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Case Series : FAHR from Truth

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We present to you the rare case of familial hypoparathyroidism in a sibling (Brother and Sister) residing in the district of Panna Madhya Pradesh , who presented with very different set of clinical signs and symptoms on different dates with the same final diagnosis of the same disease.

Case 1:

The patient, Mr. Ramnarayan, presents with chronic neuro-metabolic symptoms.

He reported recurrent generalized tonic-clonic seizures every 2 minutes and was put on four antiepileptic drugs at a medical college in Jabalpur but the epilepsy episodes were uncontrolled and had progressive gait instability.The episodes were preceded by diffuse numbness and muscle cramps. Over the last six months, neuromuscular hyperexcitability became increasingly prominent. Bedside assessment demonstrated a profoundly positive Trousseau's sign. Evaluation also revealed notable signs of cerebellar ataxia during gait testing. Examination was notable for skin thickening and tightening over the extremities. Broad laboratory evaluation was initiated to evaluate the etiology of his seizures. Results confirmed a state of profound hypocalcemia alongside hyperphosphatemia. Serum parathyroid hormone (PTH) was confirmed to be inappropriately suppressed. Normal magnesium levels successfully ruled out hypomagnesemia-induced gland suppression. Nutritional vitamin D was within normal limits, ruling out severe osteomalacia. The biochemical markers firmly established a diagnosis of idiopathic hypoparathyroidism. A brain CT was ordered to determine the structural cause of the recurrent seizures. Brain imaging confirmed classic, symmetrical calcifications of the basal ganglia and cerebellum. This distinct intracranial calcification correlates directly with long-standing metabolic hypocalcemia. Renal function panels and abdominal ultrasounds showed no signs of nephrocalcinosis. Serological testing identified positive antinuclear antibodies with a centromere pattern. These specific immunological features confirmed a concurrent diagnosis of systemic sclerosis. An electrocardiogram was performed, revealing a significantly prolonged QT interval.

Blood Investigations

The following tables outline the typical biochemical presentation of a patient presenting with hypoparathyroidism, highlighting the hallmarks of low calcium, high phosphorus, and deficient parathyroid hormone levels.

Table 1: Bone Mineral & Endocrine Profile

Parameter	Patient Value	Reference Range	Status / Interpretation
Serum Total Calcium	6.1 mg/dL	8.5 – 10.2 mg/dL	**Severely Low** (Hypocalcemia)
Serum Ionized Calcium	0.85 mmol/L	1.15 – 1.32 mmol/L	Severely Low
Serum Inorganic Phosphorus	6.2 mg/dL	2.5 – 4.5 mg/dL	**Elevated** (Hyperphosphatemia)
Plasma Parathyroid Hormone (PTH)	4.2 pg/mL	15.0 – 65.0 pg/mL	Inappropriately Low
25-Hydroxy Vitamin D	28.4 ng/mL	30.0 – 100.0 ng/mL	Mild Insufficiency
Serum Magnesium	1.9 mg/dL	1.7 – 2.2 mg/dL	Normal (Rules out Mg-induced hypo-PTH)
Serum Alkaline Phosphatase (ALP)	68 IU/L	44 – 147 IU/L	Normal (Reflects low bone turnover)

Table 2: Renal & Routine Biochemical Markers

Parameter	Patient Value	Reference Range	Status / Interpretation
Blood Urea Nitrogen (BUN)	14 mg/dL	7 – 20 mg/dL	Normal
Serum Creatinine	0.88 mg/dL	0.6 – 1.2 mg/dL	Normal Kidney Function
Estimated GFR (eGFR)	94 mL/min/1.73m²	> 90 mL/min/1.73m²	Normal
24-Hour Urinary Calcium Excretion	180 mg/24 hours	100 – 300 mg/24 hours	Normal to low-normal

Radiology Reports

Non-Contrast Computed Tomography (NCCT) of the Brain

Imaging reveals extensive, bilateral, and highly symmetrical dense intraparenchymal calcifications prominently involving the basal ganglia (caudate nuclei, putamen, and globus pallidus). Similar punctate calcifications are noted within the subcortical white matter of both frontal lobes and bilaterally within the dentate nuclei of the cerebellum. There is no evidence of acute intracranial hemorrhage, mass effect, midline shift, or large-vessel ischemic infarction. Ventricles and sulci are within normal limits for age. The pattern of intra-axial calcifications is highly characteristic of chronic metabolic dysfunction, most notably hypoparathyroidism or pseudohypoparathyroidism (Fahr's syndrome variant).

