Case Report

Unusual Presentation of Primary Hyperparathyroidism in a Young Girl: A Case Report and Review of Literature.

Rokonuzzaman SM[1], Saha A[2], *Islam MM[3] Alam MM[4], Rashid MM[5]

Touky AH[6], Alam F[7], Al Amin RM[8], Oyon RT[9]

Abstract

Primary hyperparathyroidism presenting as a severe bone disease in adolescents is rare. We herein report a case of 21 years old Bangladeshi Muslim female, initially presenting with progressive height loss, generalized weakness, gradual weight loss and then gradually developed large joint pain. Subsequent biochemical, radiographic & scintigraphic findings were consistent with primary hyperparathyroidism due to a parathyroid adenoma

Keywords: Hyperparathyroidism, Bone disease in adolescents, Parathyroid adenoma.

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Background

Primary hyperparathyroidism is a disorder of one or more of the parathyroid glands. It is the third most common endocrine disorder after diabetes mellitus & thyroid disease. The parathyroid glands become overactive & secrete excess amounts of parathyroid hormone. As a result, blood calcium rises to a level that is higher than normal. Population screening in the context of bone health has led to the identification of a new clinical entity, normocalcemic primary hyperparathyroidism (PHPT).[1] These patients have persistently elevated parathyroid hormone (PTH) levels despite normal serum calcium concentrations when causes of secondary hyperparathyroidism have been excluded. The incidence of PHPT is increasing with a rate of 42 in 100,000 per year. While in women over 60 years of age the average annual incidence approaches 190: 100,000 per year.[1,2]

1. Prof. Dr. S M Rokonuzzaman, Professor, Dept. of Medicine, Uttara Adhunik Medical College, Dhaka.
2. Dr. Avijit Saha, Assistant Registrar, Dept. of Medicine, Uttara Adhunik Medical College, Dhaka.
3. Dr. Mir Moyeedul Islam, Associate Professor, Dept. of Pharmacology, Ad-din Sakina Women's Medical College, Jashore.
4. Dr. Md Masud Alam, Medical Officer, Dept. of Medicine, Uttara Adhunik Medical College, Dhaka.
5. Dr. Md. Mamunur Rashid, Medical Officer, Department of Neurosurgery, Uttara Adhunik Medical College, Dhaka.
6. Dr. Ashfaque Hossain Touky, Assistant Registrar, Dept. of Neurosurgery, Uttara Adhunik Medical College, Dhaka.
7. Dr. Farzana Alam, Medical Officer, Dept. of Gastroenterology, Uttara Adhunik Medical College, Dhaka.
8. Dr. Rufaidah Mohammed Al Amin, Intern Doctor, Dept. of Medicine, Uttara Adhunik Medical College, Dhaka.
9. Dr. Ragib Tahmid Oyon, Intern Doctor, Dept. of Medicine, Uttara Adhunik Medical College, Dhaka.

***Correspondence:** E-mail: ofuronto.shopno@gmail.com

Case Presentation

A 21-year-old Muslim female with no known comorbidities was admitted in the Medicine ward of Uttara Adhunik Medical College Hospital on 24.02.22 with the complaints of generalized weakness for 1 year, progressive height loss of 1ft from 4.9 feet to 3.9 feet within 3 years, gradual weight loss of 15 kgs from 45kgs to 30kgs over 2 years & multiple large joints pain for 2 months which increases on movement but no associated stiffness. She does not give any significant family history.

On examination, she was markedly undernourished, short stature & had pigeon chest. Rest of her physical examination findings were unremarkable. Palpation of thyroid gland did not reveal a goiter or discrete thyroid nodule. Respiratory, cardiovascular & nervous system examination revealed no abnormality. Musculoskeletal system reveals no abnormality except pain of large joints on movement.

