Report of a Case of Pseudo-pseudo-hypoparathyroidism

by
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In 1942, Albright was attributed with naming pseudohypoparathyroidism, a disease characterized by the signs of a short stature, obesity, a round face, short fourth and fifth metacarpals, hypocalcemia and hyperphosphoremia. Subsequently, the cases with similar physical characteristics, but without hypocalcemia and hyperphosphoremia, were designated pseudo-pseudohypoparathyroidism. However, today these two diseases are regarded as identical, with the differences based simply on the time of onset of the symptoms. Very recently, we encountered a case of this disorder, which is deemed to be very rare.

The details of this case are described in the following.

The case was a forty-one year old male with the chief complaints of bilateral hip pain and knee pain.

The past history of illness of this patient revealed seven bone fractures around both elbow joints, between the ages of six and sixteen years. The patient began to lose his teeth by the age of thirty and now presented completely edentulous. There was no past history of seizures or tetany.

The family history disclosed that his mother was 146 cm in height with a round face. The patient's present symptom was right hip pain which developed in March 1982. His pain was so intensified by minor trauma that he could not walk. His knee joints were immobilized with casts for approximately three weeks (elsewhere) with a remission of the symptoms. However, in September 1982, he was again affected by right knee and right hip pain, which gradually become bilateral. The patient then visited our clinic in November 1982.

Present illness: His height was 165 cm and his weight was sixty-four kg. The patient did not present with a round face, but blue sclera was identified. There was no restriction of the range of motion in his lower extremities. There was, however, laxity in his right knee joint, but no inflammation, tenderness or joint effusion were noted. The Trousseau's and Chvostek's signs were negative.

Biochemical findings: There was no abnormalities in his general hemogram or urinalysis. His blood levels of calcium and phosphorus were within normal ranges. A rise in Al-p, a mild rise in leucocyte precipitation and a positivity of CRP were identified. There were no abnormal results in the endocrine test (Table 1).

Increased excretion of phosphorus in the urine and a rise in cyclic-AMP were observed
Radiographic findings: Plain X-rays of the skull showed diffuse hyperplasia and diffuse granular shade. Loss of teeth was also observed (Fig. 2). Cerebral C.T. examination revealed no calcification of the cerebral basal lamina. Osteoporosis was identified in lumber vertebrae, but there were no compression fractures (Fig. 3). The joint spaces of both hips were narrowed. Severe osteoporosis was identified in the femur, especially in the intertrochanteric area (Fig. 4). Osteoporosis was also noted in the tibia, but there was no osteoarthritic changes in the knee joints. Transverse lines were seen clearly in the area of proximal third of the both tibia (Fig. 5).

Malunions (possibly attributable to fractures) were seen around both elbow joints, as in the results of the Ellsworth-Howard test (Fig. 1).

Table 1: Laboratory findings

<table>
<thead>
<tr>
<th>Peripheral blood</th>
<th>Liver function</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hb</td>
<td>GOT 39</td>
</tr>
<tr>
<td>RBC</td>
<td>GPT 42</td>
</tr>
<tr>
<td>Htc</td>
<td>LDH 226</td>
</tr>
<tr>
<td>WBC</td>
<td>AL-P 23.2 (K-A-U)</td>
</tr>
<tr>
<td>Erythrocyte Sedimentation Rate 30/58</td>
<td>Isozyme II III</td>
</tr>
<tr>
<td>CRP (+)</td>
<td>r-GTP 21</td>
</tr>
<tr>
<td>Blood chemistry</td>
<td>TTT 8.1</td>
</tr>
<tr>
<td>BUN</td>
<td>LAP 206</td>
</tr>
<tr>
<td>creatinin</td>
<td>T-protein 6.8</td>
</tr>
<tr>
<td>T, Chol</td>
<td>Alb 50.4%</td>
</tr>
<tr>
<td>Na</td>
<td>α₁ 8.2%</td>
</tr>
<tr>
<td>K</td>
<td>α₂ 8.2%</td>
</tr>
<tr>
<td>Cl</td>
<td>β 11.1%</td>
</tr>
<tr>
<td>Ca</td>
<td>r 21.8%</td>
</tr>
<tr>
<td>P</td>
<td></td>
</tr>
<tr>
<td>Mg</td>
<td></td>
</tr>
<tr>
<td>Endocrine test</td>
<td>other test</td>
</tr>
<tr>
<td>%TRP 95%</td>
<td>Chromosome 46-XY</td>
</tr>
<tr>
<td>PTH 0.4 ng/ml</td>
<td>hydroxy proline (urine) 412.7 mc.mol/L</td>
</tr>
<tr>
<td>1,25(OH) vitamin 25.6 pg/ml</td>
<td></td>
</tr>
<tr>
<td>T₃ 1.0 ng/ml</td>
<td>human PTH 1000U</td>
</tr>
<tr>
<td>T₄ 6.8 g/dl</td>
<td>0-4 17 18</td>
</tr>
<tr>
<td>17-KS 7.9 mg/day</td>
<td>0.4 1.0 2.0 3.0 4.0</td>
</tr>
</tbody>
</table>

