MAR Neurology, Neurosurgery & Psychology (2026) 10:1

Case Report

A Case Report of Hypophosphatemic Rickets with Thoracic Myelopathy

Kamal Abdulazeez Mohammadamin *[1], Amanj Jamal Khidhir Pastaksor [2]

1. Specialist Orthopedic Surgeon MBChB, HDOS, FRCS (Edinburgh).

2. MBChB, DIM, FICMS (Neurology) at Hawler Teaching Hospital.

*Correspondence to: Kamal Abdulazeez Mohammadamin, Specialist Orthopedic Surgeon MBChB, HDOS, FRCS (Edinburgh).

Copyright

Received: 18 January 2026

Published: 01 February 2026

DOI: https://doi.org/10.5281/zenodo.18396992

Abstract

Hypophosphatemic ricket is x linked dominant disease due to mutation in PHEX gene, characterized by renal phosphate wasting (i.e. inability to reabsorb phosphate).

Knowledge of phosphate metabolism is critical to determine the underlying causes of hypophosphatemia and to facilitate appropriate treatment.

Keywords: *hypophosphatemic ricket, PHEX gene, Myelopathy.*

Introduction

Rickets is a metabolic disorder due to defect in mineralization of osteoid matrix caused by inadequate calcium and phosphate with failure of calcification in zone of provisional calcification. (1)

There are many types of rickets including nutritional deficiency of vitamin D, vitamin D dependent rickets and familial hypophosphatemic rickets (Familial HPR). (1).

Familial HPR is the most common heritable phosphate wasting genetic disorder.

It is an X linked genetic disorder with dominant inheritance, caused by a mutation in the PHEX gene which leads to inappropriately elevated FGF23 levels.(2)

FGF23 is the major regulator of phosphorous homeostasis which causes an increase in urinary phosphate excretion.

The condition starts in infancy or soon before or after, and causes bony deformity of the lower limbs if it is not recognized and treated.

During infancy affected children look normal but deformities of the lower limbs such as genu valgus or varus develop when they begin to walk and growth is below normal. (2)

During adulthood there is a tendency to develop heterotopic bone formation around some of the larger joints and in the longitudinal ligaments of the spinal canal that may give rise to enthesopathies and neurological symptoms via spinal cord compression.

This case study aimed at reporting a severe and very rare case of a patient with familial HPR describing its progressive nature in association with dorsal cord myelopathy with normal MRI.

Case Study

A one year old girl, a product of non-consanguineous parents, diagnosed as subluxated hip joints treated by spica cast for eight weeks.

There was radiological improvement of hip joints after spica treatment. She could walk at the age of 2.5 years with weakness i.e. delayed motor milestones, gradually she developed short stature (90cm height) and progressive deformities in both lower limbs in form of coxa vara and genu valgus.

Two of her sisters also have the same condition, her father had bowing of legs but ambulant and had a milder form of disease.

Blood Investigations showed normal serum calcium, magnesium, active B12 level, and serum folate, Copper and Zinc. Normal Thyroid function tests and parathyroid hormones but low serum phosphate and vitamin D level with high alkaline phosphatase.

Follow up radiological findings (X ray?) confirmed to be rickets associated with thoracic scoliosis and right sided convexity (cobbs angle 30).

Whole exome sequencing was conducted and detected a pathogenic variant in PHEX gene with subsequent increased expression of Exon/Intron (FGF 23).

At the age of 15 seen she started to develop progressive weakness of lower limbs that gradually made her unable to stand associated with numbness in both lower limbs upto the upper thighs. She had normal sphincters. Gradually she had deformed lower limbs and hirsutism.

Neurological exam showed floppy lower limbs, bilateral hyperreflexia, non-sustained ankle clonus and nonresponding plantar reflexes, sensory level bilateraly at L2 dermatome.

	Calcium mg/dl	Phosphate mg/dl	Alkaline phosphatase IU/L	Parathyroid hormone Pg./ml	Vitamin D ng/ml	Urinary Phosphate excretion mg/day
Normal range	(8.6-10.2)	(2.7-4.5)	<290	(15-65)	(30-70)	600-800
Sept 2022	9	2.6	790	59	11	1900
Dec 2022	8	2.7	731	32	37	
Feb 2023	9.4	1.9	588	52	19	2010
May 2023	8.9	2.1	602	48	12	
Jan 2024	9	1.5	677	45	23	1980

Table 1 Blood levels and their variable responses following treatment with phosphate and vitamin D

Head CT was normal.

Treatment with phosphate and calcitriol was carried out intermittently to a level to normalize levels of vitamin D and serum ALPase.

The plan started for correction of bone and joint deformities but she developed slowly progressive bilateral lower limb weakness and loss of sensation up to level of L2 over 2 years and eventually she became bedridden.

Dorsal Spine MRI showed no Neural tube stenosis and there was no sign of ligamentum flavum hypertrophy or abnormal calcification. So there was no compression on spinal cord and thus making surgical intervention useless.

Nerve Conduction studies and EMG were normal.

Diagnosis confirmed to be hypophosphatemic ricket with Dorsal cord myelopathy without compression.

Discussion

Familial hypophasphatemic rickets is due to renal wasting of phosphate. Inherited pattern is X-linked dominant. It is due to mutation in the phosphate regulating gene PHEX (Phosphate regulating gene with Homology to Endopeptidase) located on the x chromosome. Phosphorus plays a vital role in growth and development, bone formation and cellular metabolism (2).

