HAIR DISORDERS

A STRANGE CASE OF HYPERTRICHOSIS

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Background: Cantù syndrome, or osteodysplasia with hypertrichosis, is a rare genetic disorder featuring hypertrichosis, characteristic facial appearance, skeletal abnormalities and cardiomegaly. In most of cases it is caused by an autosomal dominant mutations of the ABCC9 gene, rarely by the KCNJ8 gene, but ex-novo mutations are also frequently reported.

Observation: We report on a 7 year old girl with congenital generalized hypertricosis and coarse facial appearance. Hypertrichosis was present since birth. Personal and maternal pharmacological anamnesis was negative for drugs correlated to iatrogenic hypertrichosis and endocrinological assessments excluded a hormonal cause. The child at birth was macrosomic, with patent ductus arteriosus, ostium secundum like interventricular defect and some hours after birth she experienced pulmonary hypertension. The patient had axial and pelvic girdle low muscle tone, resolved at the time of visit. On the basis of clinical manifestations and history we suspected the patient was affected by osteodysplasia with hypertrichosis. Sequencing of the ABCC9 gene showed c.3461G>A heterozygous mutation. The parents and sibs of the patient did not show any Cantù phenotype and genetic analysis was negative.

Key message: In patients with generalized hypertrichotic phenotype it is important to perform a thorough medical history and detect concomitant malformations. Dysmorphic facial appearance or cardiac defects can be important clues in order to suspect a genetic syndrome. In particular, Cantù syndrome is a rare genodermatosis which diagnosis is important because of its systemic implications.