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STAT5B Mutation; GH Insensitivity with Severe Growth Retardation and other Medical Problems

Ranya Abuhlegas*

Endocrinology Department, United Arab Emirates

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***Corresponding author:** Ranya Abuhlega, Endocrinology Department, Shaikh Khalifa Medical City, Abu Dhabi, United Arab Emirates

Introduction

Severe primary IGF deficiency (IGFD) is rare. Patients with severe primary IGFD typically present with normal to high GH levels, height below 3 standard deviations (SD), and IGF-1 levels below 3 SD. Severe primary IGFD includes patients with mutations in the GH receptor, post-receptor mutations or IGF mutations. As a result, these patients cannot be expected to respond to GH treatment.

Case Presentation

A 5 year old female presented with severe growth retardation. She was born at 25 weeks gestation, birth weight 650g. She had severe failure to thrive with swallowing difficulties and delayed gross motor milestones. She developed recurrent lower respiratory tract infections from the age of 2 years and was found to have T cell lymphopenia immunodeficiency, asthma and eczema. Levothyroxine was started at 4 years for autoimmune hypothyroidism. She has two siblings with similar features. She was very small and thin (height 80.4cm (-6.8 SDS), MPH=152cm, weight 9.3kg (-7.5 SDS)), with depressed nasal bridge with a prominent forehead and mandible with hypertrichosis.

Investigations revealed low IGF1 3nmol/L and high random GH level 9.5mIU/L. Bone age delayed by 2 years. Genetic testing identified a novel STAT5B c.1892G>A mutation, confirming GH insensitivity. She was started on Mecaseramin (Increlex) 0.9mg SC BID and will be followed up to assess height response and IGF1 level regularly. She will need regular immunology review.

Conclusion

Primary IGF deficiency disorders are rare but are important to recognize as some may be associated with other complex non-endocrine medical problems.

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