A CASE WITH ACRODYSOSTOSIS ASSOCIATED WITH HORMONE RESISTANCE
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Acrodysostosis
- Rare, <50 patients in literature
- Heterozygote mutations in \( PRKAR1A \) or \( PDE4D \)
- Characteristic features
  - Short tubular bones in hands and feet
  - Cone-shaped epiphyses
  - Broad nasal root
  - Various abnormalities of mandible, skull and spine
  - Short stature
  - Mental retardation
  - Might be confused with pseudohypoparathyroidism due to hormone resistance (\( PRKAR1A \)).

12-year-old

Complaint / History
- Shortness in hand and foot fingers since birth
- No additional complaint, no similar patient in the family

Past medical history
- Normal birth weighing 2800 g
- Poor school performance

Family history
- Parents were not relatives but from the same village

Physical examination (see Figures)
- Weight 45 kg (SD score 0.50)
- Height 143.7 cm (SD score -0.83)
- Upper/lower segment 0.95, normal 1.05 – 0.82
- Synophrys, curved eyebrows, low-set ears and short hands and feet
- Testes 8 cc/ 10 cc
- Bilateral optic atrophy, more apparent on the left

Imaging/laboratory
- Cone-shaped epiphyses and short tubular bones in hands and feet (Figures)
- Bone age was compatible with chronological age

Laboratory Analysis

<table>
<thead>
<tr>
<th>Result</th>
<th>Normal interval</th>
</tr>
</thead>
<tbody>
<tr>
<td>Calcium</td>
<td>9.5 mg/dl</td>
</tr>
<tr>
<td>Phosphorus</td>
<td>6 mg/dl</td>
</tr>
<tr>
<td>ALP</td>
<td>304 IU/L</td>
</tr>
<tr>
<td>PTH</td>
<td>441 pg/ml</td>
</tr>
<tr>
<td>25(OH)D</td>
<td>12.4 ng/ml</td>
</tr>
<tr>
<td>sT3</td>
<td>4.7 pg/ml</td>
</tr>
<tr>
<td>sT4</td>
<td>1.02 ng/dl</td>
</tr>
<tr>
<td>TSH</td>
<td>11.5 mIU/ml</td>
</tr>
<tr>
<td>Anti TG</td>
<td>negative</td>
</tr>
<tr>
<td>Anti TPO</td>
<td>negative</td>
</tr>
<tr>
<td>Thyroid USG</td>
<td>2.79 ml (SD score -1.52), gland echogenicity was slightly decreased, no nodules</td>
</tr>
</tbody>
</table>

Genetic Analysis

- Division of Medical Genetics, Tepecik Training and Research Hospital
- Heterozygous \( c.1102C>T \) (p.R368X), 11th exon of \( PRKAR1A \).

Discussion
- Most patients are determined as sporadic though they are inherited as acrodysostosis autosomal dominant.
- Hormone resistance can be detected together with acrodysostosis.
  - Dysfunction of cAMP regulator protein kinase A due to heterozygote \( PRKAR1A \) mutations
  - First reported in 2011
- Given elevated PTH and TSH levels and optic atrophy in our patient, \( PRKAR1A \) mutation was suspected and a known mutation was identified.
  - It is the first acrodysostosis case diagnosed with \( PRKAR1A \) mutation in our country

Conclusion
- Acrodysostosis may accompany hormone resistance and should be kept in mind in the differential diagnosis of pseudohypoparathyroidism.

References