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•Case report•

Behavioral and emotional manifestations in a child with Prader-Willi syndrome

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Summary: Prader-Willi syndrome is a neurodevelopmental disorder characterized by mental retardation and distinct physical, behavioral, and psychiatric features. Maladaptive behaviours, cognitive impairment, and impediments in speech and language seriously affect the early development and long-term functioning of individuals affected by the illness. We present a case of a 9-year-old child with Prader-Willi syndrome whose behavioural symptoms were treated with low-dose antipsychotic medications.

Keywords: Prader-Willi Syndrome; psychiatric symptoms; childhood disorders; case report; India

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1. Introduction

Prader-Willi syndrome (PWS) is a genetically determined neurodevelopmental disorder with a prevalence of 3 to 7 individuals per 100,000 births.[1] It is usually the result of a paternally transmitted deletion at chromosome 15-q11-q13. Characterized by mental retardation and distinct physical, behavioral, and psychiatric features, individuals with PWS are typically short and obese, have small hands and feet, and have other dysmorphic features including a narrow bifrontal diameter, full cheeks, and almond-shaped eyes.[2] They have borderline to moderate mental retardation, have impaired speech and language,[3] and exhibit more behavioral disturbances than individuals with other intellectual disabilities,[4] including excessive interest in food, skin picking, difficulty with changes in routine, temper tantrums, obsessive and compulsive behaviors, and mood fluctuations.[5,6] The severity of the behavioral problems increases with age and body mass index,[7] and then diminishes in older adults.[9] Recent evidence suggests that autism spectrum disorders (ASD) may be common in individuals with PWS.[9] Psychosis occurs during young adulthood in 5-10% of individuals with the syndrome.[10] The cognitive impairment, limited speech and language skills, and behavioral abnormalities seriously affect the early development and long-term functioning of individuals with PWS. Psychiatric and behavioral problems are the most common cause of hospitalization.

2. Case history

A 9-year-old girl was brought to our hospital with complaints of irritability, stubbornness, emotional lability, temper tantrums, and increased speech. Her father also reported hyperactivity, a history of over-eating and stealing food, and sudden mood changes including outbursts of laughter and crying without any obvious reason. She had had an uneventful birth history and no family history of neurological or psychiatric illness, but she had delayed development of gross motor functions and language skills. Her academic performance in primary school was poor.

Physical examination revealed an obese female (weight 54 kg, height 112 cm, body mass index 43.1) with small hands and feet, a narrow nasal bridge, and

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almond-shaped palpebral fissures. The skin on her face, hands, and arms had many excoriated papules from repetitive skin picking. Her speech had imprecise articulation and hypernasality. Her neurological examination, routine blood tests, thyroid function tests, liver function tests, and computed tomography (CT) of the brain were all within normal limits. Ultrasonography of her abdomen and pelvis revealed a fatty liver and a hypoplastic uterus.

Intelligence testing indicated that her IQ was 40. After consultation with an endocrinologist, she was diagnosed with Prader-Willi syndrome. Her management included hormonal therapy and dietary advice directed by the endocrinology department, skin treatment directed by the dermatology department, and speech therapy. She was also given risperidone 1 mg/d for behavioral control. Her family was educated about the illness. After 8 weeks of this multi-phased intervention, her irritability, stubbornness, temper tantrums, increased speech, and self- injurious behavior improved significantly. She tolerated the risperidone well without any significant adverse reaction. After 4 months of treatment the dose of risperidone was reduced to 0.5 mg per day.

3. Discussion

In the past the Prader-Willi syndrome was diagnosed based on the clinical presentation, but genetic testing can now more accurately diagnose the condition. In high-income countries, genetic testing is recommended for all infants with pronounced hypotonia; however, in most low- and middle-income countries genetic testing is not available, so the diagnosis still depends on the correct identification of the typical clinical symptoms. Given the relative rarity of the disorder and the unfamiliarity of most clinicians with the condition, many cases go undiagnosed.

It is not feasible to correct the genetic abnormality, so most treatments are aimed at suppressing unwanted symptoms. Given the frequent occurrence of difficult-to-manage behavioral problems in PWS, clinicians often try low-dose antipsychotic medication. One study unexpectedly found that antipsychotic medications – which often lead to weight gain in patients with schizophrenia – was associated with weight loss in patients with PWS.[11] However, the small numbers of individuals with PWS make it difficult to conduct formal evaluations of the effectiveness of antipsychotic medications or other interventions, so it has not been possible to develop evidence-based treatment guidelines for the condition. In most cases, clinicians must use their judgment to individualize the treatment to the needs of each patient. As shown in the current case, the ongoing involvement of multiple disciplines along with educational and psychological support for the care-givers is often needed to address the complex needs of these patients and their families.

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Conflict of interest statement

The authors declare that they have no conflict of interest related to this manuscript.

Informed consent

The father of the patient signed an informed consent form and agreed to the publication of this case report

Authors' contributions

SM drafted the manuscript. UKP critically reviewed the manuscript. SM and UKP both carried out the clinical diagnosis and the psychiatric evaluation. Both authors read and approved the final manuscript.

Prader-Willi 综合征患儿的行为与情绪表现

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概述： Prader-Willi 综合征是一种神经发育障碍，以精神发育迟滞以及明显的躯体、行为与精神方面的表现为特征。适应不良性行为、认知损害以及言语和语言障碍严重影响患者早期发育，也会影响患者的长期功能。本文报告一例 9 岁的 Prader-Willi 综合征患儿，以低剂量抗精神病药物治疗其行为症状。

关键词： Prader-Willi 综合征；精神症状；儿童期障碍；病例报告；印度

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