Fig. 1: Inorganic P reaction and C-AMP test are both positive.
well as associated osteoporosis and a mild shortness of the fourth and fifth metacarpals (Fig. 6, 7).

Some deep absorption fossae were observed histologically on the surface of the trabecle taken from the left ilium. Few osteoblastic faces surrounded by osteoids were observed, and the osteoid seam were thin (Fig. 8).

Following admission to our hospital, Elcatonin (eel calcitonin) was administered to relieve hip and knee pain and to inhibit bone resorption. Subsequently, Alfarol (1α-OH-D3) was given and his pain resolved. During this treatment, the level of serum calcium was unstable and often below normal limits (Table 2).
In 1942, Albright reported a disease characterized by short stature, obesity, a round face and bradyphalangia as well as hypocalcemia and hyperphosphoremia, and named the disorder pseudohypoparathyroidism. Subsequently, the cases with similar physical characteristics, but without hypocalcemia or hyperphosphoremia were named pseudo-pseudohypoparathyroidism. Today, the two diseases are deemed to be identical, the differences due to the time of evaluation.

Generally, the diagnosis of pseudohypoparathyroidism is made by (1) the presence of hypoparathyroidism, (2) its separation from other types of hypoparathyroidism and (3) physical finding associated with pseudohypoparathyroidism.

In our case, the history of hypocalcemia was suspected, evidenced by multiple fractures...
in childhood and the loss of teeth in early adulthood. The patient was still prone to hypo-
calcemia. The presence of pathologic hypoparathyroidism was thus confirmed. Using
blood levels of calcium, phosphorus, and parathyroid hormone and the results of the Ells-
worth-Howard test, this case was diagnosed as pseudo-pseudohypoparathyroidism. However,
calcification of the cerebral basal nucleus was not observed on X-ray, nor was there short-
ness of height, obesity or a round face. But osteoporosis occurred in the entire skeleton.
And a shortness of the metacarpals, as claimed by Albright, was identified.

It was assumed that both the chronic hypocalcemia and the effects of an increased
endogenous parathyroid hormone prompted resorption of bone mineral.

Administration of the Elcatonin (eel calcitonin) and activated type of Vitamin D (1α-
OH-D3) was effective to relieve pain in the lower extremities. However, no increase in
bone volume was not confirmed histologically. Further investigation is thought to be neces-
sary regard to these respects in future.

Summary

(1) A case of pseudo-pseudohypoparathyroidism was reported.
(2) A generalized bone resorption pattern was seen with the chief complaints of bila-
teral hip and knee pain.
(3) The diagnosis of pseudo-pseudohypoparathyroidism was based on the findings of
Albright's hereditary osteodystrophy, normal levels of serum calcium, phosphorus and nor-
mal Ellsworth-Howard test, though this case was an atypical one in respects of Albright's
criteria.

Acknowledgement

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References


