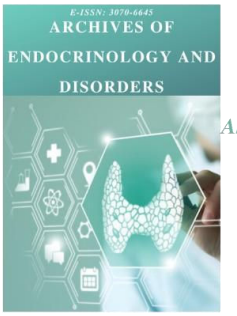

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Case Report

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The Enigma of FGF23 Dysregulation in Von Recklinghausen's Disease: A Case of Hypophosphatemic Osteomalacia with a Neurofibromin 1 Gene (c.6772C>T; p.Arg2258Ter) Mutation

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Abstract

Background: Neurofibromatosis type 1 (NF-1) or Von Recklinghausen's disease is an autosomal dominant disorder characterised by a variety of systemic manifestations including skeletal abnormalities. Hypophosphatemic Osteomalacia (HO) is a rare but significant complication associated with NF-1 primarily driven by Fibroblast Growth Factor 23 (FGF23)-mediated phosphate wasting.

Case Presentation: We report the case of a 54-year-old male with NF-1 presenting with progressive proximal muscle weakness and osteomalacia. Diagnostic workup revealed insufficiency fractures, severe hypophosphatemia and increased FGF23 levels, suggestive of FGF23-mediated HO. Additional findings included a peri-ampullary neuroendocrine tumor and a Gastrointestinal Stromal Tumor (GIST). Genetic testing identified a heterozygous pathogenic variant (c.6772C>T; p.Arg2258Ter) in the NF-1 gene. The patient was managed with phosphate and calcitriol supplementation, leading to clinical and radiological improvement.

Conclusion: Very few patients of Neurofibromatosis type 1 expressing HO is mysterious. NF-1-associated HO requires heightened clinical awareness for early diagnosis and intervention to alleviate the morbidity.

Keywords: Neurofibromatosis Type 1; Hypophosphatemic Osteomalacia; FGF23; Phosphate Wasting; Skeletal Manifestations; Genetic Mutation

Introduction

Neurofibromatosis type 1 (NF-1), also known as Von Recklinghausen's disease, is a common autosomal dominant genetic disorder with an estimated prevalence of 1 in 3,000 individuals worldwide. It is caused by mutations in the NF1 gene (350kb) located on chromosome 17, which encodes neurofibromin (2818 amino acids), a tumor suppressor protein involved in regulation of the RAS signaling pathway. Although NF-1 demonstrates nearly complete penetrance, its clinical manifestations are highly variable, ranging from cutaneous findings such as café-au-lait macules and neurofibromas to multisystem involvement. Skeletal abnormalities are well-recognized features of NF-1 and may include long bone dysplasia, congenital bowing, scoliosis and sphenoid wing dysplasia.

In addition to structural skeletal defects, metabolic bone disorders such as osteoporosis and rarely, Hypophosphatemic Osteomalacia (HO) have been described in adults with NF-1. Hypophosphatemic osteomalacia is characterized by renal phosphate wasting, impaired bone mineralization and resultant bone pain, muscle weakness and fractures. Fibroblast growth factor 23 (FGF23), a phosphaturic hormone produced primarily by osteocytes, plays a central role in the pathogenesis by reducing renal tubular phosphate reabsorption and suppressing 1,25-dihydroxyvitamin D synthesis.

The association between NF-1 and FGF23-mediated hypophosphatemic osteomalacia is rare, with fewer than 50 cases reported in the literature to date. The exact mechanism remains incompletely understood, although emerging evidence suggests that NF1-deficient osteocytes may contribute to increased FGF23 production via dysregulated RAS/PI3K signaling pathways. Historically, ectopic production of phosphaturic factors by neurofibromas or associated tumors was proposed; however, recent studies have challenged this hypothesis.

Given the rarity and diagnostic complexity of this condition, delayed recognition is common, leading to significant morbidity due to fractures and progressive disability. Early identification of FGF23-mediated phosphate wasting is crucial, as appropriate treatment with phosphate supplementation and active vitamin D analogues can result in significant clinical improvement.

In this report, we present a case of NF-1 associated with FGF23-mediated hypophosphatemic osteomalacia in a middle-aged male, highlighting the diagnostic challenges, underlying pathophysiology and therapeutic considerations in this uncommon but clinically significant association.

Case Report

A 54-year-old male presented with a two-year history of gradually progressive groin pain and low backache, accompanied by increasing difficulty in climbing stairs. He was initially evaluated by an orthopedician and after fractures were ruled out, was treated with calcium and vitamin D supplementation. Despite this, his symptoms progressively worsened, leading to significant weakness and eventual inability to ambulate without support. He was subsequently referred to a neurophysician and MRI of the spine revealed features suggestive of diffuse osteomalacia involving the lumbar spine. During this period, the patient also developed urinary retention requiring catheterization. Further evaluation with ultrasonography of the abdomen and pelvis revealed a urinary bladder diverticulum along with an incidental lesion in the head of the pancreas.