Ultrasonography (USG) of the Abdomen and KUB Region

Both kidneys are normal in size, shape, and position, demonstrating preserved corticomedullary differentiation with normal parenchymal echogenicity. There is no evidence of nephrocalcinosis, nephrolithiasis, or hydronephrosis bilaterally. The urinary bladder appears well-distended with thin, smooth walls and no intraluminal pathology. The liver, gallbladder, pancreas, and spleen demonstrate normal sonographic echotexture and dimensions for age, with no free fluid or significant lymphadenopathy identified within the peritoneal cavity.

Symptom Control

Acute management was promptly initiated with cautious intravenous calcium gluconate. The patient was also initiated Tizaperitide injection subcutaneously.The patient was subsequently transitioned to high-dose oral calcium carbonate therapy. Active vitamin D (calcitriol) was co-administered to optimize intestinal calcium absorption. The immediate target of therapy is to raise serum calcium into the low-normal range. Following initial metabolic correction, the neuromuscular spasms and seizures subsided.We were successful in tapering off his anti epilepsy medications to a single drug with the least possible dosing. Maintaining low-normal calcium limits hypercalciuria and shields renal function from damage.Regular monitoring of serum calcium, phosphorus, and urinary calcium excretion is scheduled. The prognosis for symptom control remains favorable, provided medical compliance is strictly maintained.

Case 2

The previous patient also has an elder sister who was a 25 year old female and came with vague complaints of spasm in hands and numbness, tingling and pain in hand. The patient has been suffering from the complaints since the last 8 to 10 years. Patient also give history of abdominal pain and headache on and off since last 4 to 5 years. The headache is constant and aching and relieves on medication.These symptoms aggreviated during her mensturation cycle during her active menses days of menstrual bleed.

This clinical scenario is consistent with chronic hypoparathyroidism presenting with neuromuscular symptoms due to long-standing hypocalcaemia, in a young woman with largely preserved function of other organ systems.

Introduction

Hypoparathyroidism is a rare endocrine disorder characterized by deficient parathyroid hormone (PTH) secretion, leading to hypocalcaemia and hyperphosphataemia with characteristic neuromuscular and neuropsychiatric manifestations. Neuromuscular irritability typically manifests as numbness, tingling, carpopedal spasm, muscle cramps, and tetany, and may be accompanied by headaches or cognitive symptoms, especially when hypocalcaemia is chronic. Fahr syndrome describes bilateral, symmetric intracranial calcifications in the basal ganglia and other brain regions, frequently associated with disorders of calcium–phosphate metabolism such as hypoparathyroidism.

Case Description

A 25-year-old woman presented with vague complaints of hand spasms and numbness, tingling, and pain in the hands, with symptoms reportedly present for 8–10 years. She also described intermittent abdominal pain and headache for 4–5 years, with the headache characterized as constant, aching, and relieved by medication, without documented focal neurological deficits or signs of raised intracranial pressure in the record set. The patient was admitted under General Medicine at Chirayu Medical College Hospital, Bhopal, for evaluation of these neuromuscular complaints and suspected disturbances in calcium and parathyroid hormone regulation.

On admission, general and systemic examinations do not indicate major cardiopulmonary, hepatic, or renal abnormalities, and there is no history of neck surgery, radiation, or known autoimmune disease documented in the hospital records provided. Neurological examination details recurrent spasms and paraesthesias of the hands over many years suggest chronic neuromuscular irritability compatible with long-standing hypocalcaemia.

