Case report

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Williams Syndrome presenting with attention deficit hyperactivity disorder and management approach in a resource-limited setting

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Introduction

Williams syndrome, also known as William-Beuren Syndrome, is a rare genetic condition affecting approximately 1 in 20,000 births. Individuals with Williams syndrome often exhibit a unique facial appearance, including a broad forehead, upturned nose, full cheeks, and small chin the facial appearance changing with increasing with age (1). They may also have dental abnormalities, such as widely spaced teeth. Other characteristics include a vasculopathy associated with supravalvular aortic stenosis, peripheral pulmonary stenosis and renal artery stenosis, hypercalcaemia and endocrine abnormalities including hypothyroidism and diabetes mellitus. Cognitive deficits are usually associated but can be variable and behavioural disorders are associated (2).

Williams syndrome is caused by a 1.5-1.8 Mb heterozygous deletion of chromosome 7q11.23 with a loss of around 28 coding genes (1). The diagnosis can be made clinically in the presence of typical dysmorphisms, typical cardiac defects, hypercalcaemia and cognitive and behaviour phenotypes, but is usually confirmed by identification of the microdeletion using genetic testing (MLPA, FISH or microarray).

Leyfer et al. reported an 80% prevalence of at least one psychiatric disorder in a sample of children with Williams syndrome (2). The most prevalent diagnoses were attention deficit hyperactivity disorder (ADHD) in 65% and specific phobia in 54% (2). It can be postulated that shared genetic factors play a role in the development of both Williams syndrome and ADHD. The high prevalence of ADHD in individuals with Williams syndrome highlights the importance of early identification and appropriate management. By recognising and addressing both conditions, individuals with Williams syndrome can receive the support and interventions necessary to maximise their potential.

This case report describes a child who presented with hyperactivity, inattention, and impulsivity. The child was subsequently clinically diagnosed with both Williams syndrome and ADHD and received treatment in a resource-limited setting.

Case report

Master S, a nine-year-old boy from a rural village in the North Central Province of Sri Lanka, was referred to a child and adolescent psychiatrist by his school principal due to a constellation of concerning behaviours. He exhibited hyperactivity, excessive eating, a penchant for singing and dancing, and persistent difficulties focusing during academic tasks, resulting in suboptimal academic performance.

Master S's medical history began with a premature birth via emergency lower segment caesarean section necessitated by foetal distress. Later, he was diagnosed with a small ostium secundum and tight-branched pulmonary artery stenosis. He had normal audiology and vision assessments. He had global developmental delay as an infant, including difficulties sitting without support and reaching for objects at ten months.

At two years and two months, Master S was diagnosed with failure to thrive and continued to lag his peers in developmental milestones. The emergence of facial dysmorphism prompted a referral to a clinical geneticist, which, unfortunately, was not pursued. By age five, Master S's heart condition had resolved, but his growth remained below average. He also exhibited subtle dysmorphic features, including brachycephaly, a broad nose, large nares, epicanthic folds, a high-arched palate, broad palms, and stocky fingers. Master S demonstrated below average intelligence, a deviation quotient of 75, with reading and writing skills were below grade level. He exhibited hyperactivity and impulsivity, and his mood was generally stable.

Figure 1. Facial dysmorphism of the nine-year-old boy suspected to have Williams Syndrome anterior view (A) and lateral view (B).

A clinical geneticist ultimately diagnosed Master S with Williams Syndrome with atypical features that need further genetic testing for confirmation. The child and adolescent psychiatrist conducted a clinical assessment, including an intelligence and learning assessment. Master S was diagnosed with attention deficit hyperactivity disorder according to the DSM5 criteria. A family meeting was convened to discuss the diagnosis and develop a comprehensive treatment plan. Methylphenidate was prescribed to address his hyperactivity and impulsivity, and remedial teaching was initiated.

An Individual Educational Plan was commenced for Master S with specific strategies for a consistent schedule, clear expectations and visual aids. An educational therapist introduced him to hands-on activities using the multi-sensory approach. Further, tasks were broken down into smaller, more manageable steps, with appropriate seating arrangements to minimise distractions. Follow-up visits revealed significant improvements in Master S's impulsive eating and hyperactivity. However, progress in his scholastic skills was slower. The initial treatment plan was continued, with ongoing monitoring and adjustments as needed.

Discussion

Williams Syndrome is a rare genetic disorder characterised by distinct physical features, cognitive strengths and weaknesses, and behavioural challenges. Individuals with Williams Syndrome often exhibit a unique personality profile, including a gregarious and outgoing nature, a love of music, and a strong emotional connection to others (3). The diagnosis of Williams Syndrome provided a framework for understanding Master S's unique challenges and strengths, allowing for more targeted interventions and support. In Sri Lanka, many patients with Williams Syndrome are diagnosed clinically, as many parents of affected children are unable to afford currently available molecular diagnostic testing.

In addition to the challenges posed by Williams Syndrome, Master S also faced the complexities of ADHD. This condition, characterised by hyperactivity, impulsivity, and difficulty concentrating, can significantly impact a child's academic and social functioning. The combination of Williams Syndrome and ADHD presented a unique set of challenges for Master S and his family. Methylphenidate was a crucial component of his treatment, helping to manage his hyperactivity and impulsivity. Green et al. studied methylphenidate treatment of ADHD in children with Williams Syndrome and found that a clinically significant improvement was reported in more than 70% without major adverse effects (6). However, it would have been better to commence the treatment earlier in Master S to support him in acquiring scholastic skills from his school entry.

While individuals with William Syndrome may struggle with foundational mathematical concepts, they can often acquire various mathematical skills and exhibit strengths in areas like number recognition (7). Leveraging their comparative strengths in verbal abilities, individuals with William Syndrome can benefit from language-based approaches to learning certain mathematical concepts. Although individuals with William Syndrome often possess strong oral language skills, they typically have intellectual disabilities and may face challenges in reading comprehension (8). Reading abilities among individuals with William Syndrome vary significantly.

Master S exhibited average intelligence, and we implemented targeted educational interventions to enhance his academic performance. Given the scarcity of educational psychologists, trained therapists carried out the interventions.

Child and adolescent mental health services are limited to a few specialised hospitals in Sri Lanka (9). The long waiting times at multiple clinics and long distances of travelling to seek expert opinions at tertiary care centres in the city, causing a loss of daily wages, further constrict the possibilities for an ideal management plan for low socioeconomic groups such as this family. The diagnosis of Williams Syndrome and ADHD provided a clearer understanding of his unique needs, and the treatment plan was designed to address these challenges effectively. With continued support, Master S could look forward to a future filled with opportunities and personal growth. Furthermore, this report emphasises the importance of screening for child psychiatric comorbidities in Williams Syndrome early in the management to commence relevant interventions.

Statement of contribution

MSPP, PV: Writing of the initial draft, Literature review, and clinical assessments.

KDCTP, DCDS, MC: Revisions of the initial draft, clinical assessments.

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