PRIMARY HYPERPARATHYROIDISM WITH RICKETS

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Presenting Complaints

- 17 year old developmentally normal adolescent boy, first of a twin pregnancy, born to a non consanguineous marriage.

- Came to orthopaedics with both bone fracture of left forearm.

- In view of short stature he was referred to paediatrics.

- The child was apparently normal till 7 years of age, when the mother noticed he was shorter than his peers but she attributed this to the fact he was one of the twin and did not give much attention. From the age of 15 years, child developed bending of both legs which was again neglected.
Three weeks before presentation, child had a fall from a bicycle, developed fracture of left lower end of radius and ulna, when he presented to orthopaedic department.

There is no history of bone pain, polyuria and polydipsia.

There is no history suggestive of liver or kidney disease.

He consumes a mixed diet and gets 2-3 hrs sunlight exposure (9.00 am - 4.00 pm).
On Examination

Conscious / Oriented

No pallor / icterus / cyanosis / clubbing / generalised lymphadenopathy / pedal edema

Hemodynamically stable

Weight : 30.5 kgs ( < 3rd percentile acc. WHO chart )
Height : 128 cm ( < 3rd percentile acc. WHO chart )
US : LS ratio : 0.8 :1
HC : 52 cm

Mid Parental Height : 153.5 cm
- No dysmorphic facies

- No rachitic rosary / widening of wrist / frontal bossing

- Lower limbs had windswept deformity – right genu valgus deformity, left genu varus deformity, with no double malleolus.

- System Examination was within normal limits.
Diagnosis

- Bilateral lower limb deformity due to metabolic bone disease
- Then our evaluation began ..................
Investigations

<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Normal Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>SERUM CALCIUM</td>
<td>12.1 mg/dl</td>
<td>(8.5 – 10.1 mg/dl)</td>
</tr>
<tr>
<td>SERUM PHOSPHOROUS</td>
<td>2.1 mg/dl</td>
<td>(2.5 – 4.9 mg/dl)</td>
</tr>
<tr>
<td>SERUM ALKALINE PHOSPHATASE</td>
<td>2761 U/L</td>
<td>(65 – 260 U/L)</td>
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</tbody>
</table>

- Liver function tests were normal.
- Renal function tests were normal.
- Serum electrolytes were normal.
Further Evaluation

- Vitamin D levels done was 10.68 ng/ml (< 20 – deficiency)
- Thyroid Function Tests were normal.
- Parathormone levels done 1764.90 pg/ml (12-65 pg/ml).
- Serum cortisol and Serum prolactin levels done were normal.
PRIMARY HYPERPARATHYROIDISM
- USG neck - Left parathyroid adenoma.

- Parathyroid scan (Technetium 99m sestamibi) revealed hyperfunctioning of left inferior parathyroid gland.
PRIMARY HYPERPARATHYROIDISM SECONDARY TO LEFT PARATHYROID ADENOMA
Endocrinology Opinion

- Suggested 3 and a half parathyroidectomy
Management

- Three and Half parathyroidectomy was done.

- Intra op findings – Left inferior parathyroid adenoma along with ectopic parathyroid gland in the carotid sheath.

- Adenoma removed and sent for histopathological examination.

- Post op period – Child had hypocalcemia started on calcium infusion. Calcium levels were monitored and gradually made to oral calcium supplements.

- Inj vitamin D 6 lakh units was given.

## Post Op Labs

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>SERUM CALCIUM</td>
<td>8.1 mg/dl (8.5 – 10.1 mg/dl)</td>
</tr>
<tr>
<td>SERUM PHOSPHOROUS</td>
<td>3.4 mg/dl (2.5 – 4.9 mg/dl)</td>
</tr>
<tr>
<td>SERUM ALKALINE PHOSPHATASE</td>
<td>1433 U/L (65 – 260 U/L)</td>
</tr>
<tr>
<td>PARATHORMONE LEVEL</td>
<td>129 pg/ml (12 – 65 pg/ml)</td>
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</table>
Follow up