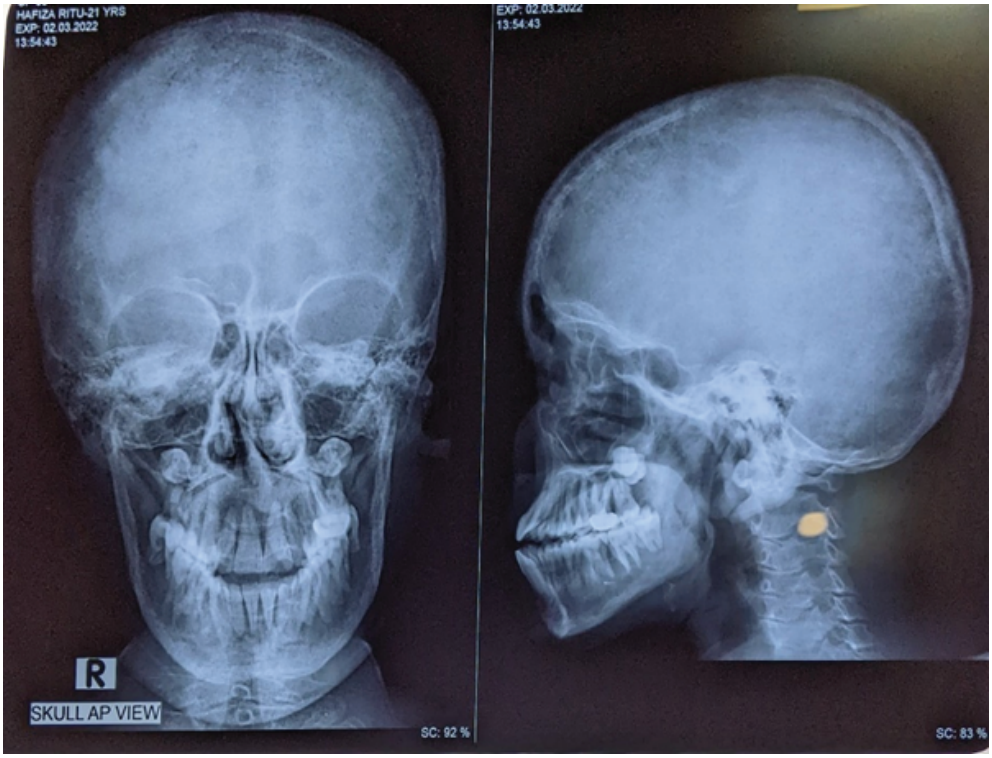
Laboratory investigation revealed as follows (Table I)

Table: I : Laboratory test results

Parameters	Test Result	Parameters	Test Result
Hb%	13.8 g/dL	**FT_4**	0.92mg/dL
ESR	15 mm in 1st hour	**HLA B-27**	Negative
TC WBC	6×10^3 / µL	**Anti-CCP**	0.9 (Negative)
TC RBC	4.8×10^6 / µL	**R.A/R.F test**	<8 IU/mL
TC Platelet	320×10^3 / µL	**S. Na^+**	138 mmol/L
S. calcium	10.3 mg/dL	**S. K^+**	3.0 mmol/L
Inorganic phosphate	1.8 mg/dL	**S. Cl^-**	106 mmol/L
Serum albumin	37 g/L	**S. HCO_3^-**	19 mmol/l
Alkaline phosphatase	2652 U/L	**S. urea**	13 mg/dL
25-OH Vit D	7 ng/mL	**S. creatinine**	0.49mg/dL
IPTH	>2500 pg/mL	**S. uric acid**	3.7mg/dL
S. TSH	4.24 µIU/mL	**GFR**	60 mL/min/1.73m²

TC; Total count, IPTH; Intact parathyroid hormone, GFR; Glomerular filtration rate, TSH; Thyroid-stimulating hormone, Anti-CCP; Anti-cyclic citrullinated peptide

