Linear Growth in a Child with Ellis Van Creveld Syndrome: Positive Effect of growth hormone therapy.

Ashraf Soliman, Fawzia Alyafei #

Alexandria University Children's Hospital, Egypt, #Department of Pediatrics, Hamad General Hospital, P O Box 3050, Doha, Qatar

Introduction

Ellis van Creveld syndrome (EVC) is a rare, multisystem disorder caused by mutations in EVC or EVC2 genes. It is characterized by severe short stature, (1) long narrow trunk and shortened arms and legs; extra fingers (postaxial polydactyly), and abnormalities of the oral region and teeth. Children with EVC may have growth hormone deficiency (GHD). Little is known on GH levels and GH treatment in patients with EVC.

Case Presentation

This boy was born at term by lower segment CS due to transverse presentation. His length = 41 cm and weight = 2.2 kg, head circumference = 33 cm.

He had features of EVC syndrome including: rhizomalic short stature, post-axial polydactyly in hands and feet, (fig 1) multiple frenulae in the mouth, hypoplastic nails, congenital heart disease (small PDA) , long filtrum, and thick upper lips.

Growth and Endocrine Assessment:

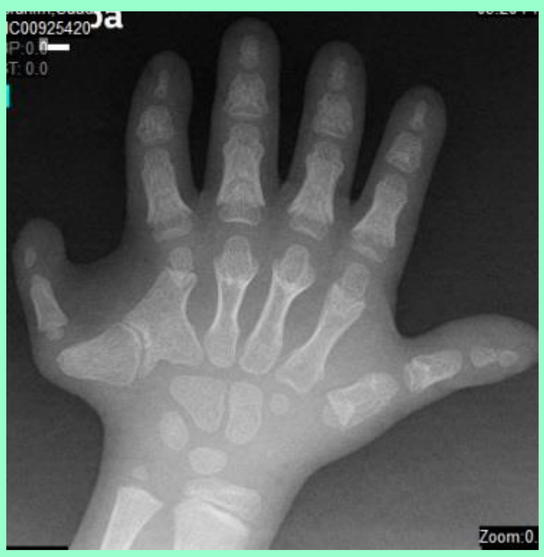
His growth chart (fig 2) showed the followings:

1. an initial spontaneous catch up during the first year of life
2. Followed by progressive deceleration of growth till the age of 6 years (htSDS = -3.6) with normal BMI (= 16 kg/m2).

Investigation showed: normal echocardiographic evaluation, normal renal and hepatic functions, and normal hemogram.

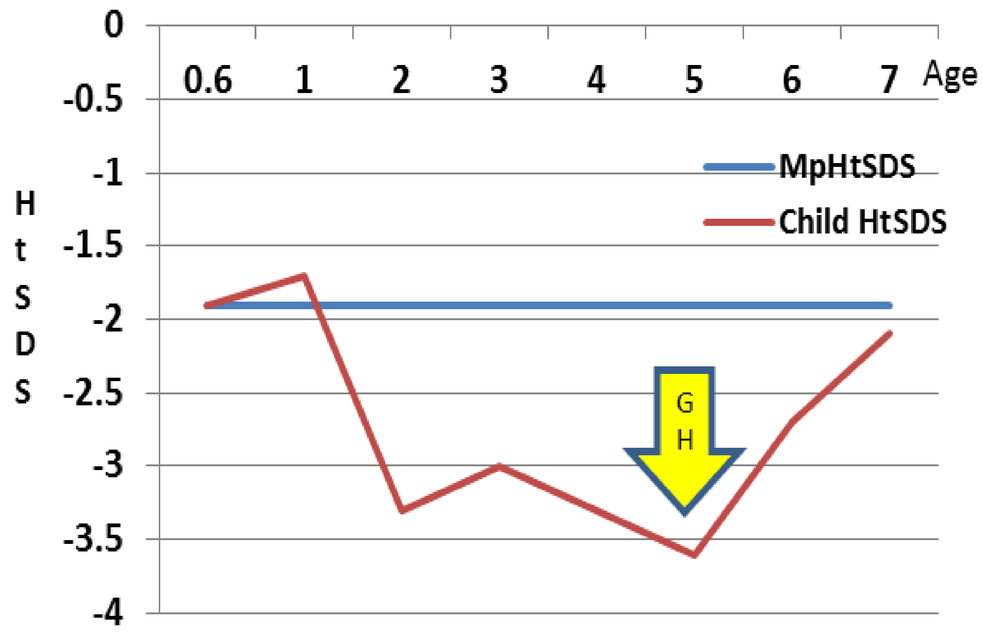
Hormonal evaluation :

Normal thyroid function,
Normal 8 AM cortisol concentrations
IGF-I = 78 ng/ml (IGF-I SDS = -1.7)
Clonidine stimulation test for growth hormone (GH) release showed a peak of 18 ug/L.



Treatment

3. Human GH therapy was started and improved his linear growth (0.05 mg/kg) (HtSDS) increased from -3.6 to -2.2) after two years of treatment. (fig 3)



Discussion

Literature review showed that EVC patients may benefit from being tested for GHD and, if indicated, treated. (2,3)
This is the first case of EVC with normal GH response to provocation that responded well to GH therapy. Previously, the association of growth hormone deficiency and ECV has been reported in one patient and, in this case, the growth hormone treatment had a favourable effect on growth.

Conclusion

It appears that growth hormone therapy can improve linear growth in GH sufficient patients with EVC syndrome.

References

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Ashraf Soliman
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