CASE REPORT

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 periorbital fullness, stellate or starburst irides among blue eyed individuals, flat nasal bridge, short up-turned nose, anteverted nares, wide mouth, long philtrum, mascrostomia, flat facial profile, full lower cheeks and a small chin [9].

We describe a Willams 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 hypertention 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, periorbital fullness, and flat nasal bridge with a short nose with anteverted nares (Figure 1).

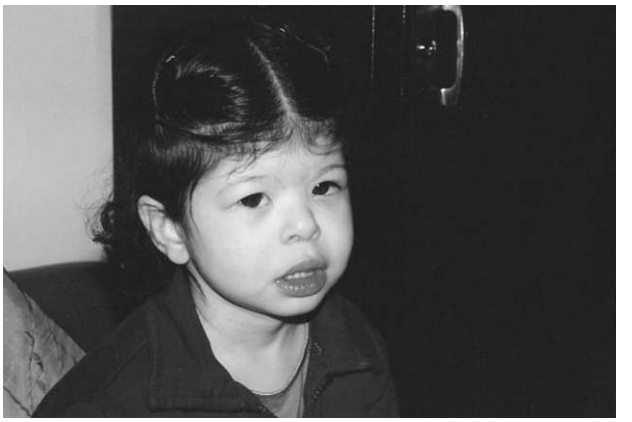

Figure-1: The craniofacial features are typical for Williams Syndrome.

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 continiues 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.

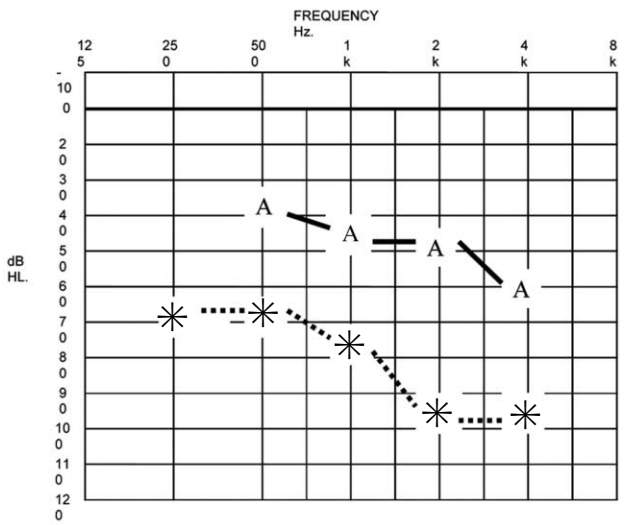

Figure-2: Hearing thresholds with and without hearing aids to warble tones from 250 to 4000 Hz in the sound field. Hearing thresholds in the soundfield are shown with. Aided thresholds in the sound field are marked with A.

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.

	500 Hz	1000 Hz	2000 Hz
Right	75 dB nHL	75 dB nHL	75 dB nHL
Left	80 dB nHL	80 dB nHL	95 dB nH

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.

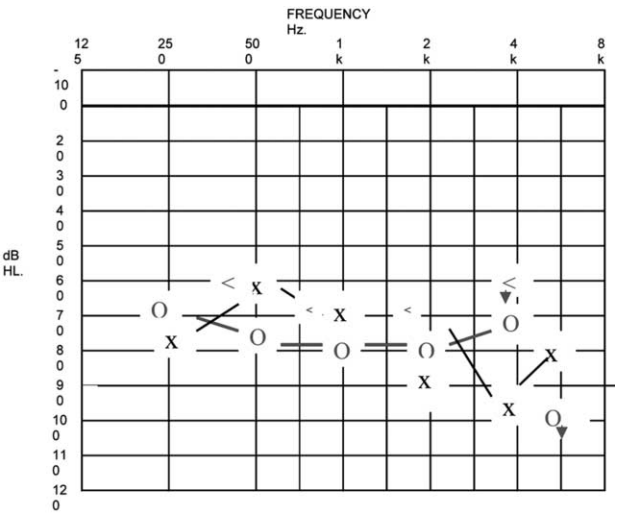

Figure-3: Hearing thresholds with play audiometry under earphones Right ear thresholds to pure tone d are shown with O and left ear thresholds with X.

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

documented in the literature [3-5,9]. Previous reports of WS patients have provided more details of their genetic analysis, cognitive delay and less about their hearing.

Few published studies have analysed the hearing sensitivity of WS patients [10-14], in most of the studies concerning WS hearing was assumed to be normal or not reported.

Klein et al. [14] reported that 61% of the subjects with WS had a history of otitis media.

Hearing loss in WS is reported not very often [8-12], all the cases reported had high frequency sensorineural hearing loss except the patient reported by Lopez-Rangel et al. had unilateral profound congenital sensorineural hearing loss [15].

In the review of Cherniske et al. [11] high incidence of sensorineural hearing loss was reported in adults with WS. They found a much higher than expected incidence of mild to moderate high frequency sensorineural hearing loss (75%) than would be expected in the general population [ISO 1984].They postulated that there might be a premature aging process with WS.

Johnson et al. [10] reported sensorineural hearing loss in 3 of 9 patients with WS; 2 patients had unilateral moderate high frequency sensorineural hearing loss and 1 patient with bilateral moderate high frequency hearing loss.

Marler et al. investigated possible age related hearing loss in WS patients. Their observation of hearing loss in adults was consistent with previous studies of moderate high frequency sensorineural hearing loss. A greater degree of hearing loss was observed in adult patients than in children suggesting that hearing loss increases in severity with age [12].

Gothelf et al.'s [13] study agrees with the earlier reports of high frequency sensorineural hearing loss in WS patients. They found that the high frequency hearing thresholds of the children with WS were higher than the control group.

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 rehabilitatior.

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

Acknowlodgements

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.

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