In young infant normal concentrations of phosphate are necessary for adequate skeletal mineralization, so hypophosphatemia result in rickets.

Mutations in PHEX result in increased synthesis of FGF23 which is responsible for phosphaturia and hypophosphatemia. FGF23 is a protein synthesized by osteoblast and osteocyte which inhibit phosphate reabsorption by renal tubule and if secreted in excess leads to hyperphosphaturia and subsequent hypophosphatemia.

Hypophasphatemic rickets is commonly misdiagnosed as nutritional rickets and usually treated by oral supplementation of vitamin D3, so biochemical investigation is necessary so as not to miss hypophosphatemia.

Thorough clinical and neurological examination of lower limb is mandatory in every case of rickets and in particular hypophasphatemic one.

There were reported cases of spinal cord compression due to calcification or ligamentum flavum hypertrophy. Our case developed thoracic myelopathy symptoms and signs without compression. Causes of Myelopathy with normal MRI (like B12 deficiency, folate deficiency, Copper and zinc derangement, ALS and cord infarction) thought, discussed and investigated by a neurologist but couldn't be determined. It is important to search for myelopathy due to cord compression in these cases because it is treatable surgically by decompression. Here the cause of myelopathy was noncompressive and complete work up for finding a cause

for myelopathy with normal MRI was nonconclusive. Literature review didn't show Familial HPR as a cause for myelopathy with normal MRI. This looks like to be the first case to be reported and a question rises whether it is a part of the illness or an association?