Bone densitometry of femur & spine by DXA showed osteoporosis with T score value: <-7.5 SD. Chest X-ray P/A view was normal. X-ray dorso-lumbar spine revealed diffuse osteopenia and multiple partially collapsed vertebrae, but the disc spaces were maintained with intact pedicles and apparently normal sacroiliac (SI) joints. Skull x-ray (both views) showed prominent trabecular markings with cortical thinning and mottled 'salt & pepper' or 'pepper-pot' appearance of cranial vault (Photograph 1)

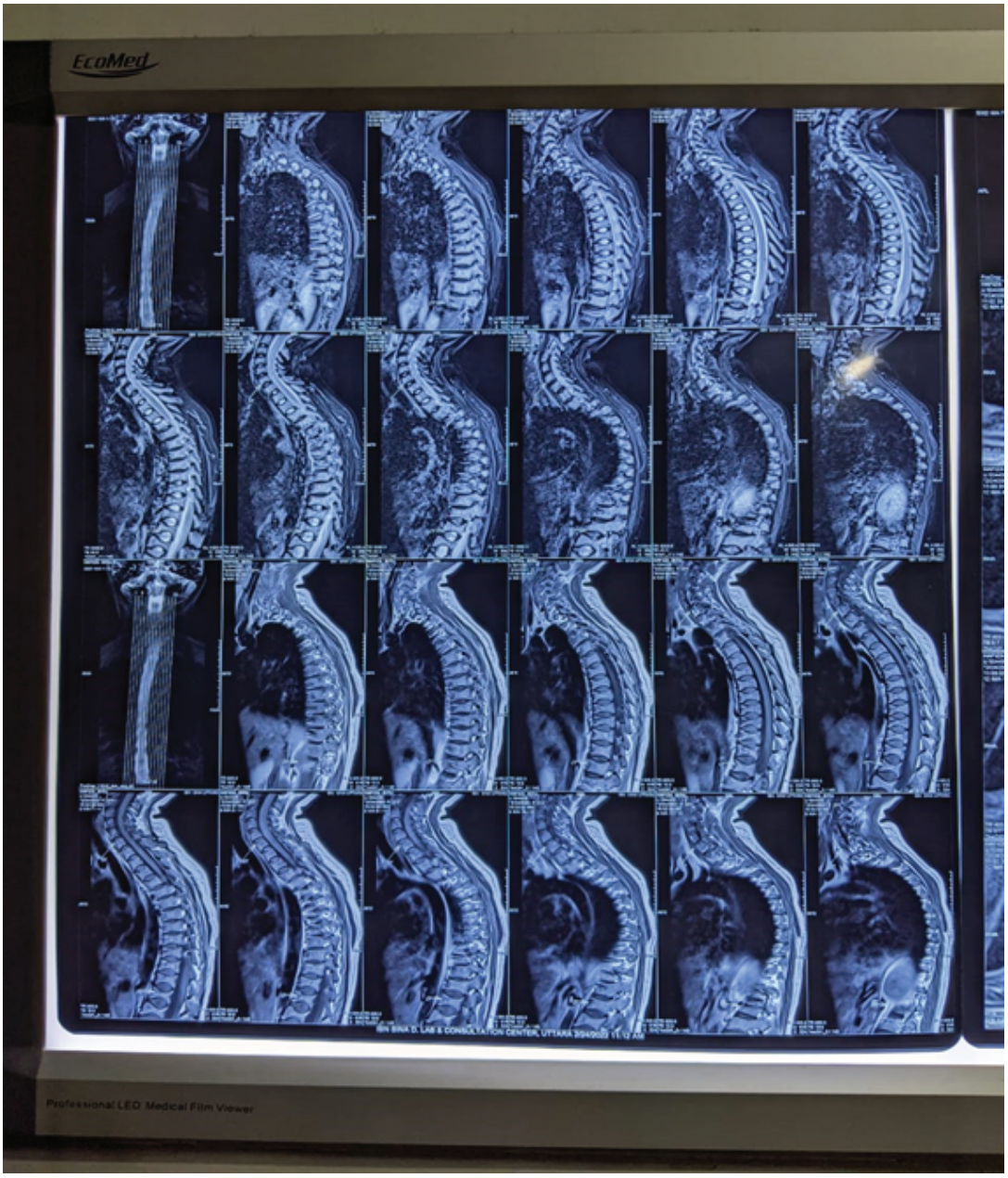


Photograph 1: X-ray skull (both view)