Patient was referred for further evaluation of osteomalacia to our Endocrinology Department. On further enquiry there was no history of renal calculi, pain abdomen or fractures. No history of decreased urine output or dental problems. No history of diabetes, hypertension or tuberculosis in the past. There was history of recurrent left lower limb swelling associated with fever since 20 years. Takes vegetarian diet and has decreased appetite, with inadequate intake of foods rich in calcium and vitamin D. Not habituated to smoking or drinking alcohol. No history of similar complaints in the family. He is married and has 2 children.

On examination weight was 50 kg, height of 163 cms and BMI-19 kg/m^2. PR- 98/min and BP- 140/90 mmHg. Pallor and non-pitting edema of left lower limb extending upto knee were present. On head to toe examination there were multiple (>6) hyperpigmented macules of >15 mm on the trunk suggestive of café-au-lait macules and multiple soft, fleshy, sessile swellings all over the body suggestive of cutaneous neurofibromas. Hyperpigmented macules were noted on palms (palmar freckling) and there were no gross skeletal deformities. On nervous system examination: higher mental functions and cranial nerves were normal. Tone was normal. Power was 4/5 at both shoulders, 4+/5 at both elbows, 3/5 at both hips, 4+/5 at knees and ankles. Deep tendon reflexes were normal. Bilateral plantar response was normal. Sensory system was normal. Cardiovascular and respiratory system examination were normal. Per abdomen was soft, no mass, organomegaly or free fluid. Urinary bladder was catheterized in view of urinary retention (Fig. 1-4).

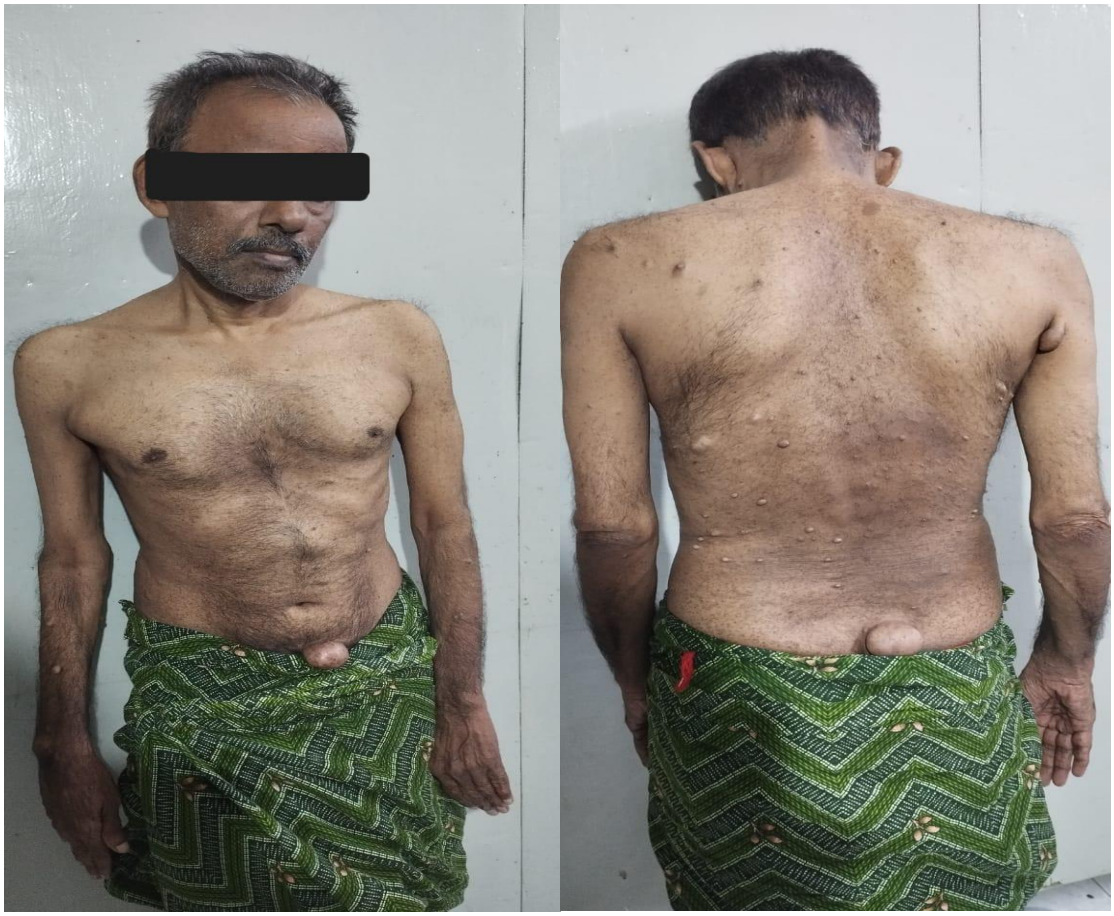

Figure 1: Multiple (>6) hyperpigmented macules of >15 mm on the trunk suggestive of café-au-lait macules and multiple soft, fleshy, sessile swellings all over the body suggestive of cutaneous neurofibromas.

Figure 2: Palmar freckling.

