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Emotional-psychological concerns of Turner's patients regarding social discrimination

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Introduction: Turner syndrome characterized by total/partial and/or homogeneous/mosaic X chromosome monosomy is associated with various physical health concerns, including facial dysmorphism, short stature, infertility, and other organ defects such as heart, kidney, bone, skin, … as well as variable degrees of cognitive impairments. Besides, social skills, communication and relationships are usually disordered.
Objectives: The aim of this study was to report social challenges of Turner syndrome particularly related to discrimination regarding physical and cognitive impairments.
Methods: A retrospective analysis of clinic data and karyotypes were carried out for the patients diagnosed with Turner syndrome among patients who consulted at our genetic counselling at the medical University of Sfax, during the last two decades.
Cytogenetic analysis were carried out using conventional methods and RHG banding with analysis of at least 20 metaphases and 3 karyotypes for each patient. Social challenges were recorded for each patient during pre-cytogenetic consultation and oriented questioning.
Results: We identified 23 cases referred with a cytogenetic diagnosis of monsomy X. The karyotyping was indicated for dysmorphism, primary or secondary amenorrhea, female infertility and recurrent pregnancy losses. Homogeneous X chromosome monosomy was recorded in 13% of cases, whereas mosaic forms with and without structural X/Y abnormalities were more frequent (82%). The mean age of patients in the study was twenty years. When the 45,X population was the predominant one (56,5%), dysmorphism and primary amenorrhea were constant. In the mosaic forms, clinical traits of Turner syndrome were insignificant. Discrimination based on physical appearance, intellectual disability, and failure to conceive were the three types of social challenges revealed by patients of our study. Parents of Turner patients were also concerned at the psychological level. They in fact revealed their emotional distress face to stressful experiences of their children with Turner syndrome regarding the social discrimination they encountered particularly in schools.
Conclusions: Social challenges related to discrimination based on physical appearance, intellectual disability, and failure to conceive in Turner syndrome lead to depression, low self-esteem and anxiety.

Disclosure of Interest: None Declared

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16p13.11 microduplication including NDE1 gene in autism spectrum disorder: A case report

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Introduction: Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by the deficit in communication and social interaction as well as restricted and repetitive interests and behaviors. In addition to the involvement of the environmental component in ASD, it is currently established the significant contribution of genetic factors such us the 16p13.11 duplication. We report the case of a patient carrying this anomaly in whom an ASD was diagnosed, the genetic study of the parents objectified the same anomaly in a clinically healthy father.
Objectives: This case report aims to expand the clinical findings associated to this genomic abnormality and provide further knowledge of the pathogenic involvement of this duplication.
Methods: Our patient, aged 4 years and 10 months, presented developmental peculiarities from birth. The difficulties became more evident with the language delay, the deficit of social interactions and the appearance of motor stereotypies as well as sensory specificities. The diagnosis of ASD was confirmed by the passation of the ADI and the ADOS.
Further genetic exploration with a CGH-Array request was performed due to a normal 46 XY karyotype. She objectified a 16p13.11 duplication comprising 25 genes including NDE1. The assessment was completed by the search for potentially associated malformations, particularly cardiac and skeletal. We continued the family investigation with genetic samples for the parents and the siblings finding the same anomaly in the father who does not present any particular phenotype.
Results: The 16p13.11 duplication is associated with a variable clinical spectrum including behavioral disorders, attention deficit/hyperactivity disorder, intellectual disability, cardiac and skeletal malformation, epilepsy as well as ASD and language delay presented by our patient. Eight annotated genes are present in this region including NDE1, the candidate gene for the neurocognitive phenotype. This microduplication can be found in the normal population, but it is increasingly detected in patients with ASD, schizophrenia or presenting cognitive disorders due to incomplete penetrance that can explain the presence of the duplication despite the absence of any disorders in the patient's father.
Conclusions: The link between the 16p13.11 duplication and ASD is increasingly recognized in the literature. The heterogeneity of its clinical expression and especially its incomplete penetrating make genetic counseling difficult. Collaboration between child psychiatrists and geneticists remains essential to detect, link their clinical evaluation and optimize care.

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