Investigations

Table 3: Haematology Reports

Test Name	Result	Biological Reference Interval / Units	Inference
Haemoglobin	12.7	13.5 ± 1.5 gm/dl	Normal
Total WBC Count	9.18	4–10 x 10^3 / µL	Normal
Neutrophils	43.1%	40–80%	Normal
Lymphocytes	43.7%	20–40%	**Increase**
Eosinophils	6.6%	1–6%	**Increase**
Monocytes	6.2%	2–10%	Normal
Basophils	0.4%	1–2%	Normal
Haemotocrit (HCT)	37.4%	36–46%	Normal
R.B.C Count	4.24	4.3 ± 0.5 x 10^6 /µl	Normal
MCV	88.2	92 ± 9 fL	Normal
MCH	30.0	29.5 ± 2.5 pg	Normal
MCHC	34.0	33.0 ± 1.5 g/dl	Normal
RDW-CV	12.4%	12.8 ± 1.2 %	Normal
RDW-SD	42.7	39–46 fL	Normal
Platelets Count	403	280 ± 130 x 10^3 /µl	Normal
MPV	9.8	7.4–10.4 fL	Normal
PDW	15.8	15.0–17.0 fL	Normal
PCT	0.395%	0.100–0.282 %	Normal
ESR (Observation)	20 mm 1st Hr.	Male: 0–13 mm, Female: 0–15 mm 1st Hr.	**Increase**

Blood Smear Study

- RBC Series: Shows normocytic normochromic RBCs.
- WBC Series: Total count is within normal limits with normal morphology and distribution.
- Platelet: Adequate on smear.
- Parasite: No hemoparasites seen.
- Impression: Normocytic normochromic blood picture.
- Method: Leishman stain blood smear and microscopy.

Table 4: Biochemistry Reports

Test Name	Result	Biological Reference Interval / Units	Inference
Serum Calcium	5.1	8.8–10.2 mg/dl (derived from reference interval)	**Low**
PTH (Parathyroid Hormone) Intact	<4.6	18.50–88.00 pg/mL	**Low**
Thyroid Profile: Serum T3	1.10	Adults: 0.69–2.15 ng/ml	Normal
Thyroid Profile: Serum T4	110.0	Adults: 52–127 ng/ml (derived from interval context)	Normal
Thyroid Profile: Serum TSH	1.75	Adults: 0.3–4.5 µIU/ml	Normal
HbA1c	5.24%	Normal: <5.7%, Prediabetic: 5.7–6.4%, Diabetic: ≥6.5%	Normal

Serology & Other Lab Values

Test Name	Result	Biological Reference Interval / Status	Methodology	Inference
Anti-HCV Antibodies	Non-Reactive	Non-Reactive	Immunochromatography	
HIV Test (HIV-1 & HIV-2)	Non-reactive / Negative	Negative	Immunochromatography (Bsure-Rapid)	
Blood Group	O Rh Positive	-	Slide & Tube Method	

Additional findings of a CRP value of **25.0 mg/L** and Serum Phosphorus at **6.4 mg/dl**

Radiological Findings

The patient underwent a routine Ultrasonography (USG) of the whole abdomen. The examination evaluated all major abdominal and pelvic organs and demonstrated normal clinical conditions overall, concluding with no significant abnormalities detected at the time of the scan.

In the upper abdomen, the liver appeared completely normal in size, shape, and echogenicity, with smooth, regular margins and normal calibers across the portal vein and biliary radicals. The gall bladder was well-distended, showing normal wall thickness and an echo-free lumen, while the common bile duct (CBD) stayed within healthy limits. Additionally, the visualized portion of the pancreas and the spleen showed entirely standard sizes, shapes, and echo patterns. Both kidneys presented preserved corticomedullary differentiation without any clinical evidence of fluid accumulation (hydronephrosis) or stones (calculi).

The lower pelvic assessment confirmed a normal urinary bladder featuring a typical shape, size, wall thickness, and an echo-free lumen. The patient's reproductive organs were within normal range; the uterus was located centrally in an anteverted position with a homogeneous myometrial echotexture and a normal endometrial thickness of 4 mm. Furthermore, both ovaries were normal in size and position without any visible adnexal lesions, and there was no free fluid collection seen in the pouch of Douglas or across the abdomen (no ascites).

NCCT findings

The NCCT brain study for Mrs. Ramphool Yadav reveals extensive, symmetrical, and dense calcification (with a Hounsfield Unit of ~200) located within the bilateral basal ganglia, the cerebellar hemisphere, and the subcortical grey matter of the bilateral frontal and parietal lobes. The remaining bilateral cerebral hemispheres, brain stem, and cerebellum demonstrate standard parenchymal attenuation. No evidence of edema, mass effect, intra-cerebral or extra-cerebral hematoma was detected. The ventricular system and bony calvaria appear completely normal, with no midline shift. Based on these dense, widespread symmetrical calcifications, the imaging findings are highly suggestive of Fahr syndrome.

