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Alwadei Syndrome - A Genetic Cause Of Intellectual Disability

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Abstract : Intellectual disability (ID) is characterized by deficits in intellectualfunctioningassociatedwithalterations in the adaptive behaviour, whose onset is inthedevelopmentalperiod. Itaffects 3% of the population, ofwhich 10% have a geneticaetiology. One of those causes isAlwadeiSyndrome, with 3 cases describedworldwide. It results from a homozygous nonsense mutation in theRUSC2 gene andisassociatedwithintellectualdisabilityanddysmorphic facialfeatures. Theauthorsreportthe case of a 5-year-old-boy, born to a healthymotherafter a full-termuneventfulpregnancy, thatwasreferred to Neurodevelopmentalconsultationdue toglobal developmentaldelay. Familyhistoryrevealedlearningdifficulties in the paternal brotherhood. Milddismorphicfeatureswereevidentsuch as darkinfraorbitalregion, low-set ears, beakednose, retrognathism, high-archedpalateandjointhyperlaxity. WechslerIntelligenceScale for Children III fullscaleIQ quoted 61. Karyotypeandchromosomalmicroarrayanalysiswerenormal, as well as the fragile X molecular study. DNA sequencingwasthenperformedandallowedtheidentificationof amutation in the RUSC2 gene. Theetiologicaldiagnosisof ID remains unknown in up to 80% of cases, creatinguncertainty in children'sfamilies. Theadvances in DNA sequencingtechnologieshaveincreasedourknowledgeofthegeneticdiseasesinvolved, as theAlwadeisyndromewasonlydescribedsince 2016. Thegeneticdiagnosisof ID allowsfamilygeneticcounselingandenablesthedevelopmentof target therapeutic approaches.

Keywords : intellectual disability, genetic aetiology, alwadei syndrome, RUSC2
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