

<b>XLH</b>	<b>BELGIUM</b>
	<b>BELGIE</b>
	<b>BELGIQUE</b>
	<b>BELGIEN</b>

Association of patients with hypophosphatemic rickets

*General Assembly  
December 2022*

*Assemblée Générale  
Décembre 2022*

*Algemene Vergadering  
December 2022*

 [www.xlhbelgium.be](http://www.xlhbelgium.be)

 [xlhbelgium@gmail.com](mailto:xlhbelgium@gmail.com)

 [XLH Belgium](#)



XLH Belgium aims to contribute to the well-being of people with XLH and their relatives. It pursues the following actions:

*Gathering information and explanations about XLH, the treatments and therapies recommended, and all the available means of help.*

*Communicating and sharing this information and explanations with people with the disease*

*Form a national and international support network*

*Representing patients and their families to medical, political, social security and other authorities.*

XLH Belgium a pour but de contribuer au bien-être des personnes atteintes de XLH et de leurs proches. Elle poursuit les actions suivantes :

Recueillir des informations et des explications sur le XLH, les traitements et thérapies préconisés, et tous les moyens d'aide disponibles.

Communiquer et partager ces informations et explications avec les personnes atteintes de la maladie.

Former un réseau de soutien national et international

Représenter les patients et leurs familles auprès des autorités médicales, politiques, de sécurité sociale, etc.

XLH Belgium wil bijdragen tot het welzijn van mensen met XLH en hun verwanten. Zij voert de volgende acties:

Het verzamelen van informatie en uitleg over XLH, de aanbevolen behandelingen en therapieën, en alle beschikbare hulpmiddelen.

Het communiceren en delen van deze informatie en uitleg met mensen met de ziekte

Een nationaal en internationaal ondersteuningsnetwerk vormen

Patiënten en hun families vertegenwoordigen bij medische, politieke, sociale zekerheids- en andere instanties

## Achievements 2022

### **[1] Evolve the association**

Meeting with XLH Netherlands

### **[2] Communicate about the association**

[www.xlhbelgium.be/](http://www.xlhbelgium.be/)  
Donation page

### **[3] Supporting patients and their relatives**

RIZIV/INAMI hearing

### **[4] Give voice to the association and its members**

Scientific article  
Contact with Kyowa Kirin  
Attendance to XLH alliance encounter

## Réalisations 2022

### **[1] Faire évoluer l'association**

Réunion avec XLH Netherlands

### **[2] Communiquer sur l'association**

[www.xlhbelgium.be/](http://www.xlhbelgium.be/)  
Page de don

### **[3] Soutenir les patients et leurs proches**

Audition par INAMI

### **[4] Porter la voix de l'association et de ses membres**

Article scientifique  
Contact avec Kyowa Kirin  
Présence à la réunion de la XLH Alliance

## Prestaties 2022

### **[1] Ontwikkeling van de vereniging**

Vergadering met XLH Nederland

### **[2] Communicatie over de vereniging**

[www.xlhbelgium.be/](http://www.xlhbelgium.be/)  
donatie

### **[3] Ondersteuning van patiënten en hun familieleden**

RIZIV hoorzitting

### **[4] Een stem geven aan de vereniging en haar leden**

Wetenschappelijke artikel  
Contacten met Kyowa Kirin  
XLH Alliance vergadering

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Association of patients with hypophosphatemic rickets	
<p>X-Linked Hypophosphatemia (XLH) is a rare genetic disorder that affects about one in 20,000 people. The key characteristic of XLH is a low level of phosphate in the blood, called Hypophosphatemia. When a person is affected by XLH, their kidneys do not properly handle Vitamin D and phosphate. This causes a variety of symptoms and disorders that usually affect their bones and teeth.</p> <p><i>Source: Learn About XLH (xlhnetwork.org)</i></p>	

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*Gathering information and explanations about XLH, the treatments and therapies recommended, and all the available means of help.*

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**XLH**  
Vereniging Nederland

<https://xlh-vereniging.nl/>

	<a href="http://www.xlhbelgium.be">www.xlhbelgium.be</a>	
	<a href="mailto:xlhbelgium@gmail.com">xlhbelgium@gmail.com</a>	
	<a href="#">XLH Belgium</a>	

**Partners and other associations:**

**RadiOrg** Umbrella organisation for patient organisations concerning rare disease - [www.radiorg.be](http://www.radiorg.be)

**Orphanet** The portal for rare diseases and orphan drugs - [www.orpha.net](http://www.orpha.net)

**International XLH Alliance** Alliance of international patient groups for people affected by X-linked hypophosphatemia and related disorders.- [xlhalliance.org](http://xlhalliance.org)

Rencontre virtuelle avec XLH Netherlands. Echange sur les difficultés rencontrées pour créer l'association et sur les activités organisées et à venir. Par exemple, organisation de XLH café, un meeting virtuel pour échanger entre patients. Nous retenons cette idée. Nous aurons d'autres échanges en 2023.

