A RARE CASE OF DELETION IN 2q24.1: CLINICAL FEATURES AND RESPONSE TO GH HORMONE TREATMENT

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Chromosomal imbalances are often due to sub microscopic deletions or duplications not evidenced by conventional cytogenetic methods. CGH array can help in the diagnosis of severe short stature, associated with mental retardation and dysmorphisms.

We describe the clinical case of a 13.1-year-old girl, born at 35 weeks, from a triplets pregnancy. She was 127.5 cm (< -5 SDS), 33 kg (< -3 SDS); SPAN: 122 cm; PH2B2, bone age: 11 years; mild psychomotor delay, facial dysmorphism (malformed years with a low-set, microcephaly) and feet malformations (flexion deformities, broad halluces). Born SGA, with a growth velocity < -3 SDS, a severe short stature she was a candidate to GH treatment. She started GH at the dosage of 0.035 mg/kg/day with a significant improvement of growth velocity. She had FSH, LH, TSH, fT4: in the normal range; low IGF-1 levels: 139 ng/ml (n.v. for age: 183-850).

CGH array evidenced a microdeletion of chromosome 2 (2q24.1), interesting genes UPP2, CCDC148,CCDC148-AS1, partially gene PKP4. MRI of CNS and pituitary revealed a small hypophysis with an intrasellar arachnoid cyst. After 11 months of GH treatment she was 134.8 cm (-3SDS), 37 kg; PH3B2.

A few cases of deletion of 2q24 are reported in literature, and the association of low birth weight, growth delay, mental retardation, facial dysmorphism, cardiac malformations, feet and hands deformities is specific of this deletion. The mild phenotype of our patient could be explained by the small deletion (2q24.1). For this reason, it could be considered a continuous gene syndrome. At our knowledge this is the first case reported in the literature treated with GH and showing a satisfactory growth.

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