Williams Syndrome with Severe Sensorineural Hearing Loss

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Williams or Williams-Beuren Syndrome (WS) is a very rare syndrome associated with a microdeletion on chromosome 7q11.23. Williams syndrome is characterized by multiple congenital anomalies including distinctive facial features, cardiovascular anomalies, neurodevelopmental delay and mental retardation. Sensorineural hearing loss is not reported very often in WS patients. The purpose of the study is to present a Williams syndrome patient with severe sensorineural hearing loss.

Bilateral severe sensorineural hearing loss was diagnosed in a child of 4 years old with WS. The child was fitted with binaural hearing aids and began to receive auditory habilitation. The child benefited from the amplification. Audiological evaluation is recommended for children with multiple anomalies including WS in order to prevent the harmful effects of the hearing loss.
Williams or Williams-Beuren Syndrome (WS) described firstly by Williams et al.\(^1\) and Beuren et al.\(^2\) is a very rare syndrome associated with a microdeletion on chromosome 7q11.23\(^3,4\).

The syndrome primarily involves vascular, connective tissue and central nervous systems. Genetic analysis has revealed a variable deletion on chromosome 7 at elastin gene locus and its inheritance has been shown to be sporadic rather than familial\(^3\).

The estimated incidence ranges between 1/7500 and 1/25000 newborns.\(^5,6\).

Clinical features of WS include distinctive facial appearance, cardiovascular anomalies, short stature, mental retardation, neurodevelopmental anomalies and infantile hypercalcemia.\(^1,2,7,8\). Other commonly associated findings include psychomotor problems such as attention deficit disorder with hyperactivity, hyperacusis and otitis media.

Facial anomalies can manifest in a wide range of symptoms including a coarse face with periocular fullness, stellate or starburst irides among blue eyed individuals, flat nasal bridge, short up-turned nose, antverted nares, wide mouth, long philtrum, macrostomia, flat facial profile, full lower cheeks and a small chin\(^9\).

We describe a Williams syndrome case with severe sensorineural hearing loss.

High frequency hearing loss and conductive hearing loss in Williams syndrome has been reported in some cases\(^10-14\) but to our knowledge, severe flat sensorineural hearing loss has not been documented elsewhere.

**CASE REPORT**

The patient is a 4 years old girl referred to Marmara University Medical School Audiology department for the hearing evaluation. Her otoscopic examination was reported as normal. She was the product of a full term pregnancy without any complication. There is no consanguineous marriage between the parents and no history of hearing loss or mental retardation in the family.

She has been operated because of large ventricular septal defect and pulmonary hypertension at the age of 4 months old. She had genetic analysis at the age of 5 months and was diagnosed as WS.

She has a coarse face with thick lips, wide mouth, full cheeks, puffiness around the eyes, periocular fullness, and flat nasal bridge with a short nose with antverted nares (Figure 1).\(^9\)

![Figure-1: The craniofacial features are typical for Williams Syndrome.](image)

She has sloping shoulders and short stature. She had feeding difficulties and vomiting in early childhood.

The patient, now aged 8 years old, represents neurodevelopmental delay and a lower IQ. She receives auditory and cognitive rehabilitation services and continues to a preschool for the hearing impaired children.

**Audiological evaluation**

Her free field audiogram using VRA to warble tones revealed hearing thresholds from 250 Hz to 4000 Hz in a range of 70 to 95 dB HL in her first audiological examination at the age of four years old (Figure 2). She was uncooperative to the play audiometry. No startle response was observed to 3 kHz 100 dB narrow band noise, she was not able to localize warble tones or speech. The speech detection threshold was obtained at 65dB nHL.
Acoustic Immitancemetry (with Interacoustic AZ 7) revealed type “A” tympanograms in both ears. Acoustic middle ear reflexes were absent bilaterally. No otoacoustic emissions (with ILO-96 Otodynamic Analyzer and desktop computer turned on) were obtained. Auditory brainstem response (ABR) was performed under the sedation with chloral hydrate (50 mg/kg). Amplaid MK15 evoked potential system was used. Wave V was obtained at 70 dB nHL bilaterally with click stimulus.

Frequency specific tonal ABR was performed using 500 Hz, 1000 Hz, and 2000 Hz logon stimuli. Thresholds obtained for each frequency tested were shown at the Table I.

<table>
<thead>
<tr>
<th>Frequency (Hz)</th>
<th>500 Hz</th>
<th>1000 Hz</th>
<th>2000 Hz</th>
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</thead>
<tbody>
<tr>
<td>Right</td>
<td>75 dB nHL</td>
<td>75 dB nHL</td>
<td>75 dB nHL</td>
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<tr>
<td>Left</td>
<td>80 dB nHL</td>
<td>80 dB nHL</td>
<td>95 dB nHL</td>
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Table-I: Tonal ABR thresholds for right and left ears in four frequencies tested.

The child was fitted with digital behind the ear hearing aids and enrolled for family centered auditory-oral therapy. Her sound field audiogram with the hearing aids is shown in the Figure 2. She is able to detect speech at 40 dB HL when aided.

Her latest play audiometry under earphones revealed bilateral severe sensorineural hearing loss (Figure 3). The speech reception threshold was obtained at 75 and 70 dB HL for right and left ears respectively.

She only wears her hearing aids at school she shows overt aversion response when the hearing aids are on although they are properly fine-tuned according to her hearing status.

Her speech and language development progressed rapidly after hearing aids. Her vocabulary increased, she is able to express herself in 3 to 4 word short sentences.

The Meaningful Auditory Integration Scale (MAIS) was administered at the beginning of hearing aid use and was repeated at one, two and three years after amplification. MAIS score increased from 8/40 to 30/40 after 4 years of hearing aid use.

**Discussion**

Williams syndrome is a rare, sporadic congenital anomaly. It is characterized with dysmorphic facial features, cardiovascular anomalies, neurodevelopmental delay and infantile hypercalcemia. Dysmorphic characteristics and cognitive anomalies and hyperacusis, the gene studies performed in WS patients have been well
To our knowledge, the case that we have documented is the only case with severe flat sensorineural hearing loss in the literature.

Since WS is often accompanied by neurodevelopmental delay and mental retardation, hearing loss may be mistaken and the poor speech understanding and delayed language development can be easily attributed to the mental retardation. The possibility of hearing loss should be kept in mind for WS children.

Another feature commonly reported in these patients is hyperacusis which may disturb the use of hearing aids as in our case. Desensitisation for hyperacusis may help these patients to wear their hearing aids.

Early diagnosis of hearing loss and early amplification will enable the child to develop speech, language and communication skills and facilitate the rehabilitation.

Our case suggests that infants with dysmorphic facial features, cardiovascular anomalies and neural development delay would benefit from an audiological evaluation. WS patients like all other patients with different syndromes also benefit from the team approach.

**Acknowledgements**

We informed the patient’s family of the purpose of this report and our intent to protect the patient’s privacy. We obtained full consent to describe the present case.

**REFERENCES**


