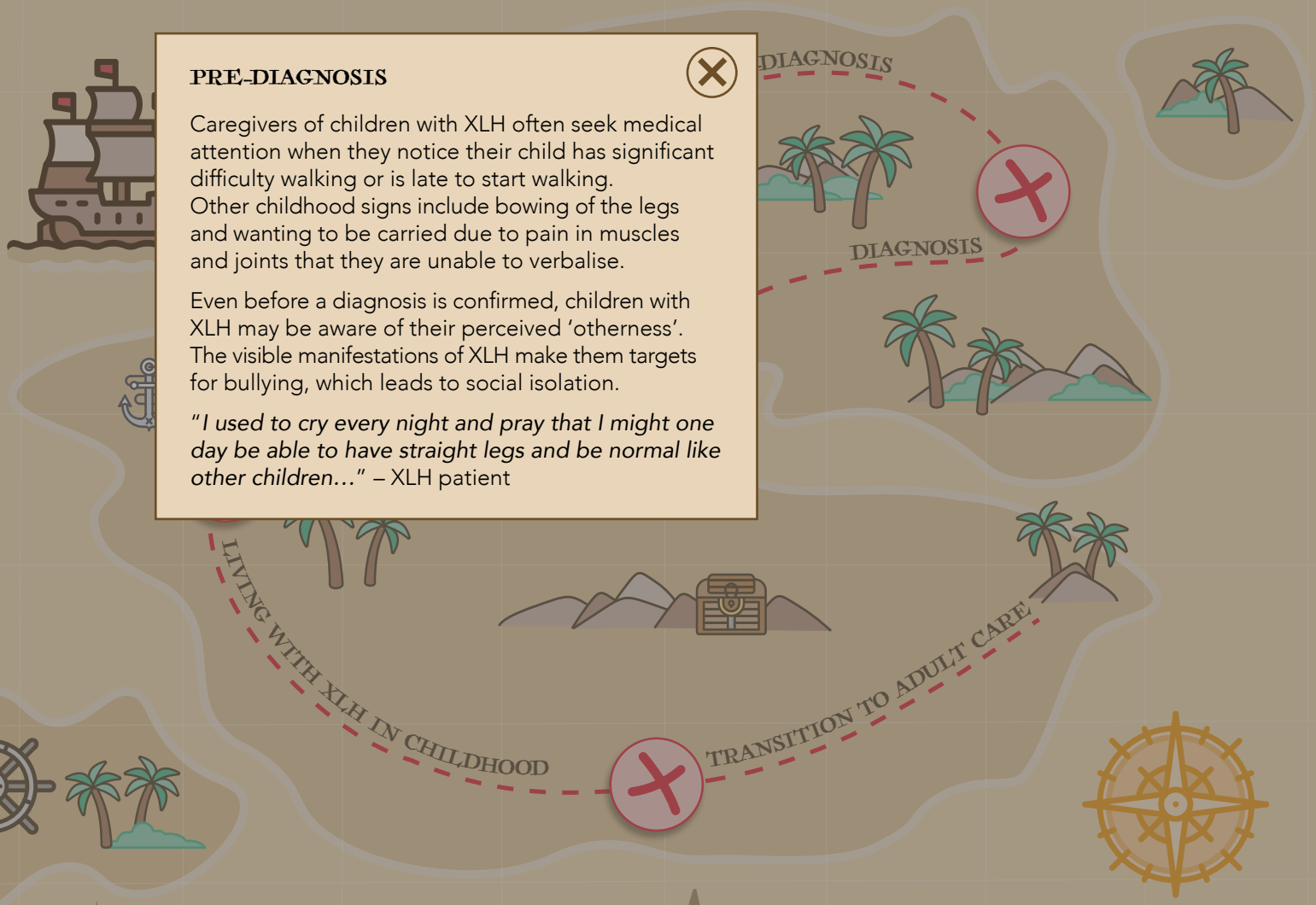

PRE-DIAGNOSIS

Caregivers of children with XLH often seek medical attention when they notice their child has significant difficulty walking or is late to start walking. Other childhood signs include bowing of the legs and wanting to be carried due to pain in muscles and joints that they are unable to verbalise.

Even before a diagnosis is confirmed, children with XLH may be aware of their perceived 'otherness'. The visible manifestations of XLH make them targets for bullying, which leads to social isolation.