Figure 3: Left lower limb- non-pitting edema.

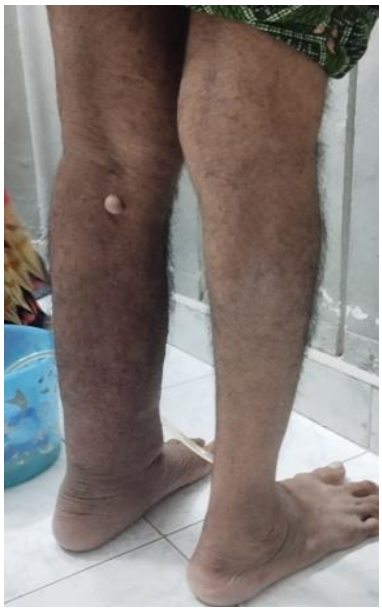

Figure 4: Neurofibroma on posterior aspect of left leg.

At the end of history and examination the diagnosis of neurofibromatosis with proximal muscle weakness, with investigations showing osteomalacia, urinary bladder diverticulum and mass at head of pancreas was made. On further enquiry son had similar lesions over the back, suggestive of café-au-lait macules and cutaneous neurofibromas (Fig. 5, Table 1).

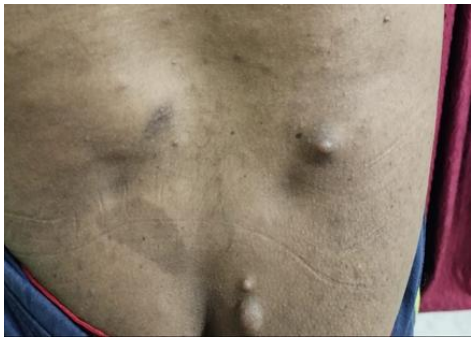

Figure 5: Son later realised that he also had café-au-lait macules and cutaneous neurofibromas on hic back.

Parameter	Value	Normal range
Hb	10.4 gm/dl	12-15 gm/dl
Serum creatinine	0.72 mg/dl	0.74-1.35 mg/dl
Serum calcium	8.5 mg/dl	8.5-10.5 mg/dl
Serum albumin	3.8 mg/dl	3.5-5.5mg/dl
Serum phosphorous	1.45 mg/dl	2.5-4.5 mg/dl
Serum alkaline phosphatase	652.2 IU/L	44- 147 IU/ml
25-OH Vitamin D	53.55 ng/ml	30-100 ng/ml
iPTH	203.2 pg/ml	15-65 pg/ml
Serum sodium	135 mEq/l	135-145 mEq/L
Serum potassium	3.98 mEq/l	3.5-5.5 mEq/L
PSA	<2 ng/ml	1-1.5 ng/ml (>2.5-abnormal)

Table 1: Initial investigations.

MRI Pelvis: bone marrow edema and well defined STIR hyperintense fracture line across medial cortex of proximal left femoral shaft and bilateral pubic rami, suggestive of severe diffuse osteomalacia with insufficiency fractures (Fig. 6).

MRI lumbar spine: features suggestive of severe osteomalacia DEXA scan (Table 2).

