The tricho-rhino-phalangeal syndrome type 1 (TRPS 1) also known as Langer-Giedion syndrome, is an uncommon genetic disorder. This disease is characterized by short stature, cone-shaped ends of the long bones (epiphyses) and distinctive facial features linked to skeletal abnormalities in early childhood. The experience about recombinant growth hormone (rGH) therapy in TRPS 1 is rather poor.

**INTRODUCTION**

The tricho-rhino-phalangeal syndrome type 1 (TRPS 1) also known as Langer-Giedion syndrome, is an uncommon genetic disorder. This disease is characterized by short stature, cone-shaped ends of the long bones (epiphyses) and distinctive facial features linked to skeletal abnormalities in early childhood. The experience about recombinant growth hormone (rGH) therapy in TRPS 1 is rather poor.

**AIM**

The aim of the study is to present the results of rGH therapy in a 14-year-old boy with TRPS1, coexisting with growth hormone deficiency and osteoporosis.

**METHOD**

This is a case report of a patient diagnosed with TRPS1. We made us of the available literature in the Medline database. The height standard deviation score (htSDS) for chronological age was calculated using Polish references [1]. Bone age was estimated due to Greulich and Pyle method. Height predictions were estimated due to Bayley- Pinneau method.

**RESULTS**

The boy was born at 37th HBD, with a birth weight 2890g. Target height based on parental height was 174 cm. He presented delayed motor development in the first moths of life. Since the age of 2 y, the height was classified below the 3rd centile.

At the age of 4y, because of distinctive facial dysmorphism such as round face, hypertelorism, wide eyebrows, bulbous nose, long philtrum, thin upper lip, low-set posteriorly rotated ears, hypertrichosis and short stature TRPS 1 was suspected. Molecular testing revealed submicroscopic deletions localized on the long arm of chromosome 8 (region 8q23.1-q23.3), which involved TRPS1 gene, confirming the diagnosis.

Due to short stature and deceleration of growth velocity at the age of 3y, GH secretion testing was performed. After clonidine and arginine stimulation, the peak GH level was 5.7 ng/ml and 2.1 ng/ml, respectively. GH deficiency was diagnosed. IGF-1 level was also below the range for age and sex (32 ng/ml). MRI of the pituitary was normal. rGH treatment was started at the age of 5.5y, when his height was 101cm (htSDS = -3 SD). At the age 6y, he had a fracture of the clavicle and elbow bone. Low mineral density was observed in densitometry (Z-score of lumbar region -2.3). Vitamin D3 and calcium oral supplementations were initiated.

Currently, at the age of 14y, his height reached 155.5 cm (htSDS = -1.4 SD) and the predicted adult height (PAH) is 180 cm. The current pubertal stage is as follows: A3, P3, and testicular volume 15 ml. No side effects of GH treatment were observed to date.

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Normal (males)</th>
<th>Normal (females)</th>
<th>Observed (rGH)</th>
<th>Expected (rGH)</th>
</tr>
</thead>
<tbody>
<tr>
<td>htSDS</td>
<td>0.8–1.9</td>
<td>0.7–1.6</td>
<td>0.8</td>
<td>1.7</td>
</tr>
<tr>
<td>Bone age</td>
<td>10–15</td>
<td>10–15</td>
<td>12</td>
<td>14</td>
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<tr>
<td>Bone p.a.</td>
<td>20–30</td>
<td>20–30</td>
<td>22</td>
<td>22</td>
</tr>
<tr>
<td>PAH</td>
<td>200–210</td>
<td>190–200</td>
<td>198</td>
<td>174</td>
</tr>
</tbody>
</table>

**CONCLUSIONS**

Almost nine-year observation of rGH therapy confirms efficacy of the treatment in TRPS 1. Increase of growth velocity and early age of the patient give a satisfactory prognosis for the further treatment.

**REFERENCES**


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