GENETICS AND GENODERMATOSES

CARDIO-FACIO-CUTANEOUS SYNDROME : ABOUT A RARE CASE

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Background: Cardio-facio-cutaneous syndrome (CFC) is one of the RASopathies caused by gene mutations in the Ras/mitogen-activated protein kinase pathway. The major features of CFC include characteristic craniofacial dysmorphology, congenital heart disease, dermatologic abnormalities, growth retardation, and intellectual disability.

Observation: We report a case of a 12-year-old boy, followed in cardiology department of children's hospital since infancy for atrial septal defect with valvulopathy and hypertrophic cardiomyopathy with growth retardation. He consulted in dermatology for xerosis, eczema plaques and hyperkeratosis evolving for many years in relapses followed by periods of partial recovery. Clinical examination also revealed a curly hair scalp, sparse eyebrows, macrocephaly, tall forehead, bitemporal narrowing with hypertelorism, epicanthal folds and wide mouth. A CFC was suspected and then confirmed by genetic consultation and testing. Laboratory tests and radiological imaging were performed to evaluate the neurological, ophtalmological, renal, gastro-intestinal and endocrinological status of our patient. Multidisciplinary care and long-term follow-up were established.

Key message: CFC is a variable and genetically heterogeneous disorder caused by mutations of genes in the Ras/MAPK pathway. Like other RASopathies, recognizable facial features, congenital heart disease, and short stature characterize CFC. In addition, CFC also has cutaneous and neurologic involvement. It is essential that this condition be differentiated from other RASopathies, as a correct diagnosis is important for appropriate medical management, and determining long-term prognosis and recurrence risk. Because of the many aspects of CFC, multidisciplinary care is essential. The prognosis of CFC is still unknown, and full understanding of care expectations during childhood and adulthood is yet to be established.