"I used to cry every night and pray that I might one day be able to have straight legs and be normal like other children…" – XLH patient

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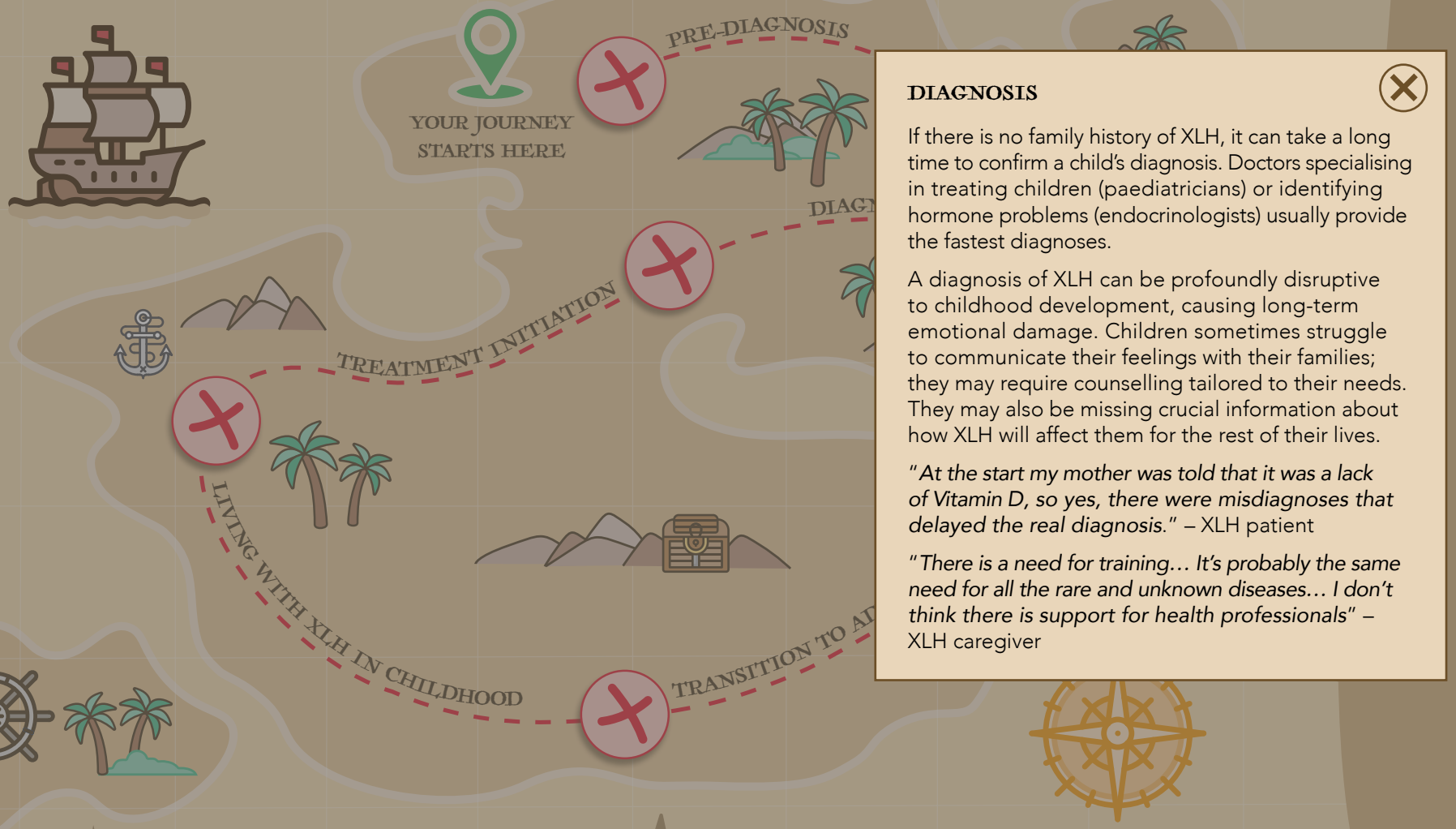

DIAGNOSIS

If there is no family history of XLH, it can take a long time to confirm a child's diagnosis. Doctors specialising in treating children (paediatricians) or identifying hormone problems (endocrinologists) usually provide the fastest diagnoses.

A diagnosis of XLH can be profoundly disruptive to childhood development, causing long-term emotional damage. Children sometimes struggle to communicate their feelings with their families; they may require counselling tailored to their needs. They may also be missing crucial information about how XLH will affect them for the rest of their lives.

"*At the start my mother was told that it was a lack of Vitamin D, so yes, there were misdiagnoses that delayed the real diagnosis.*" – XLH patient

"*There is a need for training… It's probably the same need for all the rare and unknown diseases… I don't think there is support for health professionals*" – XLH caregiver

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Treatments for XLH may have serious side effects, including chronic diarrhoea, stomach pain and recurrent urinary tract infections, which can interrupt children's school and social lives.

The overall burden of treatments, some of which might need to be taken multiple times a day, can be a constant reminder to children that they are unwell.

"I hope children with this disease in the future will not have to spend their childhood in hospital waiting rooms, I hope they will not have to go through what I had to go through." – XLH patient

TRANSITION TO ADULT CARE

IN CHILDHOOD

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LIVING WITH XLH IN CHILDHOOD

XLH may delay bone development to the point some children can't take part in games and activities with their peers.

