Clinical evolution of a patient with isolated growth hormone deficiency type IA treated with rIGF1 for 5 years after the development of GH-antibodies

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INTRODUCTION and OBJECTIVE

The main causes of isolated growth hormone deficiency are shown below.

Isolated growth hormone deficiency (IGHD)

<table>
<thead>
<tr>
<th>Type</th>
<th>Endogenous GH</th>
<th>Heritage pattern</th>
<th>Genes implicated (chromosome)</th>
<th>Other characteristics</th>
</tr>
</thead>
<tbody>
<tr>
<td>IA</td>
<td>Absent</td>
<td>AR</td>
<td>GH1 (17q22-24)</td>
<td>- Neonatal hypoglycaemia</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td>- Parental consanguinity</td>
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<td></td>
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<td></td>
<td>- Variable presence of GH antibodies at the start of the treatment</td>
</tr>
<tr>
<td>IB</td>
<td>Low</td>
<td>AR</td>
<td>GH1 (17q22-24)</td>
<td>- Normal response to GH stimulation tests</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td>GHRH-R (7p14)</td>
<td>- Good response to exogenous GH based treatment.</td>
</tr>
<tr>
<td>II</td>
<td>Low</td>
<td>AD</td>
<td>GH1 (17q22-24)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>- Hypogammaglobulinemia</td>
</tr>
</tbody>
</table>

Isolated Growth Hormone deficiency type IA causes a severe growth retardation. Their initial good response to exogenous GH is hampered by the development of anti-GH-antibodies leading to treat with IGF1 as the only therapeutic option. Here we present the evolution of a patient with IGHD type IA treated with IGF1r for more than 5 years.

CLINICAL CASE

Personal background

5-year-old patient from Pakistan
Normal pregnancy and birth: RNAT (unknown PN and TN)
Normal psychomotor development
No pathologic background

Family background

Consanguineous parents (close cousins)
Mother height: 158 cm / Father height: 168 cm
3 brothers with a normal height. Healthy.

Good initial response to GH: VC. 7.74 cm/year (+1.5 SD)
After 6 months of treatment with GH, response decreases: VC 4cm/year (- 2.1 SD)

Genetic study

Absence of the GH gene in homozygosis (ENSG000000189162) by joint amplification of the GH1 and GH2 genes and digestion with BamHI, as well as the methodology of Vnenc-KJones

Study of Antibodies anti-GH

Positive by using RRA technique in dilution 1:10000, with a 35% inhibition

Tests performed at the start of the treatment with rIGF1 and during the follow up:
- ECG, Echocardiography
- Abdominal ultrasound
- Fundoscopy and audiometry
- Carotid ultrasound: Increased systemic arterial rigidity (p75)

Final diagnosis:
Type IA IGHD with formation of Ab anti-GH

CONCLUSIONS

- Treatment with recombinant IGF-1 for 5 years has shown good results without adverse reactions, in a patient with IGHD type IA, with GH-antibodies.
- In our patient we, could detect a significant increase in BMI possibly related to the treatment that was controlled with dietary support.