USG of neck showed mildly swollen (28×12) mm right lobe of thyroid gland. USG of whole abdomen revealed multiple bright echogenic structures with posterior acoustic shadow within the lumen of gallbladder suggestive of cholelithiasis.

MRI of spine was suggestive of kyphoscoliosis at dorsolumbar region with variable degree of compression collapse at multiple dorsal & lumbar vertebrae. There were no evidence of disc herniation, neuroforaminal narrowing or canal stenosis (Photo 2). MRI of brain was unremarkable with diffuse thickened calvarium.

Contrast CT scan of abdomen showed generalized osteopenia, intracortical lucency / ground glass opacity was seen in ribs, vertebrae, pelvic bones



Photograph 2: MRI of spine

and sacrum. Subchondral bone resorption was also noted. Collapses of all dorso-lumbar vertebral bodies were seen (Cod-fish vertebrae). Fracture of superior pubic ramus was also noticed on the right side. Right renal calculus (2mm) seen in mid calyx. Tc-99m sestamibi scanning was positive for parathyroid adenoma within the right lobe of thyroid gland.

Considering the history, clinical examination findings and investigation reports, we diagnosed her as primary hyperparathyroidism most likely due to parathyroid adenoma. Before reaching the confirmatory diagnosis, we gave her symptomatic treatment that included non-steroidal anti-inflammatory drug (NSAID) for the joint pain, nutritional support for undernourishment, vitamin D injection for severe vitamin D deficiency and bisphosphonate for osteoporosis. Her symptoms improved slightly following the treatment. We also consulted neurosurgeon after the report of MRI of spine. After confirming the diagnosis by parathyroid scintigraphy test report, we transferred her to the department of ENT.

Discussion

The incidence rate of primary hyperparathyroidism varies from 34 to 120 cases per 100,000 individuals, and it increases with age. In any case, Primary hyperparathyroidism is relatively rare in the clinical setting. As a result, some clinicians have inadequate understanding of this disease, fail to make a timely and correct diagnosis, and are prone to misdiagnose. Because this disease mainly manifests as pathological fractures most patients are first diagnosed in the department of orthopedics or bone oncology, resulting in a high rate of delayed treatment or misdiagnosis.[3] So, once confirmed, timely excision of the diseased parathyroid glands is considered to be the most preferred method for the treatment of primary hyperparathyroidism with severe bone disease, rather than treatment of skeletal system disease only.[4]

The discussed case is a relatively rare case of multiple skeletal destructions in addition to primary hyperparathyroidism. This patient had longer duration and continuous bone destruction due to misdiagnosis. She already presented with indicative bone manifestations in imaging scans and bone deformities. Therefore, the potential pathological mechanisms underlying bone destruction in primary hyperparathyroidism patients should be further explored. In patients suffering from primary hyperparathyroidism over a long-term period, it always co-exists with severe vitamin D deficiency, which could have devastating consequences on the skeleton as seen in this very case.[5,6]

This degree of severity of skeletal disease in the Asian population affected by primary hyperparathyroidism has consistently been observed across various studies from the region. A study from India showed that bone pains and painful proximal myopathy were the commonest presentation (47%), followed by pathological fractures in 23.5% cases.[7]

Conflicts of interest

The authors declare no conflict of interest regarding the publication of this paper.

Conclusion

In conclusion, primary hyperparathyroidism should be kept in mind in all patients presenting with longstanding history to bone problems, from simple bone pain to severe bone disease and hypercalcemia. Elevated serum levels of PTH confirm the diagnosis.

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