

**X-LINKED HYPOPHOSPHATAEMIC RICKETS (XLH) PREVALANCE QUESTIONNAIRE****(A one-off survey conducted by the Australian Paediatric Surveillance Unit)**APSU Office  
Use Only

If you have any questions about this form please contact:

Prof Craig Munns on (02) 9845 3200 or [craig.munns@health.nsw.gov.au](mailto:craig.munns@health.nsw.gov.au) or APSU (02) 9845 3005 or [SCHN-APSU@health.nsw.gov.au](mailto:SCHN-APSU@health.nsw.gov.au)Date of  
Report:*Instructions: Please answer each question by ticking the appropriate box or writing your response in the space provided.*

Y = Yes, N = No, DK=Don't Know; NA = Not Applicable

Version Dated:  
V3\_17.06.2020**1. X-LINKED HYPOPHOSPHATAEMIC RICKETS CASE DEFINITION (< 18 years )****Rickets during childhood (Please indicate criteria present):**

- Radiological evidence rickets
- Alkaline phosphatase (ALP) above the normal age and gender-matched limits of the local laboratory range
- Serum phosphate below normal limits of the local laboratory range

**AND (at least 1)**

- Pathogenic mutation in the PHEX gene (Result: \_\_\_\_\_ City where test performed: \_\_\_\_\_)
- FGF23 levels above limits of the local laboratory range
- Family history supporting X-linked inheritance (*if yes*, in whom: \_\_\_\_\_)

**REPORTING CLINICIAN'S DETAILS:**

2. APSU Dr Code/Name: \_\_\_\_\_ / \_\_\_\_\_ 3. Date questionnaire completed: \_\_\_\_ / \_\_\_\_ / \_\_\_\_\_

**PATIENT DETAILS:**

4. First 2 letters of first name:   5. First 2 letters of surname:
6. Date of Birth: \_\_\_\_ / \_\_\_\_ / \_\_\_\_\_ 7. Sex:  Male  Female  Indeterminate
8. Date of diagnosis: \_\_\_\_ / \_\_\_\_ / \_\_\_\_\_ 9. Post code of family:

If this patient is primarily cared for by another physician who you believe will report the case, please complete the questionnaire details above this line and return to the APSU. Please keep the patient's name and other details in your records.

If no other report is received for this child we will contact you for information requested in the remainder of the questionnaire.

The primary clinician caring for this person is: **Name:** \_\_\_\_\_**Hospital:** \_\_\_\_\_**10. BIOCHEMICAL DATA AT DIAGNOSIS**

Parameter	Date	Units	Normal range	Don't know (DK)
25-Hydroxyvitamin D				
Alkaline phosphatase				
Total calcium				
Albumin				
Serum Phosphate				
Parathyroid hormone				
Urine TMP/GFR				
Urine calcium: creatinine ratio				
FGF23				

**11. CURRENT BIOCHEMICAL DATA**

Parameter	Date	Units	Normal range	Don't know (DK)
25-Hydroxyvitamin D				
Alkaline phosphatase				
Total calcium				
Albumin				
Serum Phosphate				

Parathyroid hormone			
Urine TMP/GFR			
Urine calcium: creatinine ratio			
FGF23			

## 12. ORAL HEALTH

12. Frequency of dental review:  NA  6 monthly  12 monthly  Other (please specify): \_\_\_\_\_

12a. Age when teeth first appeared (months): \_\_\_\_\_  NA

12b. Tooth abscess:  Yes  No Number: \_\_\_\_\_  NA Age at first tooth abscess: \_\_\_\_\_

12c. Dental extraction:  Yes  No Number: \_\_\_\_\_  NA Age when first tooth extracted: \_\_\_\_\_

12d. Dental capping:  Yes  No Number: \_\_\_\_\_  NA Age when first tooth capped: \_\_\_\_\_

12e. Other dental history:  Yes  No  NA

12e.i. If Yes, specify:  Toothache  Caries  Extractions

Hypodontia; number missing teeth (excluding 8's) \_\_\_\_\_  Other (please specify) \_\_\_\_\_

## 13. CLINICAL FEATURES PRESENT AT ANY TIME

System	Clinical Feature	At any time		
Musculoskeletal	Short stature (height <3 <sup>rd</sup> centile)	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Bone or joint pain	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Muscle pain	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Bowing of legs	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Flaring of wrists	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Motor delay or Reduced activity levels	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Abnormal gait	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Use of mobility aid	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Myopathy/ Muscle weakness	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Rachitic chest/ deformed ribs	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Fractures (number)	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK <i>If yes, number:</i>
	Pseudofractures (number)	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK <i>If yes, number:</i>
	Fractures with delayed healing (number)	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK <i>If yes, number:</i>
	Scoliosis	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Kyphosis	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Craniosynostosis	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Xanthoma	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Arthritis	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
Spinal Stenosis	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK	
Renal	Nephrocalcinosis	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Kidney stones	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
Other	Hearing Impairment	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK
	Hyperparathyroidism	<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> DK

14. Other clinical features (please specify): \_\_\_\_\_

**15. TREATMENT OF X-LINKED HYPOPHOSPHATAEMIC RICKETS**

15. Was the child/adult commenced on medical treatment for XLH?  Yes  No  Don't Know

15a. If Yes, what is the most recent medication used? (please complete table below):

Medication	Indication	Dose (units)	Frequency	Date started

15b. Was the child/adult treated with Burosumab?  Yes  No  Don't Know

15c. If Yes, when was treatment commenced? \_\_\_\_\_

15d. If Yes, is treatment ongoing?  Yes  No  Don't Know

16. Which health professionals (medical and allied health) have ever been involved in care?

- Physician                       Paediatrician                       Geneticist                       Orthopaedic surgeon  
 Dentist                               Physiotherapist                       Occupational therapist                       Psychologist  
 Other (please specify): \_\_\_\_\_

17. How many times has the child/adult been hospitalised in the last 12 months?:  1  2  3  4 or more

17a. Reason(s) for hospitalisation and length of stay, including orthopaedic surgery (please complete table below):

Admission #	Reason for hospitalisation	Length of Stay (days)
1		
2		
3		
4		
5		
6		
7		

18. Any other relevant clinical information: \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

**Thank you for your help with this research project.**  
**Please return this questionnaire to the APSU via email: [SCHN-APSU@health.nsw.gov.au](mailto:SCHN-APSU@health.nsw.gov.au)**  
**or fax to 02 9845 3082, even if you don't complete all items.**  
**Australian Paediatric Surveillance Unit, Kid's Research (SCHN), Locked Bag 4001, Westmead NSW 2145.**

The APSU is affiliated with the Royal Australasian College of Physicians (Paediatrics and Child Health Division)  
 and Faculty of Medicine and Health, The University of Sydney.  
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 This study has been approved by a Human Research Ethics Committee properly constituted under NHMRC guidelines.