- Child was discharged with oral calcium and to follow up after 2 weeks
- After 2 weeks,

<table>
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<tr>
<th>Test</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>SERUM CALCIUM</td>
<td>8.2 mg/dl (8.5 – 10.1 mg/dl)</td>
</tr>
<tr>
<td>SERUM PHOSPHOROUS</td>
<td>4.1 mg/dl (2.5 – 4.9 mg/dl)</td>
</tr>
<tr>
<td>SERUM ALKALINE PHOSPHATASE</td>
<td>875 U/L (65 – 260 U/L)</td>
</tr>
<tr>
<td>PARATHORMONE LEVEL</td>
<td>94 pg/ml (12 – 65 pg/ml)</td>
</tr>
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</table>
To continue oral calcium for 3 - 6 months.

- Orthopaedic intervention for mal-union of both bone fracture of left forearm and corrective surgery for the windswept deformity.
Review Of Literature
52 patients were identified with hyperparathyroidism. 2 Had MEN type 1.

34 patients (65%) had single parathyroid adenoma

27% - 16 patients had hyperplasia.

Most common presentation was rickets in the adolescent age group.

* Primary hyperparathyroidism in children – josh kollars. Pediatrics vol 115 No.4 April 1 2005 pp 974-980
In a retrospective study of primary, from 2004 to 2010, a single institution study, 522 patients underwent surgery for primary hyperparathyroidism.

- 7 patients were under the age of 19. (4 – male; 3 – female). All had single parathyroid adenoma.

- 5 underwent parathyroid adenoma removal, 1 double parathyroidectomy and 1 subtotal thyroidectomy.

* Primary hyperparathyroidism in children and young adults – a single institution experience. Pauvonic et al; Acta chir belg 2013; jan to feb; 113(1)35-9.
In a retrospective analysis of data from the primary hyperparathyroidism registry of a north indian tertiary care teaching institute,

- Of 184 histopathologically proven primary hyperparathyroidism,

- In children and adolescents, rickets is the commonest presentation of primary hyperparathyroidism while renal stones is common in adults.

A recent case report,

- A 12 year old girl presenting with short stature, genu valgum, eversion deformity at the ankle joints and flat feet.

- Radiographs – suggestive of rickets

- Labs – hypercalcemia, hypophosphatemia, vitamin D deficiency and elevated PTH

- 99m Tc sestamibi scan – increased uptake at lower pole of right lobe of thyroid

- Right inferior parathyroidectomy was done, HPE – parathyroid adenoma.

* Primary hyperparathyroidism presenting as rickets, diagnostic challenge and treatment outcomes - Deep Dutta, Jclin Res Endocrinol 2013;266-269.
A 15 year old patient, with severe bone deformities. Radiographs suggestive of rickets.

Surgical exploration revealed a parathyroid adenoma.

It was removed and patient improved symptomatically.

A case report in 1956,

- A 12 year old presented with left forearm both bone fracture,
- Radiograph suggestive of rickets.
- Labs – hypercalcemia, hypophosphatemia and elevated PTH
- Surgical exploration – left parathyroid adenoma.
- Following surgery, values normalised.

*Parathyroid adenoma in child presenting as rickets,* - B.S.B Wood, Arch Dis Child 1958;33;46-48
Case Report of 14 year old girl, genu valgum for 6 years.

Multiple bony defects.

Primary hyperparathyroidism – hypercalcemia, Hypophosphatemia, elevated ALP and elevated PTH.

Thallium – Technitium subtraction scan – left superior parathyroid adenoma.

Parathyroid adenoma presenting as genu valgum is very rare.

* Primary hyperparathyroidism presenting as rickets – P.S. Menon; Journal of Pediatrics and Child Health 1994; 30; 441-3.
Take Home Message

• Hyperparathyroidism in adolescents presents as rickets.
• All children with radiological rickets - S.Calcium, S.Phosphorus, alkaline phosphatase
• All children with hypercalcemia need investigation for hyperparathyroidism.
THANK YOU