DISEASE BURDEN OF X-LINKED HYPOPHOSPHATEMIA

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Introduction

X-linked hypophosphatemia (XLH) is a rare genetic disease, a serious, debilitating and deforming condition, characterized by renal phosphate wasting. XLH is caused by a mutation in the PHEX (phosphate regulating endopeptidase homolog X-linked) gene resulting in enhanced secretion of the phosphaturic hormone FGF23 and leading defective bone mineralization, rickets and impaired physical function. This disease, which affects 1:20.000 people, results in impaired quality of life (QoL) of patients, since, despite the treatments, they suffer progressive and debilitating complications. (Haffner D, et al. Nat Rev Nephrol. 2019)

Objective

The main objective of the study was to determine QoL of patients, children and adults suffering XLH, as well as their caregivers.

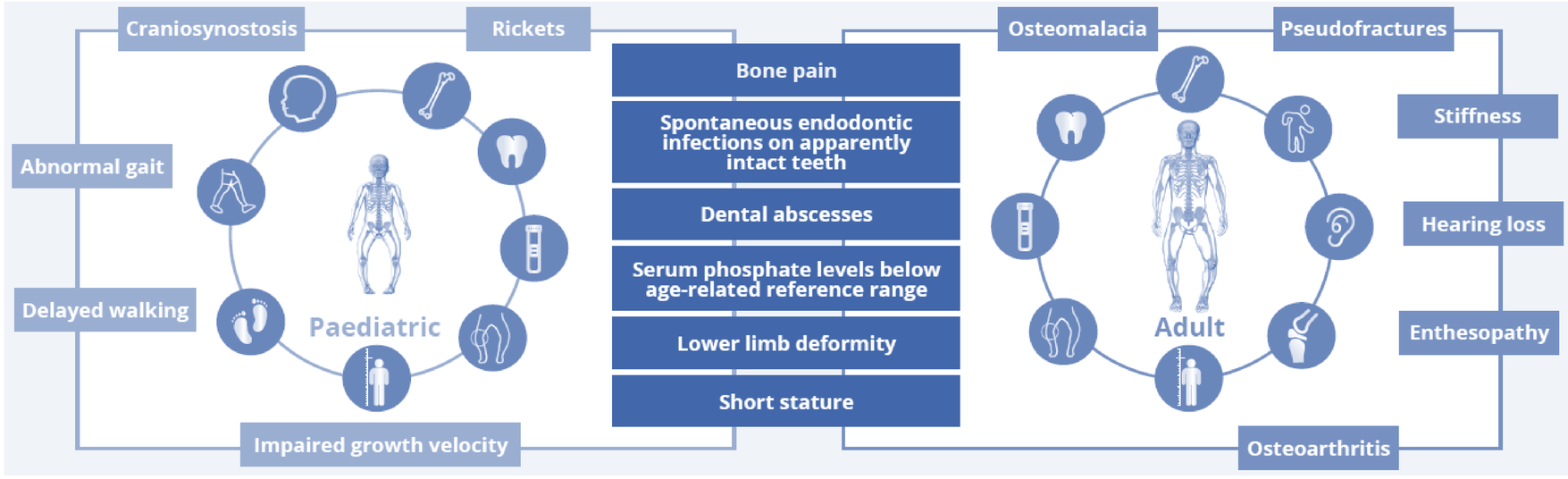


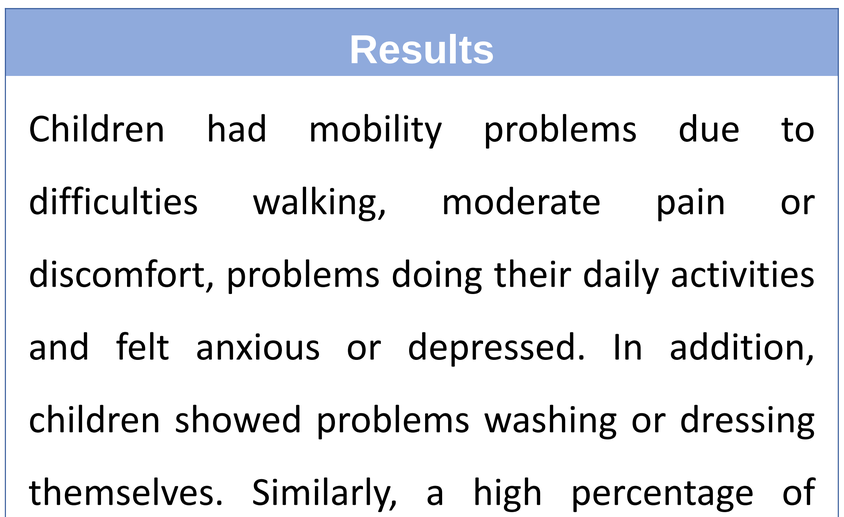
Figure 1. Clinical presentation and complications in pediatric and adult XLH patients

