DDX3X Syndrome, A Rare Genetic Disorder with New Clinical Manifestations: A Case Report

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ABSTRACT

DDX3X syndrome is a rare genetic disorder affecting females and present mainly with developmental delay and intellectual disability. This syndrome can also present with autistic features. This is a 5 years old girl with typical features of DDX3X syndrome associated with hypothyroidism and cyclical vomiting disorder, two different conditions which have not been mentioned in the literature before. The aim of this case report is to increase the awareness of pediatricians toward this genetic disorder in order to diagnose any associated condition as early as possible and formulate a planned intervention with a multidisciplinary team

Keywords: DDX3X Syndrome, Intellectual disability, Neurodevelopmental disorder

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