Virtuele bijeenkomst met XLH Nederland. Uitwisseling over de moeilijkheden bij de oprichting van de vereniging en over de georganiseerde en toekomstige activiteiten. Bijvoorbeeld de organisatie van het XLH-café, een virtuele bijeenkomst voor uitwisseling tussen patiënten. We houden dit idee. We zullen andere uitwisselingen hebben in 2023.

Virtual meeting with XLH Netherlands. Exchange on the difficulties encountered to create the association and on the activities organised and to come. For example, organisation of XLH café, a virtual meeting to exchange between patients. We will keep this idea. We will have other exchanges in 2023.



Association of patients with hypophosphatemic rickets

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<https://www.xlhbelgium.be/make-a-donation>

## Steun de Vereniging, doe een donatie

U kunt een donatie doen om de vereniging te helpen haar projecten en activiteiten uit te voeren. De vereniging en haar leden danken de schenkers oprecht voor hun vrijgevigheid.

**XLH BELGIUM ASBL VZW BE26 7320 5743 1529**

Wilt u meer weten over onze projecten en activiteiten? Wij nodigen u uit om [onze activiteitenverslagen](#) te lezen.

Donateurs ontheffen XLH Belgium uitdrukkelijk van alle beperkingen met betrekking tot de besteding van de donaties. In ruil voor donaties mogen geen goederen of diensten worden geleverd. Donaties komen niet in aanmerking voor belastingvermindering.

Notre site internet comprend maintenant une page d'information pour ceux qui voudraient nous faire un don.

Onze website bevat nu een informatiepagina voor degenen die een donatie willen doen.

Our website now includes a donation page with all the information needed.



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 XLH Belgium



Consensus Statement | [Published: 28 April 2022](#)

## Interdisciplinary management of FGF23-related phosphate wasting syndromes: a Consensus Statement on the evaluation, diagnosis and care of patients with X-linked hypophosphataemia

[Andrea Trombetti](#), [Nasser Al-Daghri](#), [Maria Luisa Brandi](#), [Jorge B. Cannata-Andía](#), [Etienne Cavalier](#), [Manju Chandran](#), [Catherine Chaussain](#), [Lucia Cipullo](#), [Cyrus Cooper](#), [Dieter Haffner](#), [Pol Harvengt](#), [Nicholas C. Harvey](#), [Muhammad Kassim Javaid](#), [Famida Jiwa](#), [John A. Kanis](#), [Andrea Laslop](#), [Michaël R. Laurent](#), [Agnès Linglart](#), [Andréa Marques](#), [Gabriel T. Mindler](#), [Salvatore Minisola](#), [María Concepción Prieto Yerro](#), [Mario Miguel Rosa](#), [Lothar Seefried](#), [Mila Vlaskovska](#), [María Belén Zanchetta](#) & [René Rizzoli](#) 

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[Nature Reviews Endocrinology](#) **18**, 366–384 (2022) | [Cite this article](#)

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➤ Trombetti, A., Al-Daghri, N., Brandi, M.L. *et al.* Interdisciplinary management of FGF23-related phosphate wasting syndromes: a Consensus Statement on the evaluation, diagnosis and care of patients with X-linked hypophosphataemia. *Nat Rev Endocrinol* **18**, 366–384 (2022). <https://doi.org/10.1038/s41574-022-00662-x>





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L'association participe à l'évaluation de Crysvida par les autorités de santé. Notre association sera brièvement consultée par l'INAMI dans le cadre de la revue du dossier déposé par Kyowa Kirin pour le remboursement de Crysvida chez les adultes. Les supports qui seront présentés sont en annexe de cette présentation

De vereniging is betrokken bij de evaluatie van Crysvida door de gezondheidsautoriteiten. Onze vereniging zal kort worden geraadpleegd door het RIZIV in het kader van de herziening van het door Kyowa Kirin ingediende) dossier voor de terugbetaling van Crysvida bij volwassenen. De presentatie materialen zijn bij dit document gevoegd,

