BARIKAT SYNDROME: AN UNCOMMON CAUSE OF HYPOCALCEMIA

Teresa Rego, Fernando Fonseca, Ana Agapito
Endocrinology Department, Curry Cabral Hospital, C.H.L.C., Lisbon, Portugal

INTRODUCTION

✓ BARIKAT SYNDROME is a very rare disease and an uncommon etiology of hypocalcemia. Also known as HDR syndrome, it is an autosomal dominant disorder characterized by hypoparathyroidism, sensorineural deafness, and renal disease.

✓ Mutations in GATA3, a gene localized in chromosome region 10p14-15, have been detected in families affected by the syndrome. GATA3 is a transcription factor that is involved in the embryonic development of parathyroid glands, kidneys, inner ears, thymus and central nervous findings.

CLINICAL CASE

1990 – 35 years old

• Admitted in Emergency department with generalized seizures and signs and symptoms of cardiac failure.

• Intracranial basal ganglia calcifications were revealed at this time and a diagnosis of HYPOCALCEMIC miocardiopathy was established.

• Started calcium replacement.

January 2014

• Hospitalized due to hypocalcemia (6.9 mg/dL) and acute heart failure.

May 2014 – 59 years old

• Admitted in the Endocrinology ward due to hypocalcemia despite being medicated with oral calcium (1 g/day of calcium carbonate and calcitriol 0.75 ug/day).

• She complained of tiredness, decreased muscle strength and unsteadiness when walking, for the last twelve months.

HOSPITALIZED

Initiated calcium carbonate 3 g/day + calcitriol 0.75 ug/dia

ABDOMINAL CT SCAN

Right kidney hypoplasia

Progressive HEARING LOSS over the last 20 years / Diagnosed with BILATERAL NEUROSENSORY DEAFNESS

LABORATORY WORK-UP:

<table>
<thead>
<tr>
<th>Item</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ca²⁺</td>
<td>7.7 mg/dL (8.4-10.2)</td>
</tr>
<tr>
<td>PTH</td>
<td>68 mg/L (100-300)</td>
</tr>
<tr>
<td>Mg²⁺</td>
<td>1.9 mg/dL (1.5-2.5)</td>
</tr>
</tbody>
</table>

DNA sequence analysis revealed on exon 5 of GATA3 gene a heterozygotic mutation c.1043T>C (p.Leu348Pro), that CONFIRMED the diagnosis of BARIKAT SYNDROME.

FAMILY HISTORY

We observed one of the two adult sons and the adult daughter of this patient:

- 38 years old, paraplegic due to a work accident. Bilateral deafness since infancy.
  - Ca²⁺ 7.4 mg/dL (8.4-10.2); PTH 11 pg/mL (11-80). Renal echography: Normal.

- 21 years old, bilateral deafness diagnosed at 15 years old.
  - Ca²⁺ 8.6 mg/dL (8.8-10.8); PTH 19 pg/mL (12-88)

The same mutation was identified in her children - c.1043T>C(p.Leu348Pro) on exon 5 of GATA3

COMMENTS

✓ Bariakat syndrome may present a variable phenotype. Renal manifestations are the most heterogeneous and usually determine disease prognosis.

✓ This patient has the classical triad. The severity of hypocalcemia since young age and subsequent irreversible cardiac involvement were determinant for diagnosis.

✓ Timely diagnosis and appropriate hypocalcemia treatment are paramount. Genetic screening of relatives takes particular relevance in this context.

BIBLIOGRAPHY