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Short Stature; Not Always Straight Forward; Clinical Cases Scenarios

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Abstract

Short stature is the commonest cause for referral to paediatric endocrinology clinic. Although the etiology can be clear on initial assessment, many children present with short stature that can be a challenge to diagnose the cause for. General blood tests for chronic diseases are important as some pathologies can have no or non-specific symptoms. An example of those is celiac disease. Getting a karyotype in a short girl (particularly if puberty is delayed) is necessary to exclude Turner syndrome. Bone age assessment is useful but there can be variation between different interpreters.

IGF1 is a helpful test to assess growth. However, its wide reference range, the need for age and gender specific ranges make it

difficult to interpret. Growth hormone stimulation tests are labor-intensive and can be unreproducible. Cut off for normal values vary and there are complicating issues about which stimulant to be used and the application of priming rules. Some children have poor growth velocity but might have normal response to growth hormone stimulation. In this group of children, growth hormone profile can be helpful. In the case of low IGF1, IGF1 generation test can be useful prior to committing to an expensive long-term treatment with IGF1 therapy. In this talk, different complex case scenarios will be discussed to highlight the difficulty in confirming the etiology of short stature in some short children.



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