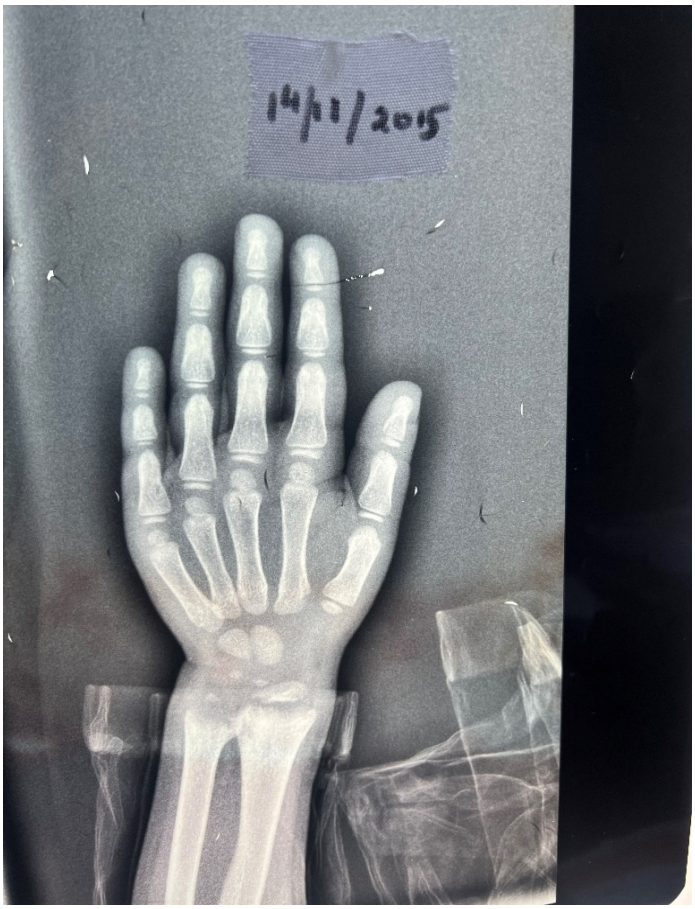

Figure1 Radiograph of wrist joint at the age of 6 years

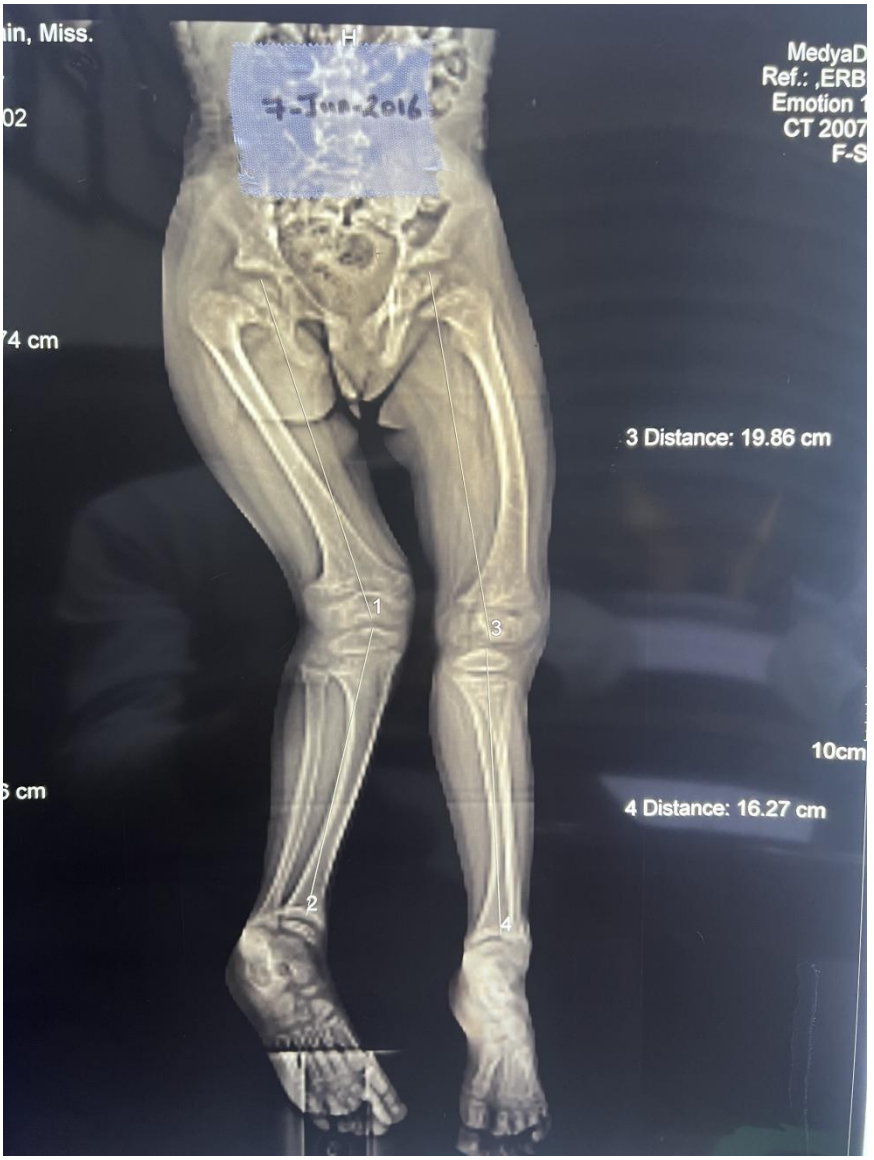

Figure 2 Radiograph of both lower limbs at the age of 7 years

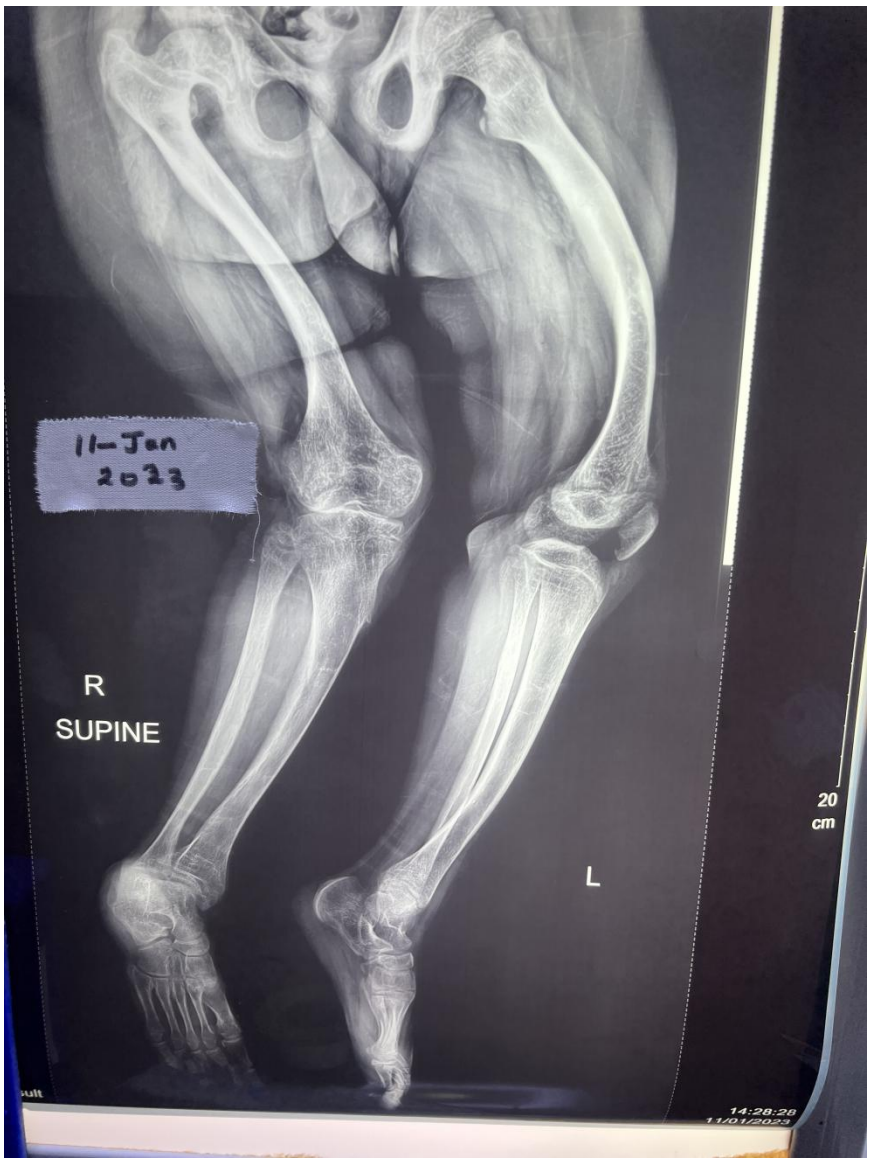

Figure 3 radiograph of both lower limbs showing coxa vara of right side and bowing of left femur at the age of 14 years