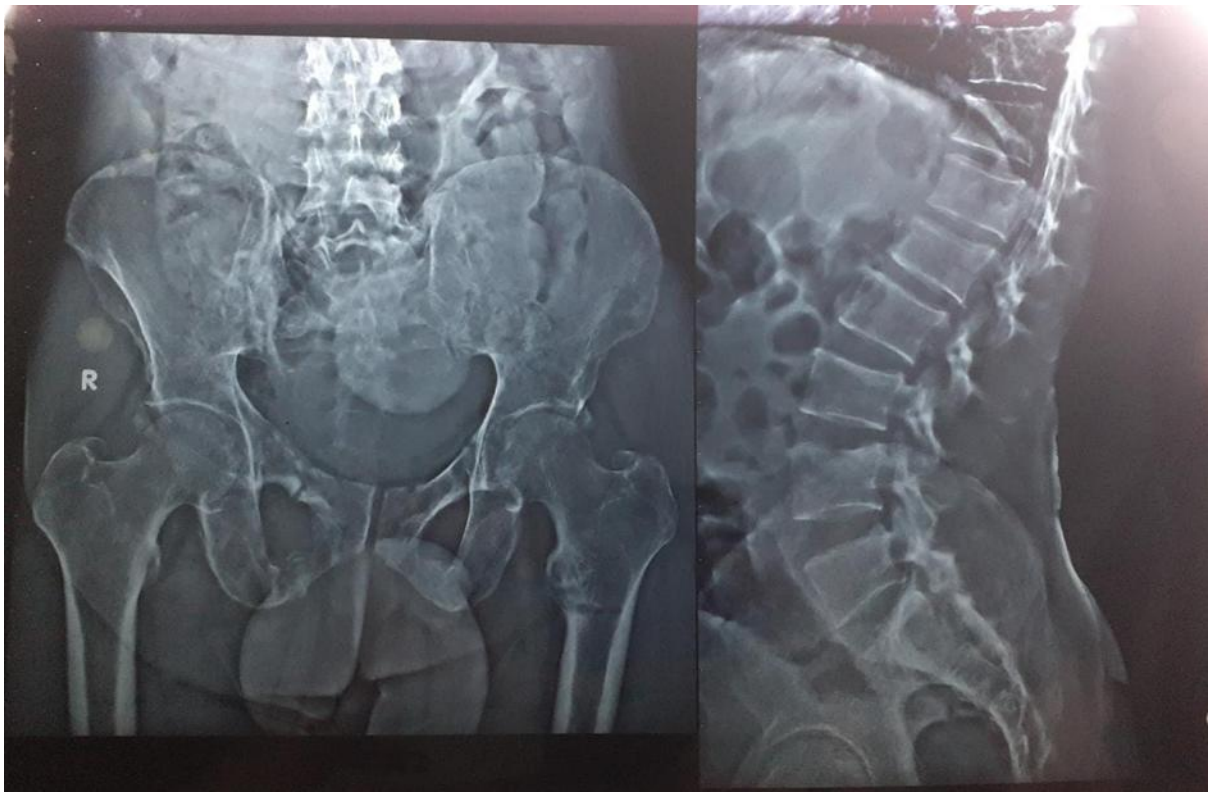

Figure 6: X-ray: pelvis-AP view (1/10/2022): showing looser zones or pseudofractures in right super pubic ramus and left sub-tronchanteric regions (pointed by yellow arrows and spine-Lateral view (1/10/2022): severe osteomalacia of vertebral bodies.

Site	BMD (g/cm²)	T Score	Z Score
L1-L4	0.926	-2.5	-1.3
Left neck of femur	0.627	-3.4	-2
Right neck of femur	0.648	-3.2	-1.9

Table 2: DEXA scan parameters at diagnosis.

Tc99 MDP Bone scan showed increased tracer uptake in thoracic spine, pelvis, left proximal femoral shaft and right ankle suggestive of metabolic superscan. As USG abdomen showed urinary bladder diverticulum of size 5.5 x 3.4 cm and focal isoechoic lesion in the periampullary region of pancreas measuring 4.4 x 4.3 cm, PET CT scan was done and showed metabolically active lesion in peri-ampullary region measuring approximately 1.9 x 1.5 x 1.5 cms (Fig. 7).

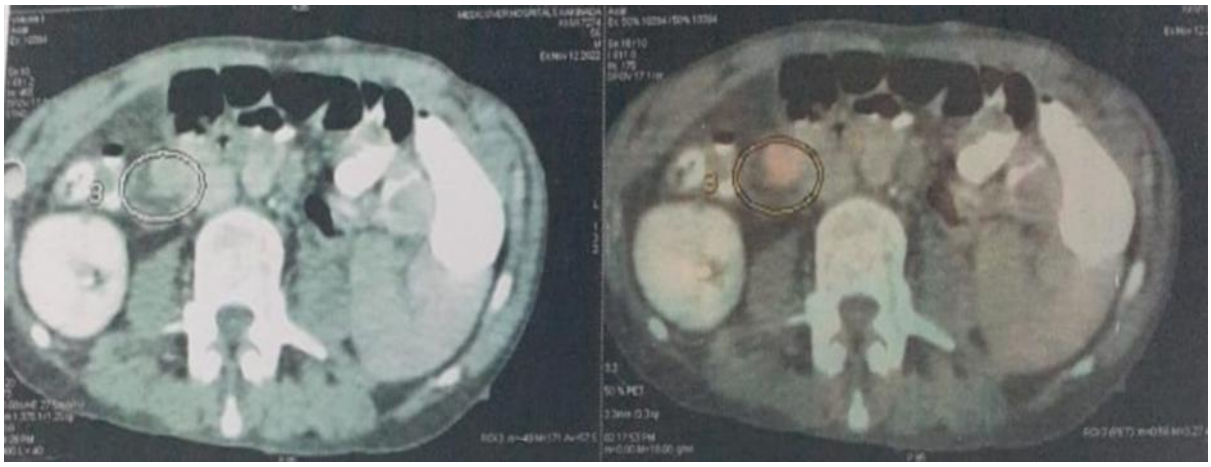

Figure 7: PET CT scan was done and showed metabolically active lesion in peri-ampullary region measuring approximately 1.9 x 1.5 x 1.5 cms.

A gastroenterology consultation was obtained. Upper Gastrointestinal (UGI) endoscopy revealed an ampullary growth, from which a biopsy was taken. Additionally, a submucosal lesion was identified in the gastric fundus, probably a Gastrointestinal Stromal Tumor (GIST). Histopathological examination of the ampullary lesion confirmed a well-differentiated neuroendocrine tumor, with a mitotic rate of <2 per 2 mm² (equivalent to 10 high-power fields at 40x magnification).