Methods

A multicentre, observational and cross-sectional study has been carried out by a scientific committee consisted of 17 health care professionals experts in XLH. QoL was assessed with the following EuroQol-5 Dimensions (EQ-5D) questionnaires: the EQ-5D-3L proxy (completed by caregivers or parents of children under 12 years), the EQ-5D-3L (completed by patients aged between 12 and 18 years), and the EQ-5D-5L (completed by adult patients and caregivers). The total number of participants for this project was 50 patients, 21 children and their corresponding caregivers, and 29 adults.

Results

Children had mobility problems due to difficulties walking, moderate pain or discomfort, problems doing their daily activities and felt anxious or depressed. In addition, children showed problems washing or dressing themselves. Similarly, a high percentage of adults were unable to walk, suffered extreme pain, showed difficulties to carry out their daily activities, and reported anxiety and depression. Moreover, the results of QoL for caregivers and parents were slightly lower than the general Spanish population.



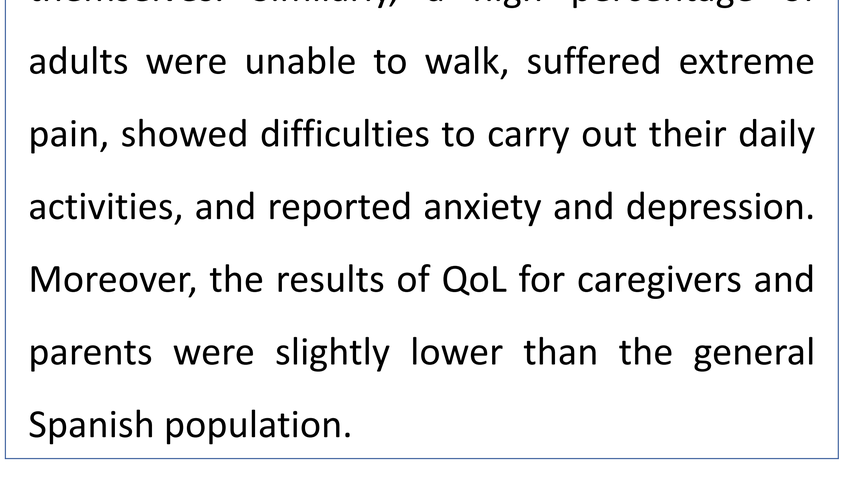


Figure 2 and 3. Comparison of the mean values of the EQ-5D-3L and EQ-5D-5L index and the visual analogue scale(VAS) in patients with XLH and their caregivers. Comparison with general population only can be done with XLH adults and caregivers results.

Items	Options	QoL in Children with XLH (%) (n=21)	QoL in adults with XLH (%) (n=29)	QoL in general Spanish population (%) (n=20,587)
Mobility	No problems	38.10	6.90	85.72
	Minor problems	61.90	37.93	6.25
	Moderate problems		31.03	4.76
	Severe problems		20.69	2.45
	Extreme problems	0.00	3.45	0.82
Self- care	No problems	90.48	48.28	93.78
	Minor problems	9.52	31.03	2.52
	Moderate problems		17.24	1.77
	Severe problems		3.45	0.93
	Extreme problems	0.00	0.00	1.01
Usual activities	No problems	66.67	20.69	88.86
	Minor problems	33.33	34.48	4.87
	Moderate problems		31.03	3.22
	Severe problems		13.79	1.58
	Extreme problems	0.00	0.00	1.46
Pain/ Discomfort	No problems	38.10	13.79	75
	Minor problems	-	10.34	12.51
	Moderate problems	61.90	34.48	8.86
	Severe problems	0.00	37.93	4
	Extreme problems	-	3.45	0.41
Anxiety/ Depression	No problems	76.19	34.48	84.97
	Minor problems	-	31.03	8.6
	Moderate problems	23.81	24.14	4.25
	Severe problems	0.00	10.34	1.68
	Extreme problems	-	0.00	0.41

Table 1. Quality of life in children and adults with XLH according to the results obtained in the EQ-5D-3L and 5Q-5D-5L questionnaire. Only XLH adults results can be compared to the general Spanish population according to the results of the National Health survey of the years 2011-2012.

Conclusion

In conclusion, children and adults with XLH, showed an important deterioration in QoL which was worse than the general Spanish population. These findings support the need for new treatments, as in many cases, conventional treatment does not prevent disease complications and the impairment of QoL.

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