The association is involved in the evaluation of Crysvida by the health authorities. Our association will be briefly consulted by the INAMI in the framework of the review of the file submitted by Kyowa Kirin for the reimbursement of Crysvida in adults. The presentation material is attached in the annex of this slide deck,

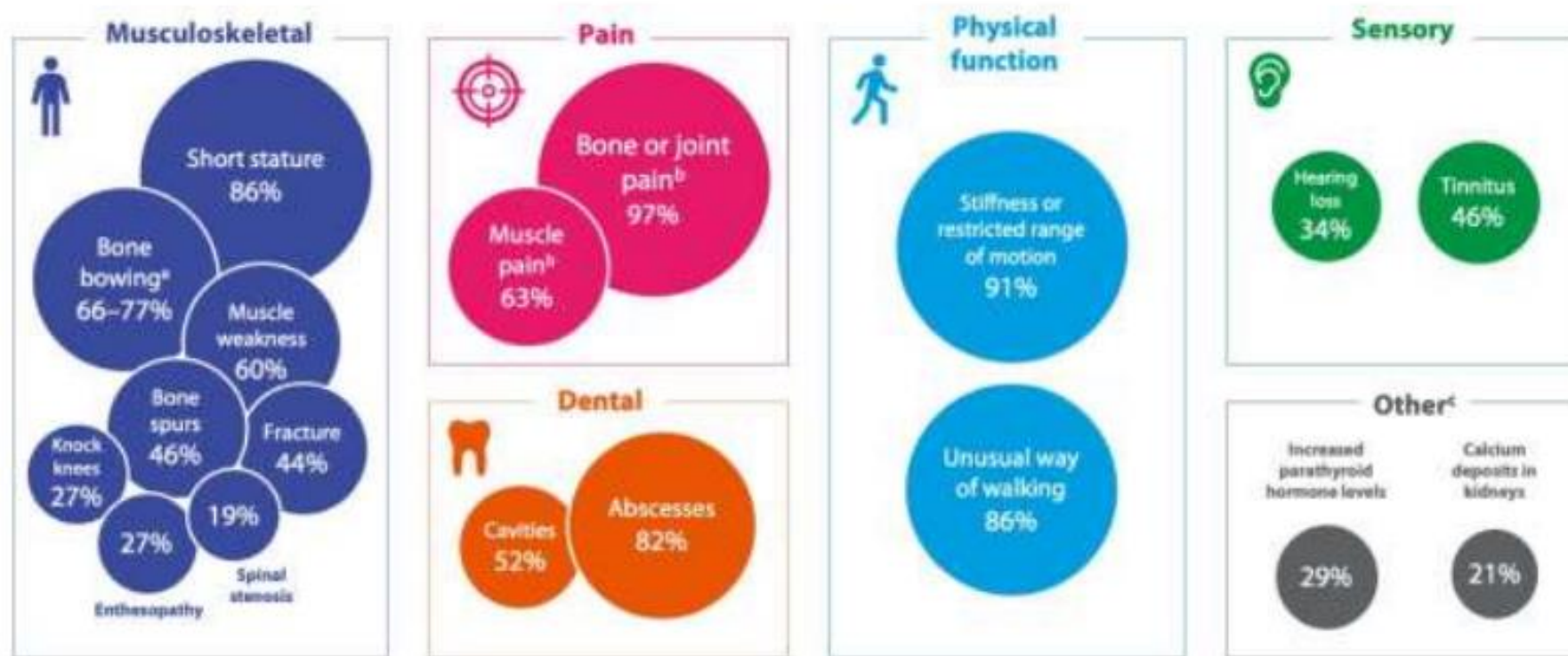
## PATIENT PERSPECTIVE

A better description of XLH in adults:

- X-linked hypophosphatemia (XLH) is a rare, lifelong, progressive disease characterised by renal phosphate wasting and abnormal bone mineralization (SH Lo et al, 2020)
- **X-Linked Hypophosphatemia, Not Only a Skeletal Disease But Also a Chronic Inflammatory State** (Meaux et al., 2022)



