Psychotic Features of Turner Syndrome

To the Editor: Turner syndrome is a genetic disorder characterized by partial or complete X chromosome monosomy, which occurs at birth in approximately 1 in 2,000 females.[1] Only a small number of studies[2] describe the psychopathologic features of Turner syndrome, with many girls exhibiting immaturity, poor self-esteem, and social relationship difficulties. Turner syndrome is 3 times more common among female schizophrenic patients,[3] and individuals with Turner syndrome are at an increased risk for autism spectrum disorders, attention-deficit/hyperactivity disorder, and intellectual disability.[2] The potential role of X chromosome genes (pseudoautosomal and nonpseudoautosomal regions) in schizophrenia pathogenesis has been investigated.[4,5] We report a case of a unspecified schizophrenia spectrum disorder (*DSM-5*) in a woman with Turner syndrome.

Case report. Ms A is a 24-year-old woman with Turner syndrome. She was diagnosed with persistent depressive disorder associated with anxious distress (*DSM-5*) at the age of 21 years and was prescribed escitalopram 20 mg/d, trazodone 50 mg/d, and mexazolam 1 mg/d. Additionally, she has a medical history of dyslipidemia, which was treated with atorvastatin, and a family history of major depressive disorder and suicide attempts.

In August 2017, Ms A was admitted to the neurology ward of a general hospital due to new-onset holocephalic headache with photophobia and right-sided half-body paresthesia. No focal signs were identified during the neurologic examination. The clinical picture was interpreted as a migraine with atypical aura. According to Ms A's family, symptomatology emerged after recent life stress events, including PhD interviews and a broken partnership.

During the first days after admission, the migraine remitted with symptomatic therapy. Physical and neurologic examinations and laboratory tests, including toxicology and serology, were unremarkable. Lumbar puncture, electroencephalogram, and cerebral magnetic resonance imaging showed no changes. Cerebrospinal fluid and serum anti–*N*-methyl-D-aspartate and anti–α-amino-3-hydroxy-5-methyl-4-isoxazolepropionic acid receptor antibodies were requested to exclude the remote possibility of autoimmune encephalitis.

Ten days later, Ms A presented with perplexity, psychomotor and speech retardation, disorganized behavior, labile mood, somatic anxiety, and psychotic symptoms, namely acoustic-verbal hallucinations and persecutory, reference, and misidentification delusions, with the patient identifying the hospital staff as family members. During a family interview with the liaison psychiatry team after the onset of psychotic symptomatology, her parents reported sporadic derealization episodes and reference delusions that started a few weeks before admission.

Ms A was transferred to the psychiatric ward and diagnosed with unspecified schizophrenia spectrum and other psychotic disorder (*DSM-5*) and was started on paliperidone 6 mg/d and bromazepam 3 mg/d. Psychotic symptomatology remitted completely in 10 days, and the patient was discharged.

This case report shares important features with the few reported cases of psychosis in Turner syndrome patients,[6] namely the presence of previous heterogeneous diagnoses, stress-precipitated onset, psychotic features that remit or respond well to antipsychotics, and labile mood and prominent anxiety symptoms. Contrary to some reported cases of Turner syndrome in which abnormalities in parietal lobe, amygdala, hippocampus, and orbito-frontal cortex were documented,[7] no changes were found on neuroanatomical imaging.

According to the available evidence, the prognosis of these psychotic episodes tends to be favorable, with a relatively benign course. However, given previous satisfactory premorbid functioning, family history of affective disorders and suicide attempts, and presence of previous depressive episodes together with this psychotic episode, a differential diagnosis of bipolar disorder must be considered.

On the other hand, the similarities between reported cases may lead us to hypothesize that the psychotic syndrome in Turner syndrome may be understood as distinct of other psychotic syndromes, with unique features. Further investigation is necessary to clarify the link between psychosis and Turner syndrome and to determine the potential unicity of this syndrome to avoid an erroneous and more serious diagnosis.

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