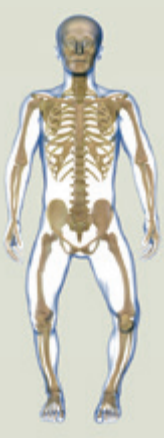
# LIVING WITH XLH

## Results of a nationwide survey of Australians impacted by XLH[1]

## What is XLH?



X-linked hypophosphataemia (XLH) is a rare, lifelong, and often debilitating genetic disease[2,3]. Whilst rare, it is the most common form of hereditary rickets[4]. It's usually passed down from a parent who carries the defective gene but can sometimes appear in individuals with no family history of the disease[5].

XLH is caused by a mutation in the PHEX* gene, leading to elevated circulating levels of the hormone, fibroblast growth factor 23 (FGF23)[2,6]. Excessive FGF23 leads to increased urinary phosphate excretion and reduces intestinal phosphate absorption[6], resulting in chronic hypophosphataemia[8]. Phosphate is a key mineral needed for maintaining many biological processes, including energy metabolism, cell signalling, regulation of protein synthesis, skeletal development, and bone integrity[9]. As a result, people affected by XLH can suffer abnormalities in the bones[2-4], joints[2-4], muscles[3], and teeth[2-4]. The burden of XLH is lifelong and progressive, and it may reduce a person's quality of life[8].

While there is no cure for XLH, therapies aimed at helping to restore phosphate to normal levels within the body may help to improve the symptoms of the disease[2,4].

*Phosphate Regulating Endopeptidase Homolog, X-Linked

## Australian Research

New research revealing the extent of the physical, emotional and social burden of X-linked hypophosphataemia (XLH) on Australian patients[1] has been released to mark Rare Disease Day (Monday, 28 February).

This first-ever, Australia-based research from XLH Australia in collaboration with Kyowa Kirin Australia aims to help increase understanding of what it is like to live with the rare genetic disease[1] which affects an estimated 1 in 20,000 people.[10,11]

### Affects ~1 in 20,000 people [10,11]

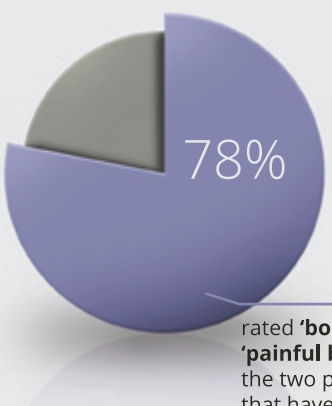


"XLH is far more than bowed legs and short stature, it's a disease that strikes at the physical, emotional and mental wellbeing of those affected."

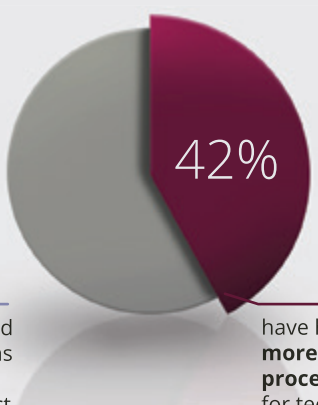
XLH Australia Vice-President, Naomi Ford

## THE BURDEN OF XLH

### Physical




rated **'bone deformities'** and **'painful bones and joints'** as the two physical symptoms that have the greatest impact on their lives.[1]




have had **three or more major dental procedures**, including for teeth removal.[1]


have undergone **five or more surgeries** relating to XLH.[1]

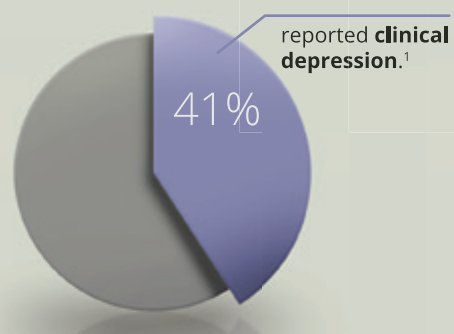
**33%**

have had three or more procedures to **break, reshape and reset leg bones** (osteotomy).[1]

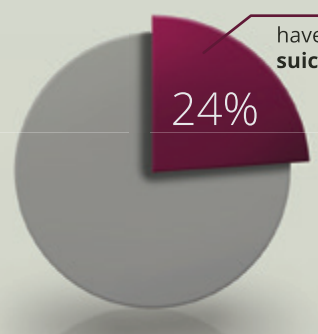
**57%**

of respondents took **longer than five months to walk unassisted** following the bone-breaking surgery.[1]

### Emotional and Mental Wellbeing




reported **clinical depression**.[1]




have reported **suicidal thoughts**.[1]


have admitted to **self harm**.[1]

**65%**

of all adults reported difficulty making **decisions about having children**.[1]

**67%**

feel their **mental health** is significantly **impacted** by the disease.[1]

**76%** of XLH patients have experienced **bullying**.[1]

### Community



**83%**

agreed that a lack of community awareness impacts support services and funding available to them.[1]

## AN ISOLATING DISEASE

"The vast majority of Australians have never heard of XLH, have no idea of what it is, and will not meet anyone living with the rare disease. It can be very isolating."

*XLH Australia Vice-President, Naomi Ford*

## When it comes to XLH, survey respondents are hoping for:[1]


## A PROGRESSIVE DISEASE

"While it's easy to focus on bone deformities in children, this research highlights that XLH is progressive, life-long and pervades almost every aspect of an individual's life."

*XLH Australia Vice-President, Naomi Ford*

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KKAU-XLH-2202083. Date of Preparation February 2022.

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12. Data on file