## The unrecognized burden of XLH in adults

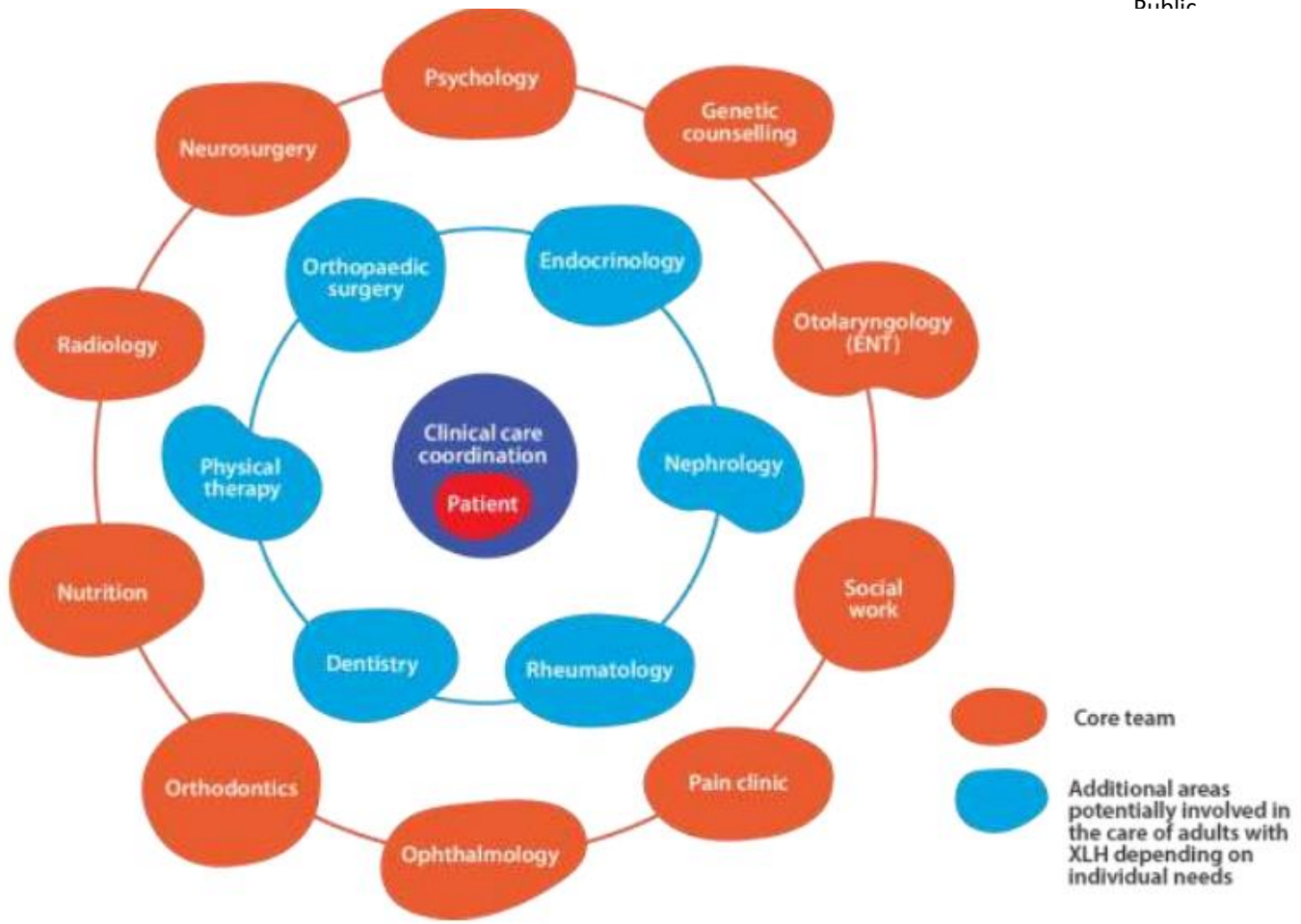


**Figure 1. Proportion of adults who reported experiencing signs and symptoms associated with XLH.**

Data shown are from the largest published study on burden of disease in adults with XLH (n = 232).<sup>3</sup>

<sup>a</sup>66% of adults experienced bowing of the thigh bone; 77% experienced bowing of the long bones of the lower leg. <sup>b</sup>Pain experienced in the year prior to participating in the survey. <sup>c</sup>Conditions thought to be related to use of vitamin D and phosphate-replacement therapies.

Source: The unrecognized burden of XLH in adults. A call to action – white paper P. Harvengt, L. Seefried, M. Smith, R. Keen (2020).



XLH requires a multidisciplinary approach

BUT...

Do we have appropriate access to the specialities that we need ?

- Pain killers: up to opioids
- Dentistry: nothing more than general population
- Physiotherapy: nothing more than general population
- Endocrinology: nothing more than the general population
- .....

We need better access to healthcare !!!