Inference: Based on the clinical presentation, imaging and histopathological findings, a provisional diagnosis of hypophosphatemic osteomalacia with pseudofractures and proximal muscle weakness was made in a patient with Neurofibromatosis type 1, associated with a periampullary neuroendocrine tumor and a probable gastrointestinal stromal tumor. In view of low serum phosphorous and osteomalacia, TmP GFR was calculated (Fig. 8, Table 3).

- Urine creatinine (Uc)- 17.92 mg/dl
- Urine phosphorous (Up)- 11.54 mg/dl
- Serum creatinine (Sc)- 0.93 mg/dl
- Serum phosphorous (Sp) - 1.54 mg/dl
- Clearance of phosphorous/Clearance of creatinine = Up x Sc / Sp x Uc =0.38
- FEPO4 (fractional excretion of phosphorous) =38%
- TRP= 0.62
- TmP/GFR= 0.4mmol/L (0.9-1.35) was derived from nomogram

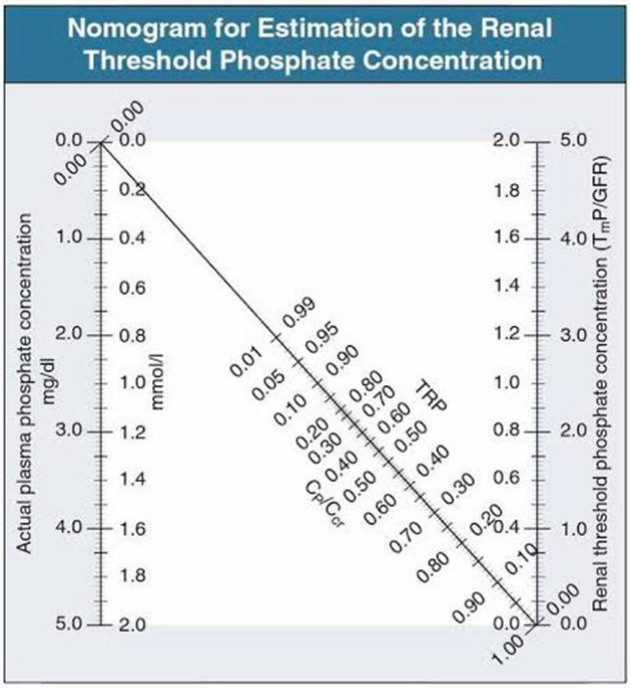

Figure 8: Nomogram for TmP-GFR.

Adults

Age	Male Range (mmol/L)	n	Female range (mmol/L)	n
25-35 years	1.00-1.35	15	0.96-1.44	15
45-55 years	0.90-1.35	15	0.88-1.42*	15
65-75 years	0.80-1.35	15	0.80-1.35	15

*Premenopausal

Table 3: Reference range of TmP-GFR as per age and sex (as per NHS).

FGF23 was 129.9 pg/ml (Normal Range: 23-95.4 pg/ml), suggestive of FGF23 mediated hypophosphatemic osteomalacia. So our final diagnosis was FGF23 mediated hypophosphatemic osteomalacia in a case of Neurofibromatosis-1 with peri-ampullary neuro-endocrine tumor and GIST. Patient was started on phosphorous supplementation at 20 mg/kg body weight in 4 divided doses (i.e; 1 gm/day of elemental phosphorous, in the form of sodium acid phosphate sachet) along with calcitriol at 1 mcg/day. Calcium carbonate (1250 mg) equivalent to elemental calcium 500 mg BD was also given. Regular monitoring of serum calcium, phosphorous and urinary calcium excretion were done. Over a span of 6 months, patient showed both clinical and radiological improvement (Fig. 9, Table 4).

Parameter	Value	Range
Serum Calcium	9.1 mg/dl	8.8-10.6 mg/dl
Serum phosphorous	2.1 mg/dl	2.5-4.5 mg/dl
ALP	195.61 IU/L	<115 IU/l
PTH	96 pg/ml	12-88 pg/ml

Table 4: Laboratory investigations 6 months after start of treatment.

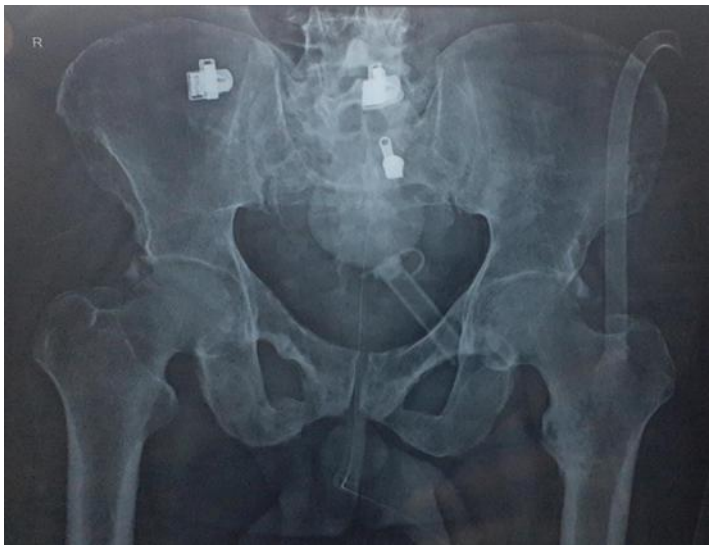

Figure 9: Fading of looser zones post treatment with phosphorous supplementation.

