Pituitary stalk interruption syndrome: diagnostic delay and sensitivity of the auxological criteria of the growth hormone research society

OBJECTIVES: To study the diagnostic delay for pituitary stalk interruption syndrome (PSIS) with growth hormone deficiency (GHD) and the sensitivity of the auxological criteria of the Growth Hormone Research Society (GHRS) consensus guidelines. METHODS: A single-center retrospective case-cohort study covering records from January 2000 through December 2007 evaluated the performance of each GHRS auxological criterion for patients with GHD and PSIS. Diagnostic delay was calculated as the difference between the age at which the earliest GHRS criterion could have been observed and the age at diagnosis of PSIS with GHD. A diagnostic delay exceeding one year was defined as late diagnosis. RESULTS: The study included 21 patients, 16 (76%) of whom had isolated GHD and 5 (24%) multiple pituitary hormone deficiencies. The median age at diagnosis was 3.6 years (interquartile range, IQR, 2.6-5.5). The median diagnostic delay was 2.3 years (range 0-12.6; IQR 1.5-3.6), with late diagnosis for 17 patients (81%). Height more than 1.5 SDS below target height was the most effective criterion: 90% of the patients met the criterion before diagnosis at a median age of 1 year, and it was the first criterion to be fulfilled for 84%. CONCLUSION: In our cohort, the delay for diagnosis of PSIS with GHD was long and could have been reduced by using the GHRS criteria, in particular, height more than 1.5 SDS below the target height. The specificity of such a strategy needs to be tested in healthy populations.
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