

## Case Report

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# Williams-Beuren Syndrome: Manifestation with Attention Deficit Hyper Activity Disorder and Voiding Dysfunction with Kidney Anomalies; Good Response to Ritalin

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Williams-Beuren syndrome is a rare neurodevelopmental disease, in which mental retardation is common with this submicroscopic chromosomal deletion genetically disease. We reported a boy with Williams-Beuren syndrome and highlighted the clinical characteristics and response to treatment with Ritaline of such an individual with ADHD and voiding dysfunction.

The patient was a six year-old boy with typical presentation of WBS and some psychiatric problems referred because of hyperactivity symptom and attention deficit.

Children with mental retardation and WBS have a comorbidity of psychiatric disorders and kidney anomalies.

Williams-Beuren syndrome can present with hyperactivity and Ritalin can improve some symptoms and voiding dysfunction.

**Keywords:** Williams-Beuren Syndrome; Attention Deficit Disorder with Hyperactivity; Ritalin; Methylphenidate.

**Running Title:** Williams-Beuren Syndrome: Manifestation with Voiding Dysfunction

## Introduction

Williams-Beuren syndrome (WBS) is a rare neurodevelopmental and genetically disease explained nearly 50 years ago. It involves central nervous system and connective tissue [1, 2].

The molecular mechanism is a submicroscopic chromosomal deletion affecting the elastin gene (ELN) in 7q11.23. The incidence is one in 20000 live births following spontaneous mutations [3].

WS and attention deficit hyperactivity disorder (ADHD) show similar patterns of neuropsychological tasks, particularly in working memory and delayed short-term memory [4].

A survey among 30037 Norwegian children born between 1980-1985 revealed three children with WBS with a prevalence of 1 in 7500 with 7q11.23 deletion in all cases. Therefore, one of the etiologies for MR is WBS [5].

20-35% of children with mental retardation have been shown to have a comorbid psychiatric disorder [6]. Because of possible limited population-based data on the occurrence of Williams-Beuren syndrome, unfortunately, we did not find any information about the epidemiology of WS in Iran and there is few reported cases about this syndrome and comorbidity with ADHD.

Therefore, this case had some characteristics:

- It is a rare case report about psychiatric problem of WS
- It is rare because of consideration of urinary problem in this syndrome
- A stimulant as suitable treatment for voiding dysfunction is suggested

## Case Report

This was a six year-old boy from Yazd (The center area of Iran) with specific appearance, short stature and dysmorphism. He was born at 38 weeks of gestational age with a birth weight of 3 kg. The symptoms included hyperactivity, attention deficit, sociability, openness, overfamiliarity, anxiety, specific phobia, mild mental retardation, impairment in verbal and visuospatial skills.

In general appearance, short palpebral fissures, medial eyebrow flare, a depressed nasal bridge and everted nares, elfin like face, thick lips and iris with stellate form were detected.

Familial history of psychiatric disorder was negative.

Although he trained for toilet rituals, his mother complained of urgency, diurnal and nocturnal enuresis. This caused problems for his family, especially his mother. As average, he had bed wetting 5 times and diurnal enuresis 3 times weekly.

**Paraclinic findings:** Thyroid abnormalities, Kidney anomalies, hyperacusis in audiometry, asymptomatic hypercalcemia and cardiovascular problems like supra aortic valve were detected. Intelligence quotient (IQ) test as Raven test was 62 (Mild mental retardation range).

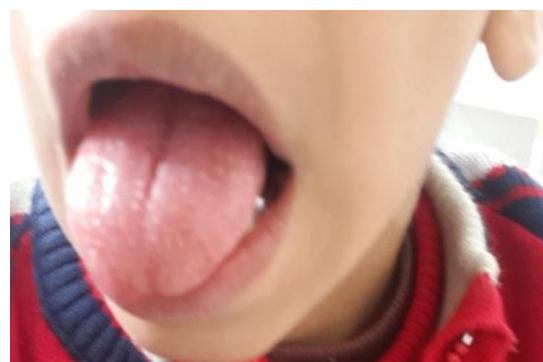
**Urinary tract findings:** The gall bladder, ureters and urethra were intact. The size of kidneys was smaller than normal; nephrocalcinosis, marked asymmetry in kidney size and some small cysts in both kidneys were detected. Urine Analysis had Normal result. CBC, BUN and Creatinine all had normal findings.

**Treatment plan:** The psychiatrist prescribed Tablet Ritalin 10 mg daily after cardiovascular consultation. He did not take antibiotics because of no urinary tract infection. After one month, he was referred again and the attention was suitable and hyperactivity and anxiety were declined. Occupational therapy and speech therapy were suggested. Moreover, it was ordered to avoid taking extra calcium and vitamin D. As history taken from his mother, nocturnal and diurnal enuresis decreased noticeably (partial response,

approximately 1 time for nocturnal and no diurnal enuresis).



**Figure 1.** Mild little-finger clinodactyly in William's syndrome



**Figure 2.** Thick lips and no abnormality in child's tongue

## Discussion

Our patient was a boy with both WBS and ADHD and more manifestations of Williams-Beuren syndrome.

His facial features were characteristic for WS. There was no nausea or vomiting, a symptom frequently seen in this syndrome.

In a study, seven types of anomalies were reported in 40 children with WBS [7]. Moreover, the risk of anatomical anomalies of the kidneys and the urinary tract is accelerated 12-36-folds in WBS [8]. Therefore, ultrasound screening of renal system should be part of the first assessment of patients with WBS for diagnosis of voiding dysfunction and abnormalities [8, 9]. Urology consultation and assessment for urinary system were considered before our visit by the

pediatricians. The urologist ruled out urinary tract system etiologies for voiding dysfunction.

ADHD patients have significantly higher rates of nocturnal enuresis, urgency and infrequent voiding [10]. We expected that ADHD with WS cause increased the risk of enuresis and voiding dysfunction because of probable impulsivity and disinhibition. Generally, an IQ of 41-80 is detected in WBS [11]. Our case was in medium range.

In a study, 43% met the criteria for ADHD [12]. Our case met full criteria for combined ADHD. Among all psychiatric and behavioral disorders, hyperactivity, impulsivity and inattention were prominent in this patient.

We suggest pediatricians and neurologists to consider psychiatric problems of children with genetic disorders. Hence, suitable psychiatric approaches can improve many problems and decline some tensions and conflicts of parents.

## Conclusion

Williams-Beuren syndrome can present with hyperactivity, inattention and kidney anomalies. Ritalin may improve some symptoms and voiding dysfunction dramatically.

## Conflict of Interest

Authors have no conflict of interest to declare.

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**Authors' contribution:** RB prepared and participated in drafting, revising, designing and submission of the manuscript. The case was presented for more evaluation by TS and she participated in discussion.

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