Ga 68 DOTATATE PET-CT was done for peri-ampullary Neuroendocrine Tumor (NET) which showed mild somatostatin receptor expressive lesion in periampullary region, with no significant tracer activity in ill-defined lytic-sclerotic lesions in pelvic bones, sacrum and proximal end of left femur.

Referred to Department of Gastroenterology for further management of neuroendocrine tumor. Endoscopic ultrasound and endoscopic excision of periampullary neuroendocrine tumor was done. Patient continued to require phosphorous supplementation even after excision of periampullary NET, so it might not be the source of FGF-23.

Whole exome sequence was sent for phenotype-genotype correlation which showed heterozygous pathological variant mutation at 6772 nucleotide (C>T, leading to p.Arg2258Ter) of exon 45 of NF-1 gene, leading to premature termination of protein coding and truncated neurofibromin production. This was rare with only 20 cases reported till date in ClinVar database (Fig. 10) [1].

Result:

Positive: An established or plausible cause of the reported phenotype was identified

Variant(s) Information

Gene (Transcript)	Location	Variation	Zygosity	Classification*	Disease (OMIM)	Inheritance
NF1 (NM_001042492.3)	Exon 45	c.6772C>T (p.Arg2258Ter)	Heterozygous	Pathogenic	Neurofibromatosis-Noonan syndrome (601321); Neurofibromatosis, familial spinal (162210); Watson syndrome (193520); Neurofibromatosis, type 1 (162200)	Autosomal dominant
					Leukemia, juvenile myelomonocytic (607785)	Somatic mutation, Autosomal dominant

Database Information

1000 Genomes	EVS*	ExAC#	gnomAD¥	Indian Exome Database^	In-house exome database†	dbSNP	OMIM	ClinVar
Absent	Absent	Absent	Absent	Absent	Absent	Present	Absent	Present
MAF: NA	MAF: NA	MAF: NA	MAF: NA	MAF: NA	MAF: NA	ID: rs876658541	ID: NA	ID: VCV000230389.40

*EVS- Exome Variant Server, #ExAC- Exome Aggregation Consortium, ¥Genome Aggregation Database, ^836 Indian exome samples, †120 Indian exome samples, MAF- Minor Allele Frequency, NA- Not Available

Figure 10: Result of whole exome sequencing.

Discussion

Metabolic bone disorders encompass a spectrum of conditions characterized by abnormalities in bone mineralization, turnover and strength, leading to increased fracture risk and skeletal deformities. Osteomalacia, specifically, results from defective mineralization of osteoid, most commonly due to vitamin D deficiency, chronic kidney disease or phosphate depletion.

Hypophosphatemic osteomalacia represents a distinct subset driven by renal phosphate wasting, frequently mediated by phosphaturic hormones such as Fibroblast Growth Factor 23 (FGF23) [1-9]. FGF23 reduces proximal tubular phosphate reabsorption and suppresses 1α-hydroxylase activity, leading to decreased calcitriol synthesis and impaired bone mineralization [10-13]. In contrast to osteoporosis, where bone matrix is reduced, osteomalacia is characterized by accumulation of unmineralized osteoid and pseudofractures (Looser's zones). In the context of genetic disorders such as Neurofibromatosis type 1 (NF-1), both structural and metabolic bone abnormalities may coexist, highlighting the complex interplay between genetic signaling pathways and bone metabolism [10,12,14,15]. Hypophosphatemic osteomalacia is amongst the rare disorders associated with NF1, with less than 50 cases reported till date. The first two cases were reported by Gould, in which post-mortem of neurofibromatosis cases showed undecalcified bone with increased osteioid [2]. In 1991, Konishi.et.al reported a case of NF-1 with HO and documented low TmPhos/GFR determined by Walton and Bijvoet nomogram (discovered in 1975), along with review of 34 cases of NF-1 with osteomalacia [3]. Initially only hypophosphatemia was documented followed by TRP (Tubular Reabsorption of Phosphate) since 1967 and later TmP/GFR was determined since 1975. Neurofibromata or an occult secondary tumor produces humoral factor (as NF-1 patients are at increased risk of secondary tumors) was the thought until the 20th century. In 2000 FGF23 was identified by positional cloning as the gene responsible for autosomal dominant hypophosphatemic rickets. In 2003 a case report by M Sovied, et al., showed premanent cure of tumor associated osteomalacia after surgical removal of two large schwannomas in a case of neurofimatosis-1 [4]. In 2015 Gupta, et al., stated that probably the largest neurofibroma or those with recent growth cause HO and their surgical removal should be tried, to achieve permanent cure (Fig. 11) [5].