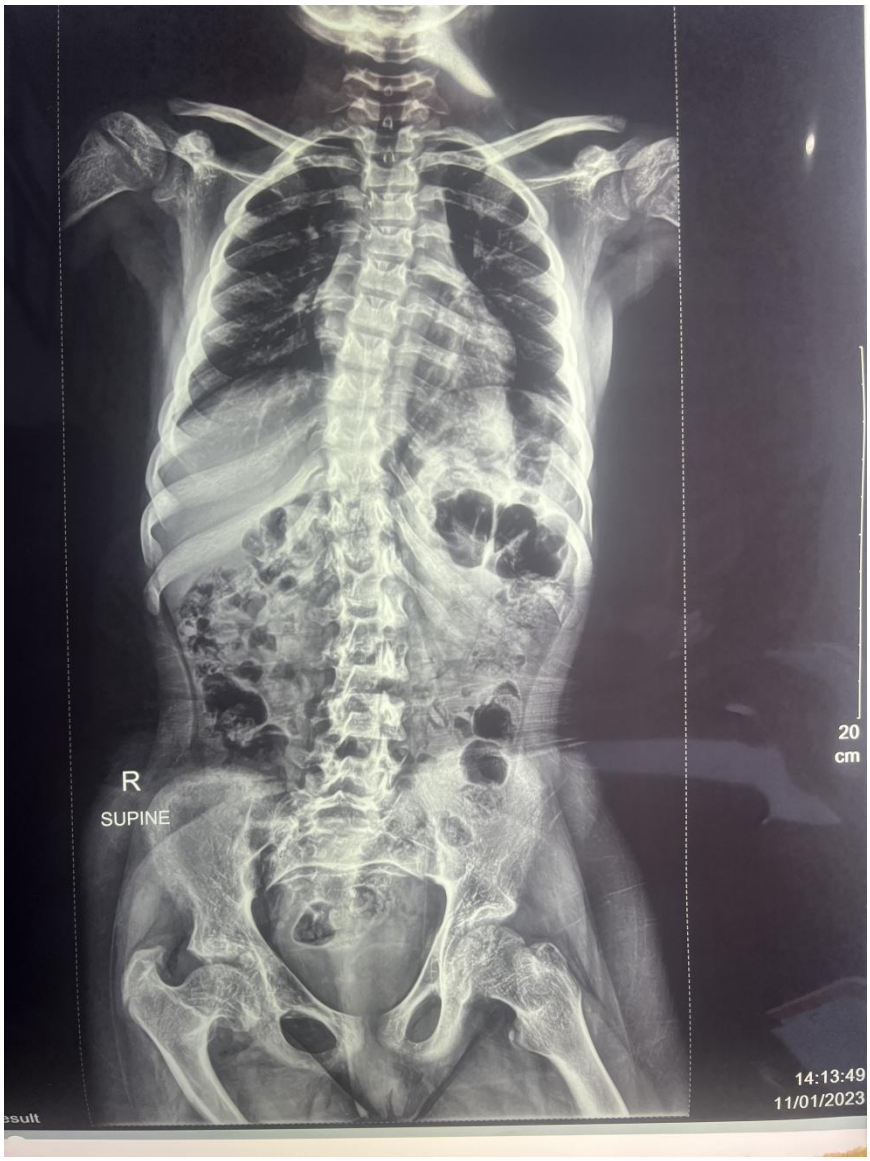

Figure 4 Early onset scoliosis

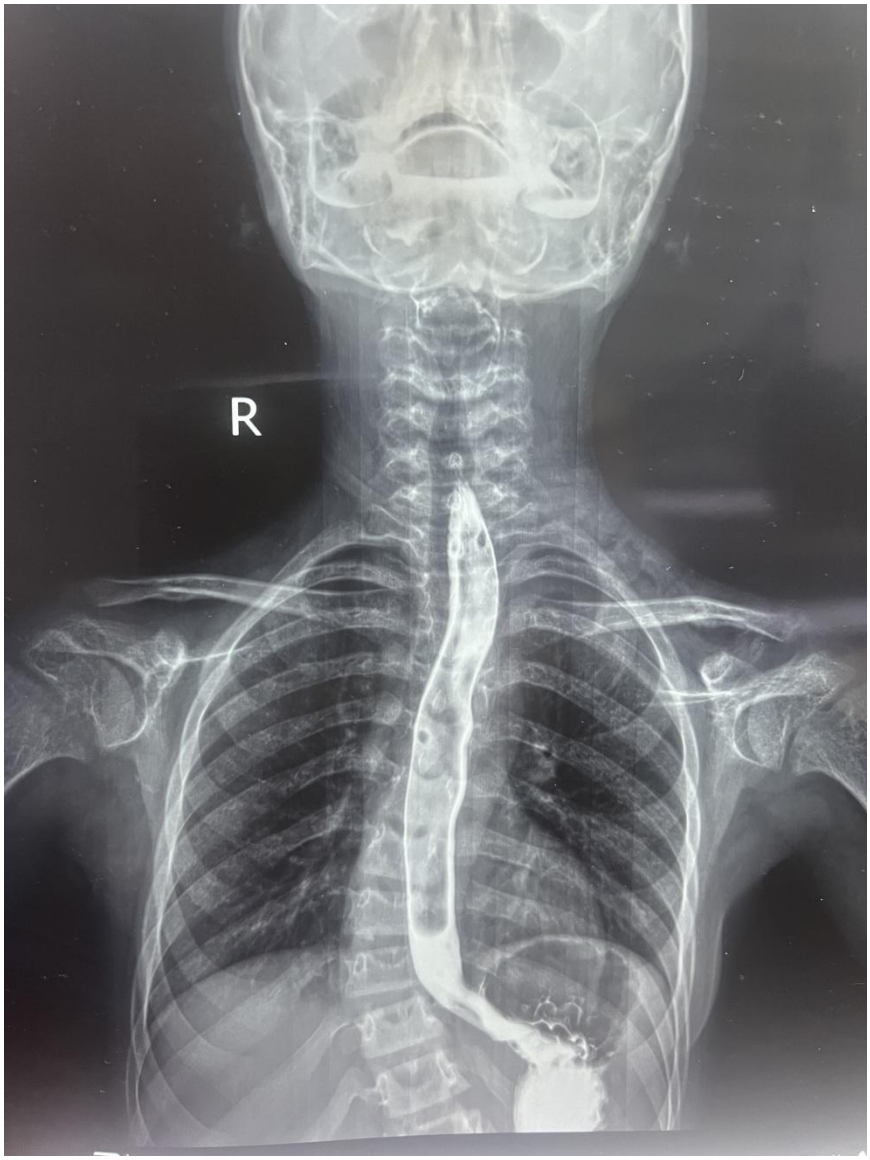

Figure 5 Barium study exclude stricture

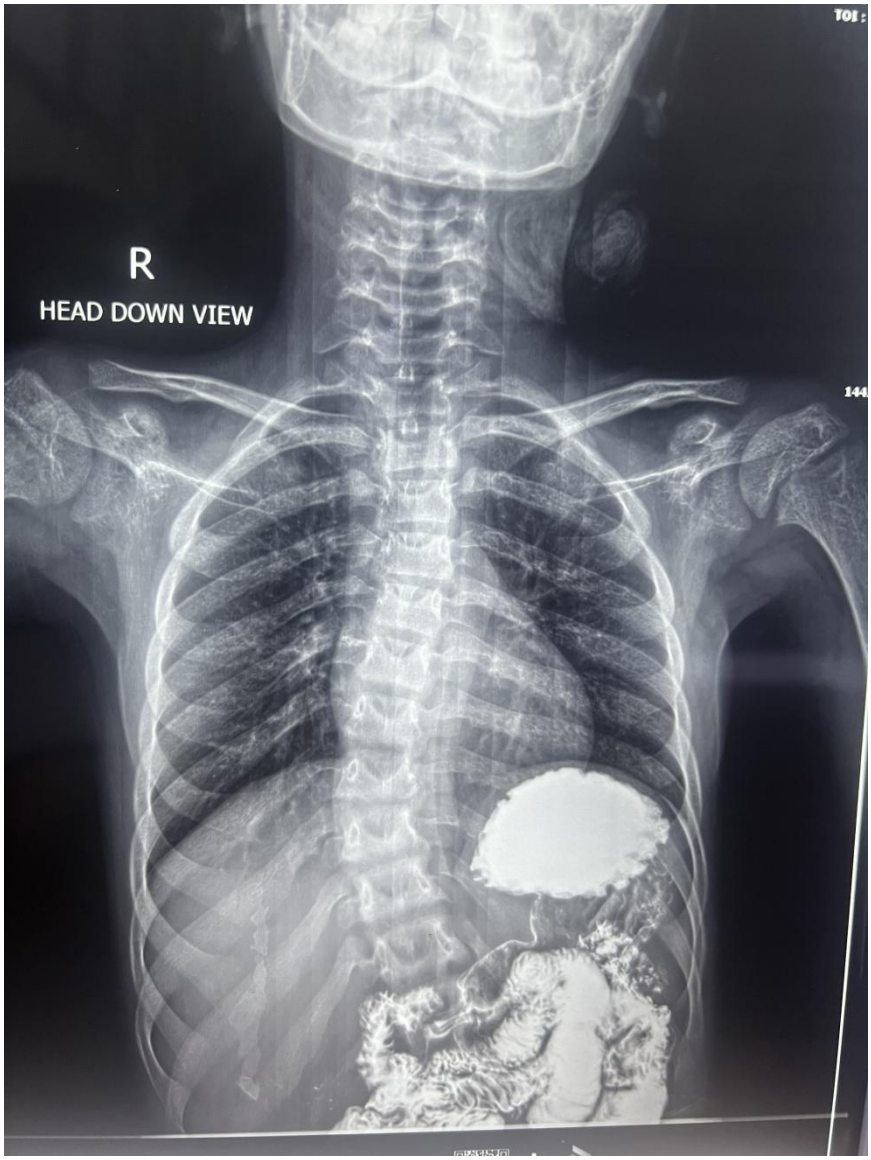

Figure 6 Barium swallow

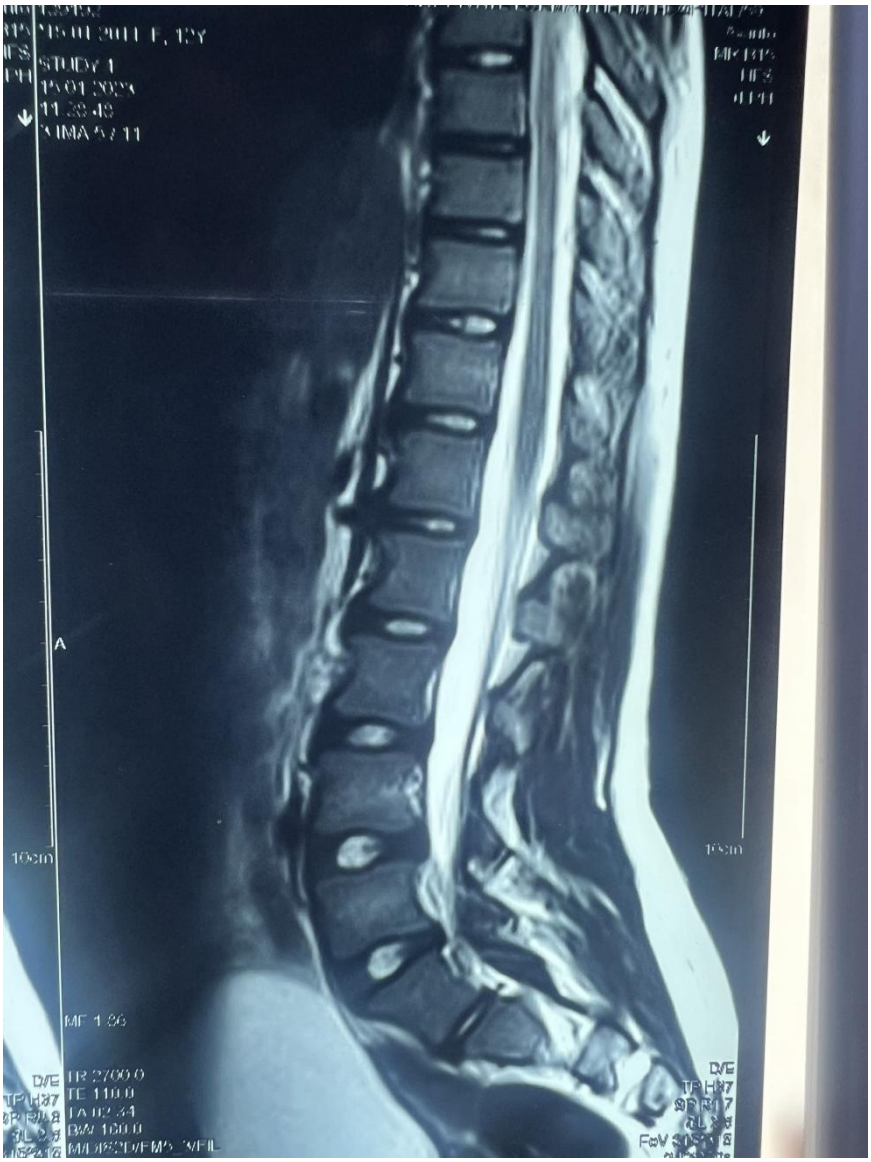

Figure 7 MRI of lumbosacral spine sagittal view

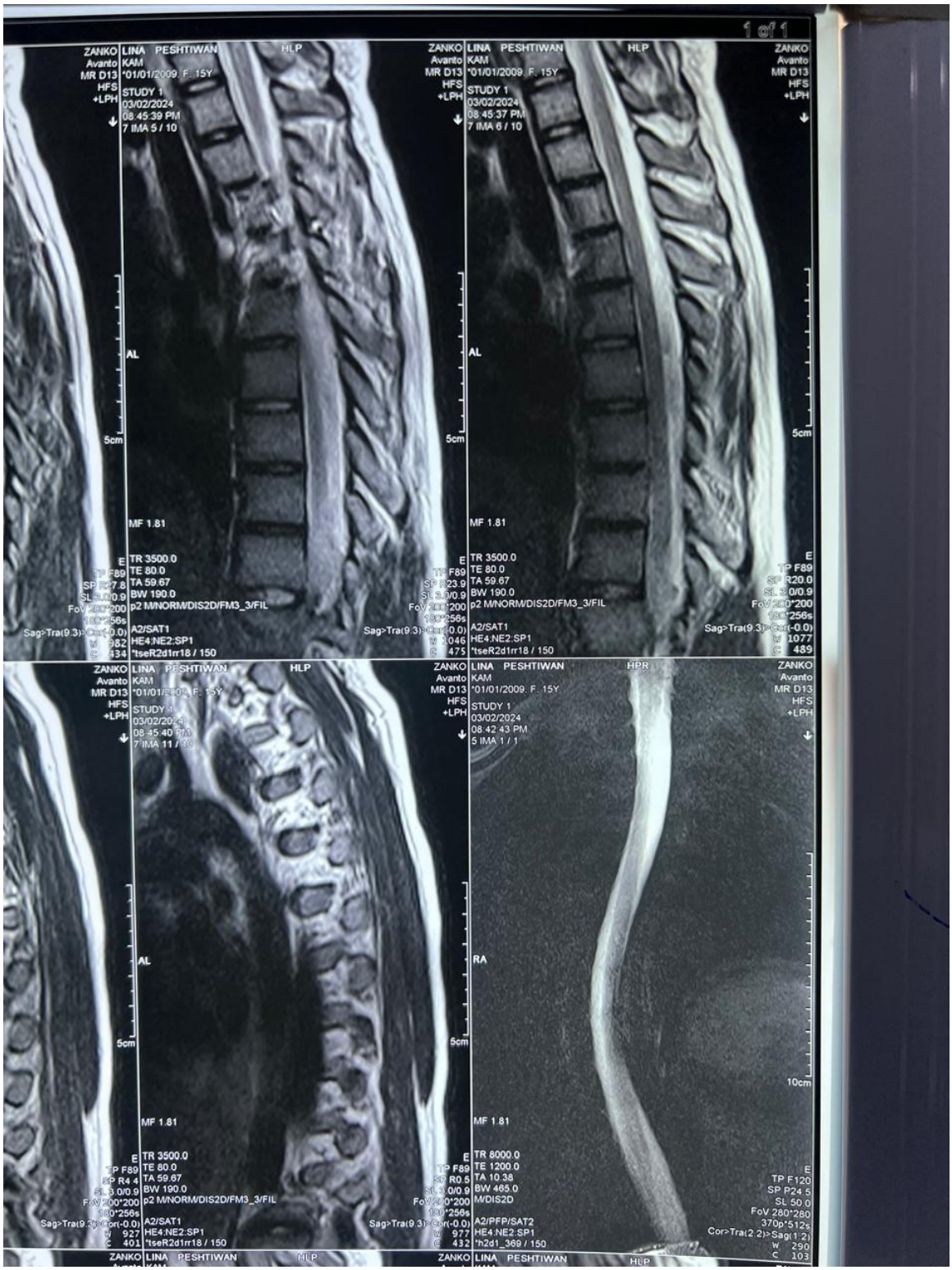

