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A Missense Mutation in LMX1A in a Patient With Moebius Syndrome: A Case Report

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Moebius syndrome is characterized by congenital complete or partial paralysis of the facial nerve and is often associated with orofacial and limb malformations. It is a rare syndrome that affects the sixth and seventh cranial nerves. Facial paralysis results in abnormal abduction of one or both eyes and facial paralysis or weakness. Moebius syndrome is an uncommon condition and only a few hundred cases have been reported in the literature. A seven-year-old girl with Moebius syndrome is featured in this report. She had asymmetrical facial expressions, ocular abduction anomalies, and swallowing difficulties. She also had mild low-set ears, hypertelorism, a short nose, and restricted jaw movements. Array-comparative genomic hybridization analysis of exosome sequencing showed a mutation p.Gln61Arg in exon 3 of LMX1A.

Moebius syndrome (MBS), a rare congenital disease, was first described by Von Graefe in 1880 and then named after Moebius in 1888 [1]. It is characterized by unilateral or bilateral facial and abducens palsy, which leads to a loss of facial expressions, and an inability to smile or swallow. Moreover, patients with MBS also suffer from orofacial and limb defects. The estimated prevalence is 1/250,000, with an equal incidence in both genders. The condition affects one in 50,000 to one in 500,000 newborns. The unclear diagnostic criteria have led to difficulty in the clinical assessment, prognosis, and genetic analysis of patients with MBS. Since 1888, the pathogenesis of the disease has been unclear; till 2007 when MBS was defined as “congenital unilateral or bilateral, non-progressive facial weakness and limited abduction of the eye”.

Furthermore, de novo mutations in the PLXND1 and REV3L have been recorded in many MBS cases. PLXND1 plays a role in regulating the migration of a wide spectrum of cell types in the striatum and is selectively expressed in direct-pathway medium spiny neurons although REV3L functions in the translation of DNA synthesis, it can also protect DNA from damage.

Biography

Ghaliah O. Alnefaie is a board certified, a licensed marital and Pathology and associate professor of Department of Pathology, SAU, and currently teaching at Taif University Therapy at Pathology program. Main research foci include resilience, creative thinking, creativity, art therapy with children and adolescents, grief and loss, couples' work, and cultural considerations in therapy.