DDX3X Syndrome

https://pubmed.ncbi.nlm.nih.gov/36434914/

DDX3X syndrome is a surprisingly common newly discovered genetic neurodevelopmental disorder associated with intellectual disability, autism spectrum disorder, language delays, attention-deficit/hyperactivity disorder, and medical comorbidities. Two hundred individuals with DDX3X syndrome have been described in the literature to date, with varied levels of detail. Individuals with DDX3X syndrome often have complex presentations including symptoms in the neurological, psychiatric/psychological, ophthalmologic, and gastrointestinal domains. Owing to this complex presentation, an overview of symptom prevalence, medical recommendations, and suggested medical surveillance is vital for the care and health of individuals with DDX3X syndrome. In this article, we summarize the present clinical knowledge of DDX3X syndrome and provide recommendations for clinical assessments and care based on a comprehensive review of the existing literature and of new, not yet published DDX3X syndrome cohorts. As more is learned about DDX3X syndrome, we anticipate that these recommendations will evolve.