Figure 8 MRI of dorsal spine sagittal view

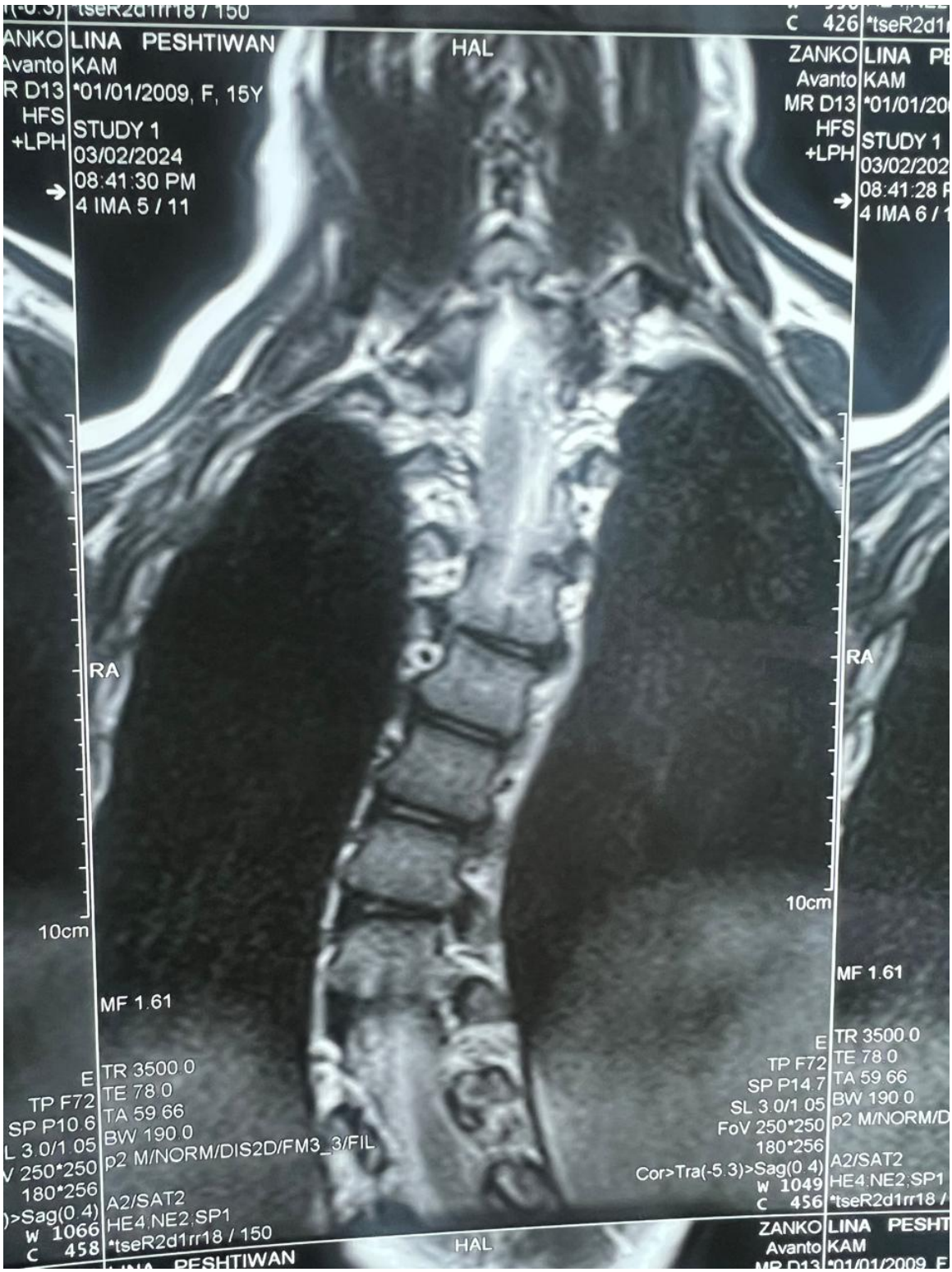

Figure 9 MRI dorsal spine coronal view

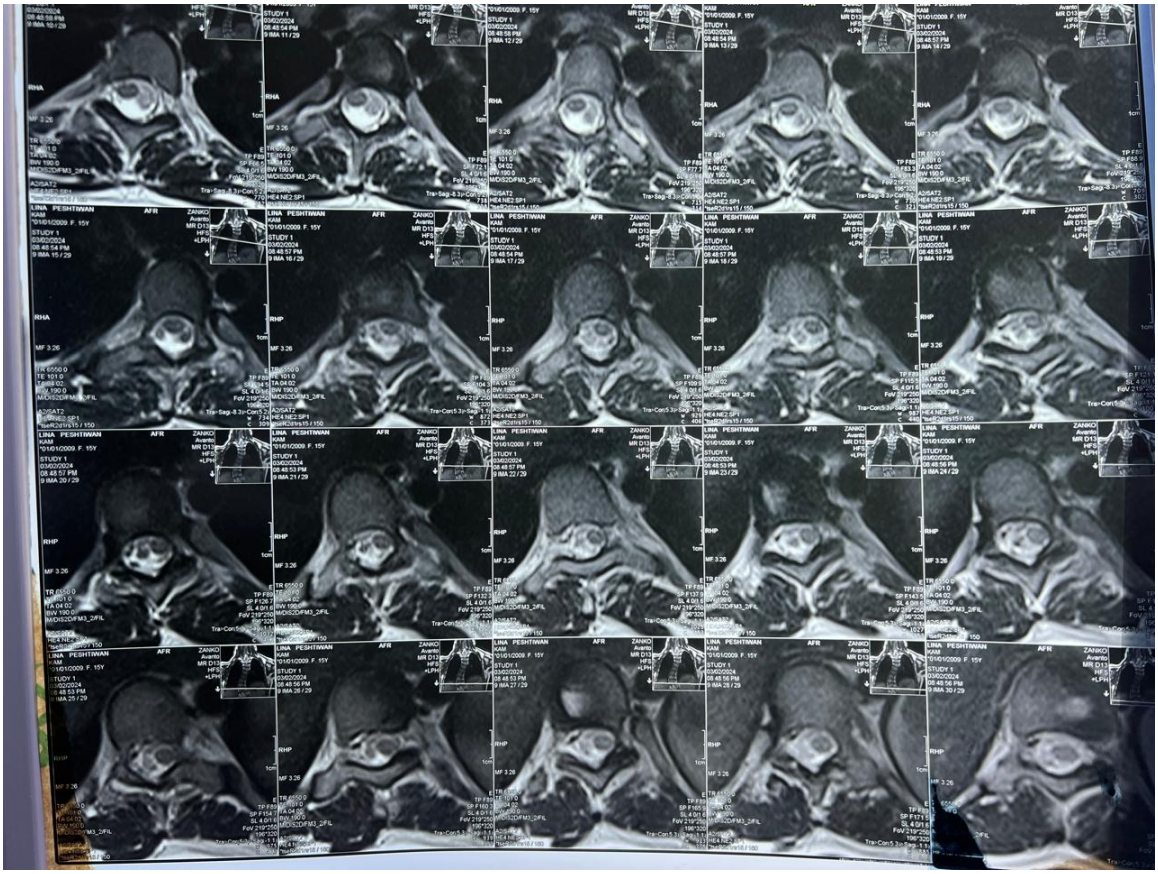

Figure 10 RI dorsal spine axial section

Conclusions

Do not forget to measure serum phosphorus in any child whose complaints suggest rickets. Identification of genetic defect is fundamental for characterization of hereditary hypophosphatemic rickets. Thorough neurological assessment of lower limbs and MRI of whole spine are essential to exclude myelopathy before decision for surgical intervention.

References

1. Arnaout, F. (2020) *Concise Orthopaedic Notes*. Warwickshire: Arnaout
2. Blom,A et al,. (2018) *Apley & Solomon's System of Orthopaedics and Trauma. 10th edn*. Bristol: CRC Press
3. Azar,F. and Beaty, J.(2021) *Campbell's Operative Ortjopaedics*. 14th edn. Philadelphia: Elsevier

4. Rafaelsen S, Johansson S, Raeder H, Bjerknes R. Hereditary hypophosphatemia in Norway: a retrospective population-based study of genotypes, phenotypes, and treatment complications. Eur J Endocrinol. 2016;174:125–136. doi: 10.1530/EJE-15-0515. [DOI] [PMC free article] [PubMed] [Google Scholar]
5. Makitie O, Doria A, Kooh SW, et al. Early treatment improves growth and biochemical and radiographic outcome in X-linked hypophosphatemic rickets. J Clin Endocrinol Metab. 2003;88(8):3591–3597. doi: 10.1210/jc.2003-030036. - DOI - PubMed

Medtronic