PP-011 https://doi.org/10.5080/61upk.ozt509

A CASE OF SMITH–MAGENIS SYNDROME FOLLOWED FOR YEARS AS CONDUCT DISORDER AND ATYPICAL PSYCHOTIC DISORDER AND DIAGNOSED IN ADULTHOOD

Mahmut Barış Koç, Melike Ceyhan Balcı Sengul, Selim Çiftcioğlu

Pamukkale University, Faculty of Medicine, Department of Psychiatry, Denizli, Türkiye

OBJECTIVE: Smith–Magenis syndrome (SMS) is a rare genetic disorder characterized by developmental delay, intellectual disability, distinctive dysmorphic facial features, behavioral dysregulation, and circadian rhythm disturbances. Due to its clinical overlap with attention-deficit/hyperactivity disorder, conduct disorder, autism spectrum disorder, and psychotic disorders, misdiagnosis is common and diagnosis is often delayed. We present a patient first evaluated at age 7 who received multiple psychiatric diagnoses throughout childhood and adulthood and was ultimately diagnosed with SMS at age 27, underscoring the importance of including SMS in psychiatric differential diagnosis, particularly in cases with longstanding behavioral and cognitive impairment.

CASE: After written informed consent was obtained from the patient and her relatives, history revealed referral to child and adolescent psychiatry at age 7 for learning difficulties, lagging behind peers, irritability, self- and hetero-aggression, and attention problems. She was managed for specific learning disorder, ADHD, and mild intellectual disability. Over time, anger outbursts, impulsivity, aggression, stereotypies, failure to achieve toilet training, and marked cognitive impairment became prominent. During childhood, multiple psychotropic treatments were administered. After age 18, she was followed in adult psychiatry with a diagnosis of atypical psychosis, and multiple antipsychotic treatments were administered for persistent behavioral and psychotic symptoms. Due to clinical worsening, she was hospitalized for reassessment and treatment adjustment.

Despite combined high-dose antipsychotic therapy, response was insufficient, and eight sessions of electroconvulsive therapy led to marked improvement. Physical examination revealed dysmorphic features, and phenotype-guided genetic testing confirmed SMS.

DISCUSSION: This case illustrates that SMS may remain unrecognized into adulthood in patients with chronic behavioral and psychotic symptoms, even after years of continuous psychiatric care. In early-onset behavioral problems accompanied by intellectual disability and sleep disturbance, careful assessment of dysmorphic features and a multidisciplinary diagnostic approach are essential for accurate diagnosis.

Keywords: Smith-Magenis syndrome, atypical psychosis, conduct disorder