DOES THE TREATMENT WITH RECOMBINANT HUMAN GROWTH HORMONE IMPROVES FINAL HEIGHT IN CHILDREN AFFECTED BY X-LINKED HYPOPHOSPHATEMIA?

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**BACKGROUND / AIM**

- X-linked hypophosphatemia (XLH) is a rare disease caused by mutations in PHEX, leading to elevated FGF23 levels, hypophosphatemia and chronic renal phosphate wasting.
- Clinically, children affected with XLH manifest leg deformities, poor growth with short stature (Figure 1), dental abscesses, hearing loss, craniosynostosis.
- Despite optimal conventional treatment (oral phosphate supplementation and active vitamin D), 25 - 40% of patients with well-controlled XLH show linear growth failure with final height ~2 SDS (1-2).
- Recombinant human growth hormone (rhGH) may be an adjuvant treatment of the growth retardation in these patients (1-2).
- **Aim of the present study** is to describe whether rhGH treatment improves final height in children with XLH.

**RESULTS**

1. **General characteristics of patients**

<table>
<thead>
<tr>
<th>Parameters</th>
<th>Absolute number or % (n) or Mean±SD</th>
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<tbody>
<tr>
<td>Number of subjects</td>
<td>34</td>
</tr>
<tr>
<td>Boys / girls</td>
<td>38 % (13) / 62 % (21)</td>
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<td>Subjects carrying a PHEX-mutation</td>
<td>85 % (29)</td>
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<td>rhGH duration (years)</td>
<td>3.4±2.9</td>
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<tr>
<td>Follow-up duration (years)</td>
<td>4.5±3.1</td>
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<tr>
<td>rhGH dose at initiation (µg/kg/d)</td>
<td>77.4±14.5</td>
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<td>Age of diagnosis of XLH (years)</td>
<td>3.4±1.4</td>
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<tr>
<td>Age before rhGH (years)</td>
<td>9.8±3.5</td>
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<td>Age at 2 years after rhGH (years)</td>
<td>11.9±3.4</td>
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<td>Age at the end rhGH (years)</td>
<td>14.2±3.1</td>
</tr>
<tr>
<td>Age at the last visit (years)</td>
<td>19.2±3.4</td>
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</tbody>
</table>

- 34 patients (13 male / 21 female) were included.
- Mean age at start of rhGH treatment was 9.8±3.5 years.
- Duration of rhGH treatment and follow up were 3.4±2.9 and 4.5±3.1 years, respectively.
- The last visit was performed at 19.2±3.4 years.
- Mean doses of rhGH at initiation and the end of treatment were 77.4±14.5 and 66.8±20.5 µg/kg/day, respectively.

2. **Main results**

- The average height of patients significantly increased from -2.4±0.9 SDS to -1.3±0.9 SDS after 3 years of rhGH treatment.
- After 3.4 years of rhGH treatment, height at discontinuation was -1.2±0.9 SDS, remaining stable thereafter and resulting in final height -1.3±0.9 SDS.
- The global height increment during rhGH treatment was 1.2±0.7 SDS.

**CONCLUSION**

- Treatment with rhGH significantly increases height in XLH children with growth failure despite optimal conventional treatment, in this cohort of 34 patients.
- The major height gain is obtained during the first 2 years of rhGH treatment and is sustained till the final height, despite treatment interruption.

**References:**
1. Linglart A et al. Endocrine Connections 2014
2. Rothenbuhler A et al. Growth Horm and IGF Research 2017

**PATIENTS / METHODS**

- Retrospective observational study
  1/ Inclusion criteria: XLH children treated during childhood with oral phosphate supplementation and active vitamin D who received rhGH for at least one year reached their final height born after 1980
  2/ Anthropometric parameters

**Figure 1**