X-LINKED HYPOPHOSPHATAEMIC RICKETS (XLH) PREVALENCE QUESTIONNAIRE

(A one-off survey conducted by the Australian Paediatric Surveillance Unit)

If you have any questions about this form please contact:
Prof Craig Munns on (02) 9845 3200 or craig.munns@health.nsw.gov.au or APSU (02) 9845 3005 or SCHN-APSU@health.nsw.gov.au

APSU Office Use Only

Date of Report:

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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

1. X-LINKED HYPOPHOSPHATAEMIC RICKETS CASE DEFINITION (diagnosed since January 1 2014)

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 <3rd centile)	☐ Yes ☐ No ☐ DK
	Bone or joint pain	☐ Yes ☐ No ☐ DK
	Muscle pain	☐ Yes ☐ No ☐ DK
	Bowing of legs	☐ Yes ☐ No ☐ DK
	Flaring of wrists	☐ Yes ☐ No ☐ DK
	Motor delay or Reduced activity levels	☐ Yes ☐ No ☐ DK
	Abnormal gait	☐ Yes ☐ No ☐ DK
	Use of mobility aid	☐ Yes ☐ No ☐ DK
	Myopathy/ Muscle weakness	☐ Yes ☐ No ☐ DK
	Rachitic chest/ deformed ribs	☐ Yes ☐ No ☐ DK
	Fractures (number)	☐ Yes ☐ No ☐ DK ***If yes***, number:
	Pseudofractures (number)	☐ Yes ☐ No ☐ DK ***If yes***, number:
	Fractures with delayed healing (number)	☐ Yes ☐ No ☐ DK ***If yes***, number:
	Scoliosis	☐ Yes ☐ No ☐ DK
	Kyphosis	☐ Yes ☐ No ☐ DK
	Craniosynostosis	☐ Yes ☐ No ☐ DK
	Xanthoma	☐ Yes ☐ No ☐ DK
	Arthritis	☐ Yes ☐ No ☐ DK
	Spinal Stenosis	☐ Yes ☐ No ☐ DK
Renal	Nephrocalcinosis	☐ Yes ☐ No ☐ DK
	Kidney stones	☐ Yes ☐ No ☐ DK
Other	Hearing Impairment	☐ Yes ☐ No ☐ DK
	Hyperparathyroidism	☐ Yes ☐ No ☐ 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 hospitalistation	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
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.