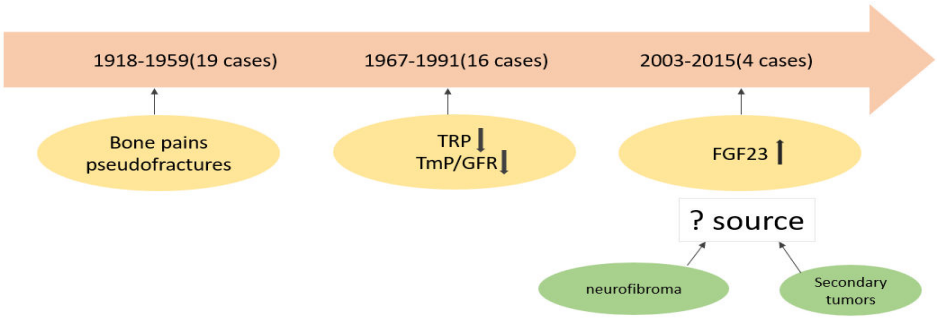

Figure 11: Flow diagram of review of NF-1 cases with osteomalacia over years.

A study by Kamiya, et al., involving Nf1 cKO (conditional Knockout) mouse with Nf1 deficient osteocytes-showed that PI3K (phosphoinositide (PI) 3-kinase) pathway activation might be responsible for increased FGF23 production [6]. As the upregulation of FGF23 was inhibited by PI3K inhibitor Ly294002, the sequence of which is shown in Fig. 12.

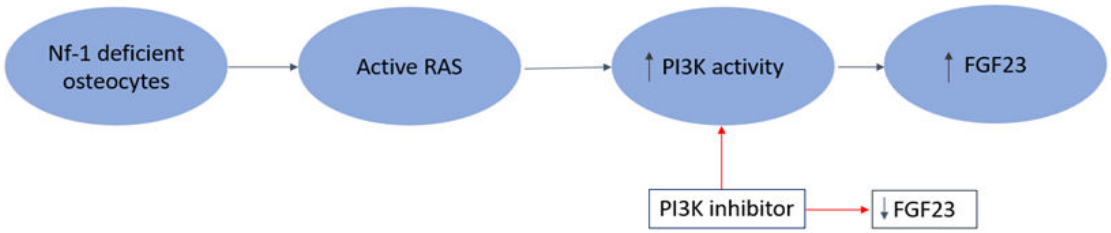

Figure 12: Upregulation of FGF23 production due to Active Ras in NF-1 deficient osteocytes.

According to the study done by Sahoo, et al., in 2019, tumour cells in the neurofibroma tissues did not stain FGF23 on IHC. So, it is unlikely for neurofibromas to contribute to high circulating FGF23 levels in NF1 [7]. In 2022 Angelos Kaspiris, et al., described that if NF-1 protein is absent in osteocytes, RAS remains in its active state [8]. Activation of RAS and its downstream signaling pathways PI3K/AKT/mTOR might be responsible for FGF23 production. Increased secretion of FGF23 from Nf-1 deficient osteocytes results in abnormal calcium-phosphorus metabolism, bone mineralization and an osteomalacia-like bone phenotype (Fig. 13).

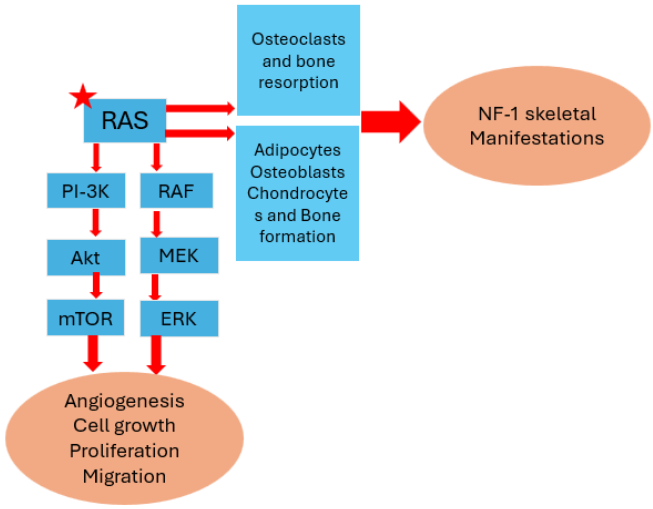

Figure 13: RAS activation leading to skeletal manifestations.

In our case, NF1-deficient osteocytes are the most plausible source of excess FGF23, rather than the coexisting neuroendocrine tumor or GIST, as these tumors are not typically implicated in tumor-induced osteomalacia. This observation is supported by prior studies. Kamiya, et al., demonstrated in an NF1 conditional knockout mouse model that osteocyte-specific NF1 deficiency leads to increased FGF23 production via activation of the PI3K pathway [6]. Similarly, Sahoo, et al., reported lack of FGF23 expression in neurofibroma tissue on immunohistochemistry, arguing against tumors as the primary source of phosphaturic factors in NF-1 [7]. Furthermore, Kaspiris, et al., highlighted that loss of neurofibromin results in persistent RAS activation and downstream PI3K/AKT/mTOR signaling, contributing to abnormal bone metabolism and elevated FGF23 levels [8]. Together, these studies support the concept that intrinsic osteocyte dysfunction, rather than tumor secretion, underlies FGF23 excess in NF-1. Our case further reinforces this hypothesis, as phosphate wasting persisted despite resection of the neuroendocrine tumor, suggesting a non-tumoral source of FGF23.

