GH therapy in Lery Weill Syndrome: report of three cases

E. Guidoni, M. Franceschini, R. Scarinci and G. Municchi

Department of Molecular Medicine and Development
University of Siena

Background:

SHOX deficiency is a frequent cause of short stature. Growth hormone (GH) therapy has been approved for growth promotion in individuals with *SHOX* mutations by FDA and EMEA.

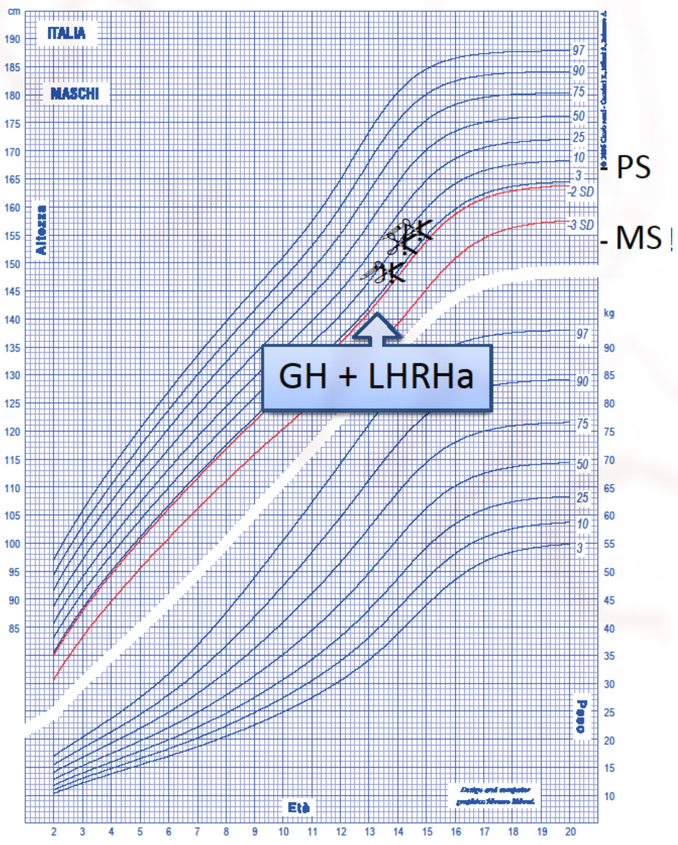
Case Report:

We present three patiens with Leri-Weill Syndrome (LWS) who have started GH therapy at different chronological ages (CA).

Patient 1

Patient 1 was referred for short stature at the CA of 13.4 yrs: height was 148.4 cm (-1.5 SD), Tanner stage 4, bone age (BA) 13 yrs. Target height (TH) was 160.5 cm (-2.5 SD), the mother was affected by LWS and her stature was 140.7 cm (-3.7 SD). He started GH treatment associated with LHRH analogue (LHRHa). After 3 months of combined therapy height was 151.7 (-1.3 SD), after 6 months height was 153.4 cm (-1.2 SD). After 1 year on LHRHa + GH therapy, height was 156 cm (-1.3 SD), BA 13.6 yrs. (Figure 1)

