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Acromegaloid Facial Appearance Syndrome

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Editorial

A rare multiple congenital anomalies/dysmorphic syndrome with a probable autosomal dominant inheritance, characterized by a progressively coarse acromegaloid-like facial appearance with thickening of the lips and intraoral mucosa, large and doughy hands. Acromegaloid facial appearance syndrome appears to be part of a phenotypic spectrum that includes hypertrichotic osteochondrodysplasia, Cantu type and hypertrichosis-acromegaloid facial appearance syndrome. Acromegaly is characterized by skin and soft tissue changes which are due to increased growth hormone levels.

Patients with similar physical findings but with an intact somatotroph axis are considered to have pseudoacromegaly. Another term used to describe this condition is acromegaloidism. These latter patients have normal serum IGF-1 levels and reveal a suppressed serum GH following an oral glucose challenge. Conditions responsible for pseudoacromegaly includes severe insulin resistance, multiple neuromas syndrome, pachydermoperiostitis, Ascher's syndrome, drug intake and hypothyroidism. Pseudoacromegaly due to severe insulin resistance is due to supraphysiologic levels of insulin which stimulate growth through an intact mitogenic signaling pathway.

The underlying etiology for excess soft tissue growth in other conditions is not known but it is probably due to the growth factors different from those of GH and IGF-1. Pseudoacromegaly is the syndrome referred to as acromegaloid facial appearance (AFA) and a variance of it, which includes terminal hypertrichosis. This syndrome has been reported so far in about eight cases/families that demonstrate an acromegaloid appearance. Patients with AFA have differing inheritance. The majority of cases seem to be inherited in an autosomal dominant pattern or autosomal dominant pattern with incomplete penetrance. A rare multiple congenital anomalies with a probable autosomal dominant inheritance which is characterized by a progressively coarse acromegaloid-like facial appearance with thickening of the lips and intraoral mucosa, large and doughy hands and, in some cases, developmental delay. AFA syndrome appears to be part of a phenotypic spectrum that includes hypertrichotic osteochondrodysplasia, Cantu type and hypertrichosis-acromegaloid facial appearance syndrome.

Acromegaloid Facial Appearance syndrome is a very rare syndrome combining acromegaloid-like facial appearance, thickened lips and oral mucosa and acral enlargement. Progressive facial dysmorphism is characterized by a coarse facies, a long bulbous nose, high-arched eyebrows, and thickening of the lips, furrowed tongue and narrow palpebral fissures, oral mucosa leading to exaggerated rugae and frenula. We report a case of acromegaloid facial appearance syndrome in a 19-year-old male patient who presented with all the characteristic features of the syndrome along with previously unreported anomalies like dystrophic nails, postaxial polydactyly and incisal notching of teeth. Acromegaloidism describes a highly heterogeneous group of disorders. When a patient presents with acromegaloid phenotype in early adulthood, growth hormone excess and insulin resistance associated pseudoacromegaly should be first ruled out by analyzing the endocrine profile of the patient. On exclusion of an abnormality of the somatotrophic axis, rare genetic syndromes associated with the AFA should be considered in the differential diagnosis.

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