XLH can have many negative emotional impacts on children: anger, anxiety, confidence issues, depression, distress, fear, guilt, stress, low self-esteem, sleep issues, mental health problems.

Accessing specialist care can require travel, which can be very burdensome and can lead to missing school. This is even more challenging if multiple family members have XLH and caregivers have to balance all their treatment and logistical needs.

"When walking in the street, people point, laugh and talk about you as if you are not there. People underestimate you based on your appearance, so people saw 'different' and equated it with not being able." – XLH patient

"It adds a lot of stress to everyone around us. There is always something to be done, something to take, somewhere to go. It's non-stop." – XLH caregiver

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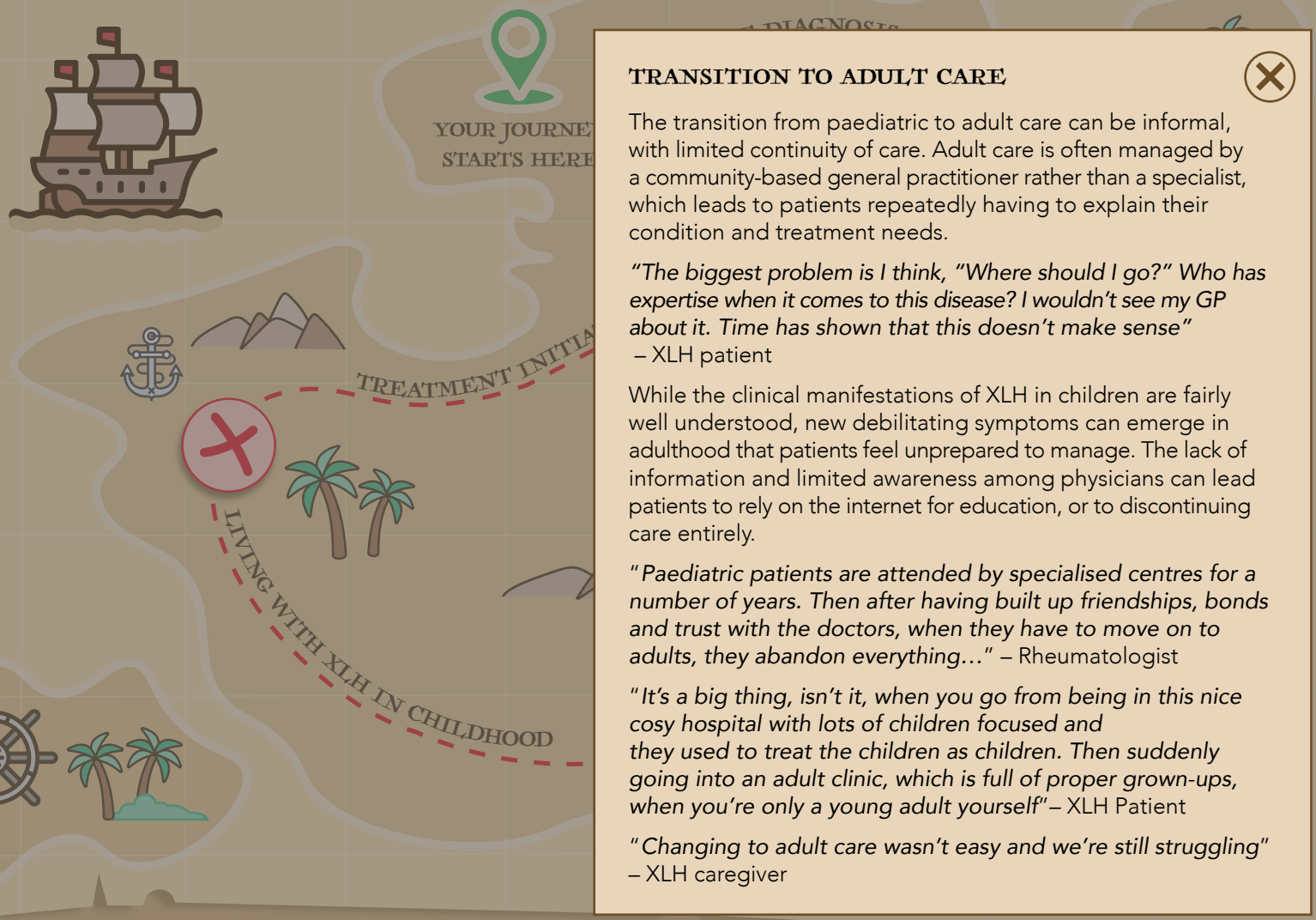

TRANSITION TO ADULT CARE

The transition from paediatric 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

"It's a big thing, isn't it, when you go from being in this nice cosy hospital with lots of children focused and they used to treat the children as children. Then suddenly going into an adult clinic, which is full of proper grown-ups, when you're only a young adult yourself" – XLH Patient

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