Conclusion

Neurofibromatosis type 1–associated hypophosphatemic osteomalacia is a rare but clinically significant entity that requires a high index of suspicion for timely diagnosis. Emerging evidence suggests that NF1-deficient osteocytes play a central role in FGF23 overproduction, leading to renal phosphate wasting and impaired bone mineralization. This case highlights the importance of recognizing metabolic bone disease in NF-1, even in the presence of coexisting tumors that may confound the diagnosis. Early identification and appropriate treatment can significantly improve clinical outcomes. The potential role of targeted therapies such as burosumab warrants further exploration. Additionally, the relationship between specific NF1 mutations, including p.Arg2258Ter and skeletal manifestations remains unclear, emphasizing the need for larger studies to establish genotype–phenotype correlations and optimize management strategies.

Conflict of Interest

The authors declared no potential conflicts of interest with respect to the research, authorship and/or publication of this article.

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None.

Data Availability Statement

The datasets used and/or analysed during the current study are available from the corresponding author upon reasonable request.

Ethical Statement

The project did not meet the definition of human subject research under the purview of the IRB according to federal regulations and therefore was exempt.

Informed Consent Statement

Informed consent was taken for this study.

Authors' Contributions

All authors contributed equally to this paper.

References

1. ClinVar Miner. Submissions by variant NM_001042492.3(NF1):c.6772C>T (p.Arg2258Ter). [Last accessed on: March 25, 2026] https://clinvarminer.genetics.utah.edu/submissions-by-variant/NM_001042492.3%28NF1%29%3Ac.6772C%3ET%20%28p.Arg2258Ter%29
2. Gould SE. Neurofibromatosis with osteomalacia. J Pathol Bacteriol. 1918;23(1):163-8.
3. Konishi K, Nakamura M, Yamakawa H, Suzuki H, Saruta T, Hanaoka H, et al. Hypophosphatemic osteomalacia in von Recklinghausen neurofibromatosis. Am J Med Sci. 1991;301(5):322-8.
4. Soveid M. Tumor associated osteomalacia in neurofibromatosis: Case report and literature review. Med J Islam Repub Iran. 2003;16(4):217-20.
5. Gupta A, Dwivedi A, Patel P, Gupta S. Hypophosphatemic osteomalacia in von Recklinghausen neurofibromatosis: Case report and literature review. Indian J Radiol Imaging. 2015;25(1):63-6.
6. Kamiya N, Yamaguchi R, Aruwajoye O, Kim AJ, Kuroyanagi G, Phipps M, et al. Targeted disruption of NF1 in osteocytes increases FGF23 and osteoid with osteomalacia-like bone phenotype. J Bone Miner Res. 2017;32(8):1716-26.
7. Sahoo SK, Kushwaha P, Bharti N, Khedgikar V, Trivedi R, Agrawal V, et al. Elevated FGF23 in a patient with hypophosphatemic osteomalacia associated with neurofibromatosis type 1. Bone. 2019;129:115055.
8. Kaspiris A, Savvidou OD, Vasiliadis ES, Hadjimichael AC, Melissaridou D, Iliopoulou-Kosmadaki S, et al. Current aspects on the pathophysiology of bone metabolic defects during progression of scoliosis in neurofibromatosis type 1. J Clin Med. 2022;11(2):444.
9. Pickering ME, Bouvier D, Puravet A, Soubrier M, Sapin V, Oris C. Hypophosphatemia related to a neuroendocrine tumor of the pancreas: A case report. Clin Biochem. 2022;104:62-5.
10. Carpenter TO. The expanding family of hypophosphatemic syndromes. J Bone Miner Metab. 2012;30(1):1-9.
11. Florenzano P, Hartley IR, Jimenez M. Tumor-induced osteomalacia. Calcif Tissue Int. 2021;108(1):128-42.
12. Minisola S, Peacock M, Fukumoto S. Tumour-induced osteomalacia. Nat Rev Dis Primers. 2017;3:17044.
13. Imel EA, DiMeglio LA, Hui SL, Carpenter TO, Econs MJ. Treatment of X-linked hypophosphatemia with calcitriol and phosphate increases circulating FGF23 concentrations. J Clin Endocrinol Metab. 2010;95(4):1846-50.
14. Razzaque MS. The FGF23–Klotho axis: Endocrine regulation of phosphate homeostasis. Nat Rev Endocrinol. 2009;5(11):611-9.
15. Feng JQ, Ward LM, Liu S. Loss of DMP1 causes rickets and osteomalacia and identifies a role for osteocytes in mineral metabolism. Nat Genet. 2006;38(11):1310-5.