GH treatment in oto-spondylo-megaepiphyseal dysplasia: a case report

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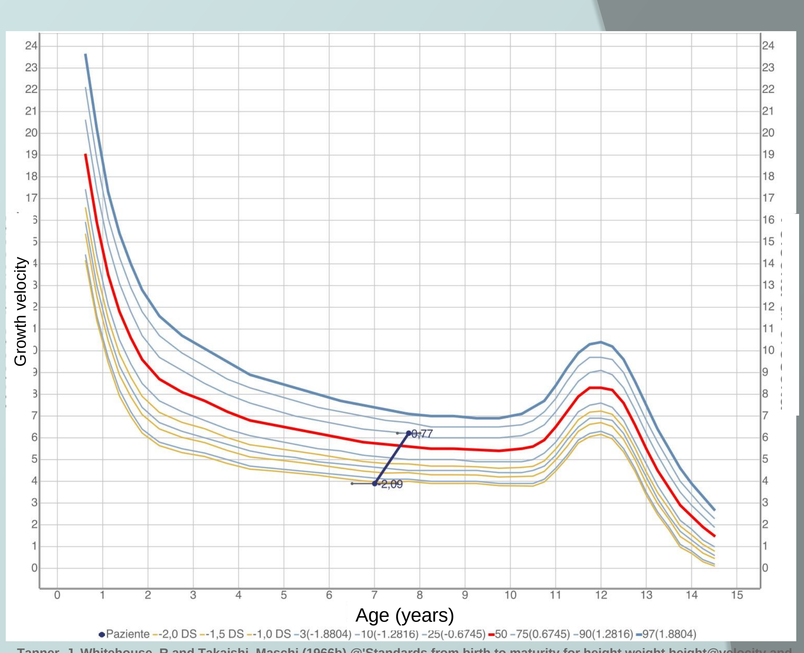
Introduction

The oto-spondylo-megaepiphyseal dysplasia (OSMED) is a rare condition with **autosomal recessive inheritance caused by congenital defect in the formation of cartilage collagen**.
OSMED is caused by mutations in the COL11A2 gene, which encodes the alpha2 chain of XI type collagen, a complex molecule that gives structure and strength to the connective tissues that support the body's joints and organs.
OSMED is characterized by **severe sensorineural hearing loss and skeletal abnormalities with distinctive facial dysmorphisms, enlargement of the epiphysis, disproportionately short limbs and platyspondyly**. Patients often experience limited joint movement, back and joint pain and arthritis beginning early in life. The prevalence is unknown.
Up to now only a few families with OSMED worldwide have been described in the literature. We describe the case of a girl affected by OSMED and growth hormone (GH) deficiency.

Case report

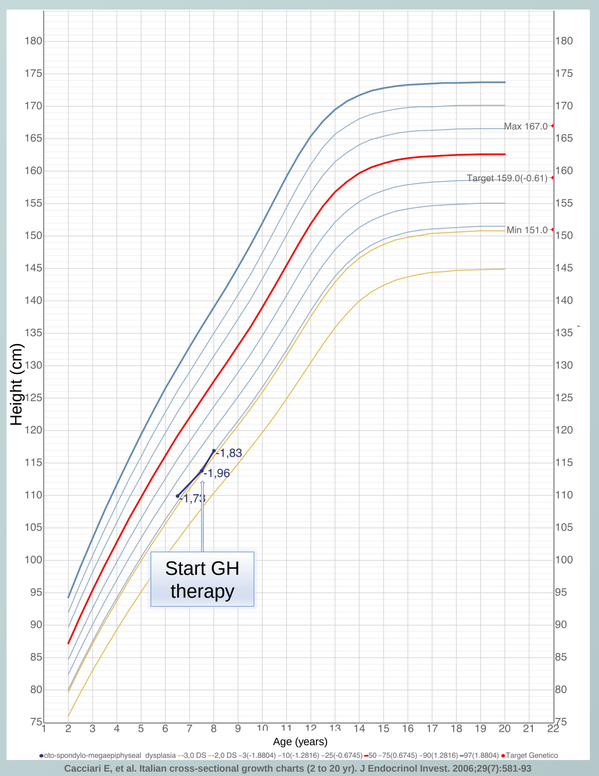
A 6 years old girl was initially referred to our department for evaluation of sensorineural deafness, language retardation and facial dysmorphisms, characterized by protruding eyes and upturned nose with flattened bridge, large and rounded tip. Genetic evaluation showed homozygous mutations in the COL11A2 gene (6p21.3), leading to the diagnosis of OSMED performed at the chronological age (CA) of 7.5 years.
Duo to the presence of short stature (-2.0 SD), with target height of 159 cm (-0.6 SD), after excluding other identifiable causes of short stature, we evaluated growth hormone (GH) secretion, spite bone age being advanced about 1 year compared to her CA.
We performed 2 pharmacologic stimulation tests that confirmed GH deficiency.
The brain MRI was normal and therefore a diagnosis of idiopathic GH deficiency was made.
The patient started **GH replacement therapy** and after 6 months on treatment her growth velocity markedly improved from 4cm/years to 6cm/years, even thought calculated over a period of only 6 months. No adverse events were reported. Figure 1-2

Figure 1



Tanner, J, Whitehouse, R and Takaishi, Maschi (1966b).@'Standards from birth to maturity for height weight height@velocity and weight velocity: British children 1965 Part II.' Arch Dis Child 41: 613-35.

Figure 2



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Conclusion

To our knowledge, this is the **first case of OSMED who presents also GH deficiency and is being treated with GH replacement therapy** reported in the literature.
In contrast to what generally happens in GH deficiency, bone age in our patient was not retarded compared to CA, but we believe that this phenomenon could be due to the syndrome itself. Only the long-term follow-up of this patient will show whether the beneficial effect of GH treatment on growth velocity observed during the first 6 months will be confirmed, without the occurrence of adverse events.

References

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Growth and syndromes (to include Turner syndrome)

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