### Financial pressure

- Medical and dental bills
- Insurance
- Home modifications
- Special equipment

### Challenges at home

- Support needed from family and friends
- Mental health support
- Family planning

### Challenges at work

- Type of work / career
- Company support
- Time off / flexible working
- Early retirement



### Lack of awareness

- Low awareness among healthcare professionals, policy makers and the general public
- Lack of resources for patients

### Pain

- Painful symptoms that interfere with daily life
- Need for ongoing pain management

### Limited mobility

- Progressive deterioration of physical function
- Difficulty getting around
- Need for mobility aids

Figure 5. Factors contributing to the burden of XLH in adulthood.

## Conclusions:

- Burden of disease in adults is significant (similar to other bone disease like OI)
- Conventional treatment is not a realistic option (QoL- and endocrinology-related)
  
- What we need
- better access to treatment (as a whole)
  - A reasonable price structure for the new treatment options
  - More than 5 minutes hearing (cfr level of expertise available from patient experts)

# XLH

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# XLH

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Association de patients atteints de rachitisme hypophosphatémique

L'hypophosphatémie liée à l'X (XLH) est une maladie génétique rare qui touche environ une personne sur 20 000. La principale caractéristique de la XLH est un faible taux de phosphate dans le sang, appelé hypophosphatémie. Lorsqu'une personne est atteinte de XLH, ses reins ne traitent pas correctement la vitamine D et le phosphate. Cela provoque une variété de symptômes et de troubles qui affectent généralement les os et les dents.

Source : *En savoir plus sur le XLH (xlhnetwork.org)*

### **Partenaires et autres associations :**

**RadiOrg** Organisation faîtière des associations de patients concernant les maladies rares - [www.radiorg.be](http://www.radiorg.be)

**Orphanet** Le portail des maladies rares et des médicaments orphelins - [www.orpha.net](http://www.orpha.net)

**International XLH Alliance** Alliance de groupes internationaux de patients pour les personnes touchées par l'hypophosphatémie liée à l'X et les troubles connexes. - [xlhalliance.org](http://xlhalliance.org)

Notre association s'appelle XLH Belgium, et a pour but de contribuer au bien-être des personnes atteintes de XLH et de leurs proches. Elle poursuit les actions suivantes :

*Rassembler des informations et des explications sur le XLH, les traitements et thérapies recommandés, et tous les moyens d'aide disponibles.*

*Communiquer et partager ces informations et explications avec les personnes atteintes de la maladie.*

*Former un réseau de soutien national et international*

*Représenter les patients et leurs familles auprès des autorités médicales, politiques, de sécurité sociale et autres.*



[www.xlhbelgium.be](http://www.xlhbelgium.be)



XLH  
Belgique





# XLH

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Vereniging van patiënten met hypofosfatemische rachitis

X-gebonden hypofosfatemie (XLH) is een zeldzame genetische aandoening die bij ongeveer één op de 20.000 mensen voorkomt. Het belangrijkste kenmerk van XLH is een laag fosfaatgehalte in het bloed, Hypofosfatemie genoemd. Als iemand lijdt aan XLH, kunnen de nieren niet goed omgaan met vitamine D en fosfaat. Dit veroorzaakt een verscheidenheid aan symptomen en aandoeningen die meestal gevolgen hebben voor hun botten en tanden.

*Bron: Meer informatie over XLH ([xlhnetwork.org](http://xlhnetwork.org))*

Onze vereniging heet XLH Belgium en heeft tot doel bij te dragen tot het welzijn van mensen met XLH en hun verwanten. Zij voert de volgende acties:

*Het verzamelen van informatie en uitleg over XLH, de aanbevolen behandelingen en therapieën, en alle beschikbare middelen om hulp te bieden.*

*het doorgeven en delen van deze informatie en uitleg met mensen met de ziekte*

*Een nationaal en internationaal ondersteunend netwerk vormen*

*Vertegenwoordiging van patiënten en hun gezinnen bij medische, politieke, sociale zekerheids- en andere instanties.*

### **Partners en andere verenigingen:**

**RadiOrg** Overkoepelende organisatie voor patiëntenorganisaties op het gebied van zeldzame ziekten - [www.radiorg.be](http://www.radiorg.be)

**Orphanet** Het portaal voor zeldzame ziekten en weesgeneesmiddelen - [www.orpha.net](http://www.orpha.net)

**International XLH Alliance** Alliantie van internationale patiëntengroepen voor mensen die lijden aan X-gebonden hypofosfatemie en aanverwante aandoeningen. - [xlhalliance.org](http://xlhalliance.org)



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XLH België