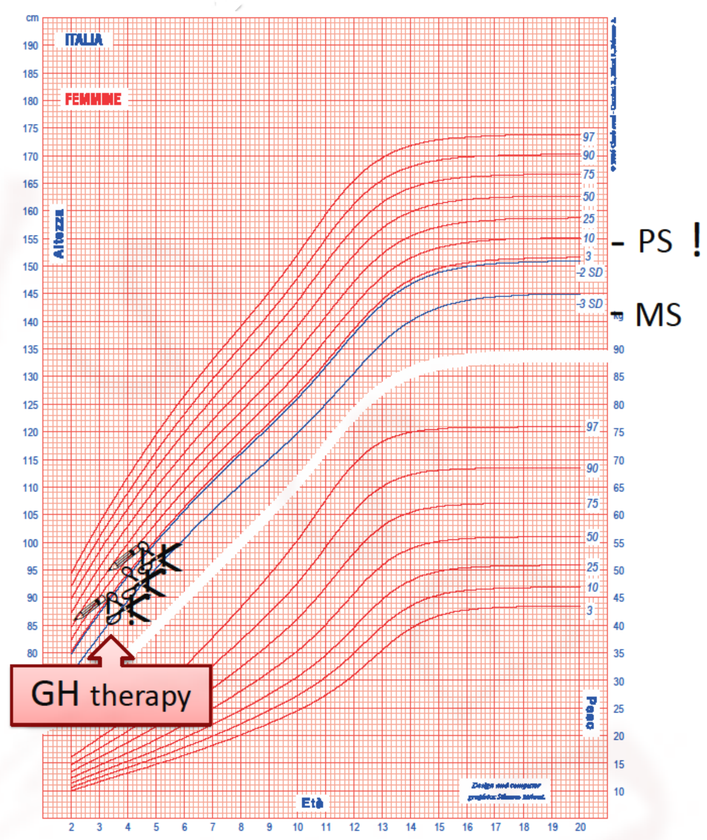
Figure 1



Patient 2

His sister, patient 2, was referred for short stature at the CA of 3.5 yrs: height was 88 cm (-2.6 SD), BA 2.5 yrs. TH was 147.5 cm (-2.6 SD). She started GH therapy. After 3 months on GH therapy the height was 90.7 cm (-2.3 SD) and after 6 months 92.8 cm (-2.2 SD). After 1 year on GH therapy the height was 97 cm (-2.1 SD), BA 3.6 yrs. (Figure 2)

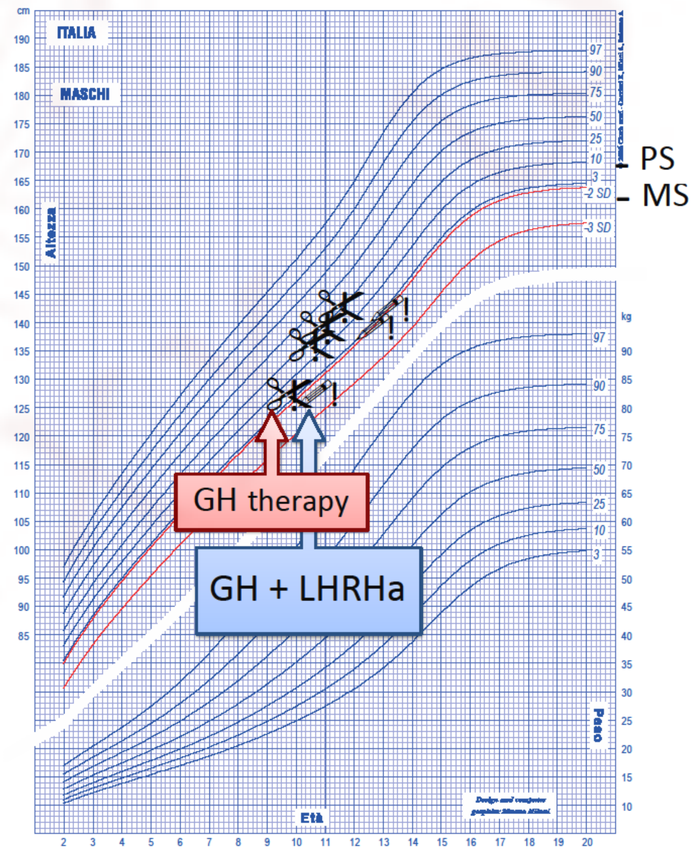
Figure 2



Patient 3

Patient 3 was referred for short stature at the CA of 9.3 yrs, prepubertal, height 127.4 cm (-1.2 SD), BA 10.5 yrs. TH was 164.5 cm (-1.8 SD). His brother (18 yrs) with a final stature of 150 cm presented the same mutation. Patient started GH therapy at the CA of 9.5 yrs. After 6 months height was 136 cm (-0.5 SD). After 10 months height was 139 cm (25°-50°ct), Tanner stage 2, BA 12.5 yrs. Due to the accelerated skeletal maturation LHRHa was associated. After 6 months of combined therapy height was 143 cm (50°ct), but BA was 13 yrs.

Figure 3



Conclusions:

GH therapy significantly improves growth rate and final height in children with SHOX deficiency. Despite the small size of our sample, our results confirm that height gain is higher in children who start GH therapy early.

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Fat 2
Elisa Guidoni
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