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Classic Crouzon syndrome and Crouzon syndrome associated with Acanthosis nigricans: Familial case report

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Crouzon syndrome is an anomaly or congenital defect with an autosomal dominant pattern that is present due to the premature closure of the cranial sutures whose alterations will give as result, morphologic changes in the cranium and face. The mutation originates in the short arm of the chromosome 10 which would cause a random change in the gene FGFR2, in the case of association with acanthosis nigricans the mutation is present in the FGFR3 compromising the short arm of chromosome 4. In this clinical report, we analyze a family consisting of: a mother with CS (Case 2) and a healthy non-consanguineous father; As a result of this marriage, three children were conceived: a 9-year-old female patient with CSAN (Case 1), a 6-year-old female patient with CS (Case 3) and a 5-year-old male patient with CSAN (Case 4). Each patient's clinical characteristics are widely described. From the study of the last three generations of this family, it is determined that case 2, who presented CS without a family history or predisposing factors such as advanced paternal age, corresponded to a novo mutation, due to its variable expressiveness. This work highlights the recognition of these two pathologies within the same family lineage; these diseases have a different genetic origin with similar clinical characteristics, being the only family case according to the medical literature in Ecuador. Genetic counseling is recommended for all craniofacial abnormalities since the moment of birth. The therapeutic options of CS and CSAN should have a multidisciplinary approach; it requires the association of the pediatric service, neurosurgery, maxillofacial surgery, and a series of specialties that promote the physical and psychosocial well-being of patients.

Biography

Rebeca Briones is an MD graduate from Universidad Católica de Santiago de Guayaquil, Ecuador graduated in 2016, she is passionate on children's health and wellbeing. During her medical career she enjoyed teaching and was chosen to become a professor's assistant for 3 years, she was also granted a scholarship due to her academic achievements. Rebeca's experience in patient care and hospital work leads her to pursue her career pathway into pediatric research.

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