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

Authors: Julia André[1], Volha V. Zhukouskaya[1,2], Anya Rothenbuhler[1,3] , Anne-Sophie Lambert[1,3], Jean-Pierre Salles[5] , Brigitte Mignot[6] , Agnès Linglart[1,3,4].

Affiliations: [1]APHP, Reference Center for Rare Disorders of the Calcium and Phosphate Metabolism, Filiere OSCAR and Platform of expertise for rare diseases Paris-Sud, Bicêtre Paris-Sud Hospital, Le Kremlin Bicêtre, France; [2]Department of Clinical Medicine and Surgery, Division of Endocrinology, University of Naples Federico II, Naples, Italy; [3]APHP, Department of Endocrinology and Diabetology for children, Bicêtre Paris Sud Hospital, Le Kremlin- Bicêtre, France; [4]Paris Sud – Paris Saclay University, Faculté de Médecine, Le Kremlin- Bicêtre, France; [5]Toulouse, CHU Purpan, Department of Endocrinology ; [6]Besançon, CHU of Besancon, Department of Pediatrics

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 abcesses, 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)

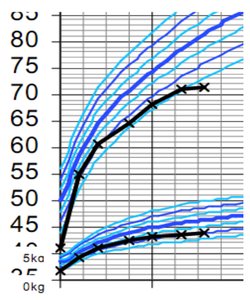


Figure 1

- ✓ **Aim of the present study** is to describe whether rhGH treatment improves final height in children with XLH.

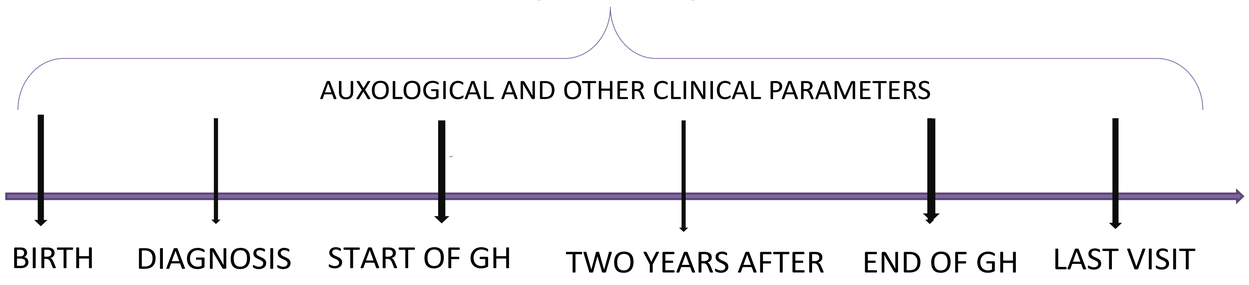
PATIENTS / METHODS

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



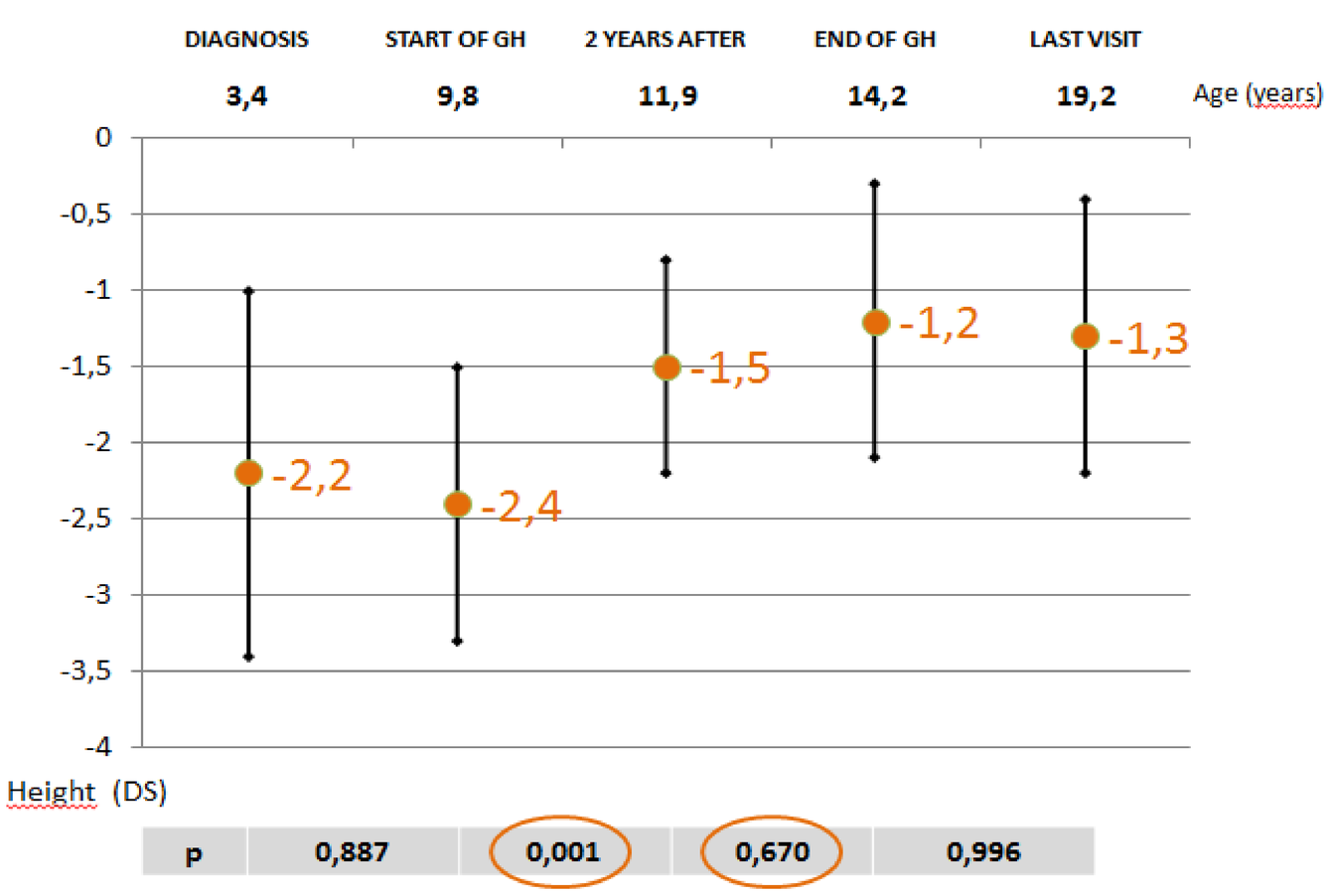
RESULTS

1. General caracteristics of patients

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

- ✓ 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



p	0,887	0,001	0,670	0,996

- ✓ The average height of patients significantly increased from -2.4±0.9 SDS to -1.5±0.7 SDS (p<0.001) after 2 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 Research2017

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Julia André
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