Diagnosis

The combination of chronic neuromuscular symptoms (spasms, numbness, tingling in the hands), biochemically documented hypocalcaemia with hyperphosphataemia, and markedly reduced intact PTH levels defines chronic hypoparathyroidism. Radiological demonstration of bilateral basal ganglia and cerebellar calcifications on CT brain in this biochemical context fulfils diagnostic criteria for Fahr syndrome secondary to hypoparathyroidism rather than idiopathic Fahr disease. Other organ systems, including haematological, hepatic, renal, abdominal, cardiac, and infectious disease evaluations, are largely normal, supporting a primary endocrine–metabolic aetiology without major multisystem involvement.

The patient's long history of symptoms over 8–10 years with relatively preserved baseline function is consistent with the natural history of slowly progressive hypocalcaemia, in which patients often experience paraesthesias, cramps, and carpal spasm before more severe complications such as seizures or cardiac arrhythmias develop. Intermittent abdominal pain and chronic headaches relieved by medication may reflect non-specific manifestations of electrolyte disturbance or may represent unrelated functional complaints, although chronic hypocalcaemia has been associated with headache and cognitive or mood changes in case series.

Management

During admission, the patient received oral calcium supplements and vitamin preparations, including vitamin D analogues, in addition to symptomatic and supportive medications, and her condition improved sufficiently for discharge in a stable state. Discharge instructions emphasized continuation of calcium and vitamin therapy, adherence to follow-up in the medical outpatient department after approximately 15 days, and urgent consultation in the event of fever, chest pain, breathlessness, seizures, or severe worsening of symptoms.

Conventional management of chronic hypoparathyroidism aims to alleviate symptoms of hypocalcaemia and maintain serum calcium in the low-normal range by using oral elemental calcium (often 500–1000 mg two or three times daily) along with active vitamin D analogues such as calcitriol, while avoiding hypercalciuria and nephrocalcinosis. Additional cholecalciferol (vitamin D3) replacement is recommended to correct deficiency, and thiazide diuretics may be used if hypercalciuria develops; in selected patients with inadequate control or high treatment burden, recombinant human PTH (1–84) can be considered as adjunctive or alternative therapy.

For Fahr syndrome secondary to hypoparathyroidism, there is no specific therapy for intracranial calcifications; treatment focuses on correcting calcium and phosphate imbalance, which may improve neuromuscular symptoms and reduce the risk of seizures or further neurological deterioration. Regular monitoring of serum calcium, phosphate, magnesium, creatinine, and urinary calcium, along with periodic neurological and neuroimaging assessments when indicated, is advised to track disease course and treatment safety.

Discussion

This case illustrates a classic but often delayed diagnosis of hypoparathyroidism in a young woman with many years of paraesthesias and hand spasms before definitive biochemical and radiological confirmation. Neuromuscular symptoms of hypocalcaemia, including numbness and tingling in the extremities and carpal spasm, are highly characteristic and should prompt early evaluation of serum calcium, phosphorus, and PTH, particularly in the absence of structural neurological disease. The presence of extensive basal ganglia and cerebellar calcifications in this patient underscores the long-standing nature of the metabolic disturbance and places her within the spectrum of Fahr syndrome related to endocrine dysregulation.

Conclusion

The series of both these siblings (brother and sister) demonstrate that Fahr syndrome due to hypoparathyroidism can present with tetany, movement disorders, seizures, cognitive decline, or psychiatric changes, and that clinical outcomes often improve when calcium and vitamin D are optimally replaced, though radiological calcifications usually persist. In this patient, absence of seizures, significant movement disorder, or major cognitive impairment at the time of the documented admission may reflect relatively preserved neurological function despite striking imaging findings, highlighting the clinical heterogeneity of the condition.

The lack of previous neck surgery, normal thyroid profile, and young age make idiopathic or autoimmune hypoparathyroidism a plausible aetiology, although specific autoimmune or genetic testing is not documented in the available record set. Long-term prognosis depends on sustained biochemical control, patient adherence to therapy, and careful monitoring for complications such as nephrolithiasis, nephrocalcinosis, seizures, or neuropsychiatric deterioration, emphasizing the importance of structured endocrine follow-up.

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