LONG-TERM EFFICACY AND SAFETY OF RGH IN CHILDREN WITH SHOX DEFICIENCY:
PRELIMINARY DATA OF A NATIONAL ITALIAN SURVEY.

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INTRODUCTION
The phenotypic spectrum of short stature homeobox-containing gene deficiency disorders (SHOX-D) ranges from non-specific short stature to Leri-Weill dyschondrosteosis. Current guidelines support recombinant human Growth Hormone (rhGH) in SHOX-D children1,2, but long-term data are still lacking3. Moreover, no correlation has been established yet between the severity of phenotype, including the response to rhGH, and the underlying SHOX pathogenic variant4.

AIMS
1) To evaluate long-term efficacy and safety of rhGH
2) To identify potential predictive factors influencing response to rhGH

RESULTS
(a) Baseline (T0) features in enrolled patients (n=86):

(b) Longitudinal data (mean rhGH duration 5.94±2.16 years):

(c) Correlations between mean H-SDS gain at T5 and GV-SDS in the first two years of treatment:

GV SDS (T0 -1.27±1.84, T1 2.39±1.37, T2 1.44±1.91, T3 0.95±1.87, T4 0.77±2.41, X2 56.65, p<0.01) and S/H ratio (T0 0.56±0.02, T1 0.55±0.02, T2 0.54±0.01, T3 0.54±0.01, T4 0.53±0.01, X2 15.77, p<0.01) improved significantly along rhGH. Mean H-SDS gain was: T4 vs. T0 +1.18±0.49 and T5 vs. T0 +0.68±0.89

(d) No adverse effects were reported a part from transient impaired glucose metabolism (2/86 cases) and transient headache (1 case).

(e) No differences in clinical and therapeutic data were detected between patients carrying mutations involving enhancers (5/86) and ones with no-sense and missense mutations in SHOX gene, both at the beginning of rhGH and along follow-up.

CONCLUSIONS
Our preliminary data confirm the efficacy and safety of rhGH in SHOX-D children. Besides wide genotype spectrum, all SHOX-D genotypes seem to adequately respond to rhGH

REFERENCES

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