CASE REPORT

The effects of Goldenhar Syndrome on hearing and speech development

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SUMMARY

Goldenhar syndrome is a congenital abnormality with an incidence of 1 in 5,200 to 26,500 births. This syndrome is characterized by facial asymmetry, ear malformation, and/or defects in the eyes and vertebrae. The hearing disorder manifests as both conductive or sensorineural due to the abnormalities occurring in the inner and outer ear. We report a case of a 1-year-3-month-old child presenting with left anotia and right microtia, severe bilateral conductive hearing loss, and global delayed development. The patient was also found to have a hemifacial microsomia, a secundum atrial septal defect (ASD), and a ventricular septal defect (VSD). The patient was advised to use hearing aids and participate in speech therapy. The management of this Goldenhar syndrome patient should be done comprehensively, appropriate to the abnormalities found to achieve the best result.

INTRODUCTION

Goldenhar syndrome (GS) is a congenital condition presenting with facial, ear, and vertebral abnormalities. It is characterised by hemifacial microsomia and ear defects causing clinical manifestations such as hearing disorder due to the incomplete formation of various ear structures.[1,2]

Although this syndrome's exact etiology and pathogenesis have not yet been pinpointed, it is suspected that this condition arises from anomalies in the first and second brachial arc and/or neural crest. The interaction between genetic and environmental factors such as infection, drug use, and alcohol could play a role in this disease.[1,2]

As GS is a sporadic disease, genetic factor is not always acting as the primary determinant. Even though this disease is incurable, it is treatable according to the presenting symptoms. If GS is accompanied with hearing problems, thus a hearing aid and speech therapy are needed to increase patients' quality of life and self-esteem.[2,3]

CASE REPORT

A one-year-three-month-old male patient came to Cipto Mangunkusumo Hospital (CMH) Community ENT division, Universitas Indonesia, Indonesia, referred from the pediatrics department for a hearing examination. The patient presented with a congenital complaint of the absence of ear canals in both ears with no left earlobe, alongside a small right earlobe. The patient could only respond to loud noise such as a forcefully closed door. The patient had not been able to say meaningful words. In speech development, he started babbling at the age of 13 months. A delay in motor development was noted as the child had only been able to sit at 15 months and could not crawl, stand, or walk.

The mother denied any history of infections, hypertension, diabetes, and drug consumption during pregnancy. The patient was born at full term with a cesarean section due to breech presentation. He cried soon after delivery and weighed 2,300 grams. One week after the delivery, the patient suffered from hyperbilirubinemia and was treated with blue light therapy for two days.

At the age of three months, the patient was brought to the paediatrics clinic in CMH with complaints of dyspnoea; the patient was further found to have a leaky heart valve. The patient was then suggested to undergo routine visits as no further surgical management was deemed needed. When referred to our division, the patient weighed 9.7kg with a length of 54cm. No sign of dyspnoea was found, and vital signs were in the normal range. However, the physical examination found that the patient was normocephalic, had a hemifacial microsomia with micrognathia, right ptosis, and lagophthalmos of the left eye. In addition, ENT clinical examination found abnormalities of both ears; those are left anotia and a second-degree microtia of the right ear (Figure 1). Neurological examination revealed a global delayed development alongside left hemifacial VII cranial nerve paresis. A grade 3/6 parasystolic murmur at the left inferior sternal border with no gallop was found during cardiac diagnostic evaluation.

Brainstem-evoked response audiometry (BERA) examination—with a click stimulus rate of 27.7/second on the rarefaction—found no electrophysiological response of the fifth wave up until 80dB stimulus on both ears. Bone conduction BERA examination found an electrophysiological response of the fifth wave at 30dB on both ears (Figure 2). Masking was unable to perform due to bilateral ear abnormalities. In paediatric patients with bilateral atresia, a BERA examination is one of the best choices to identify any residual cochlear hearing function.

A secundum atrial septal defect and a small perimembranous ventricular septal defect were revealed during routine echocardiography (Figure 3).

A suspicion of vertebral deformities at level T8-10 arose from

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Fig. 1: The patient presented with hemifacial microsomia, left anotia, and right ear microtia.
Permission from the parents of the patient were obtained for the publication of this photograph.

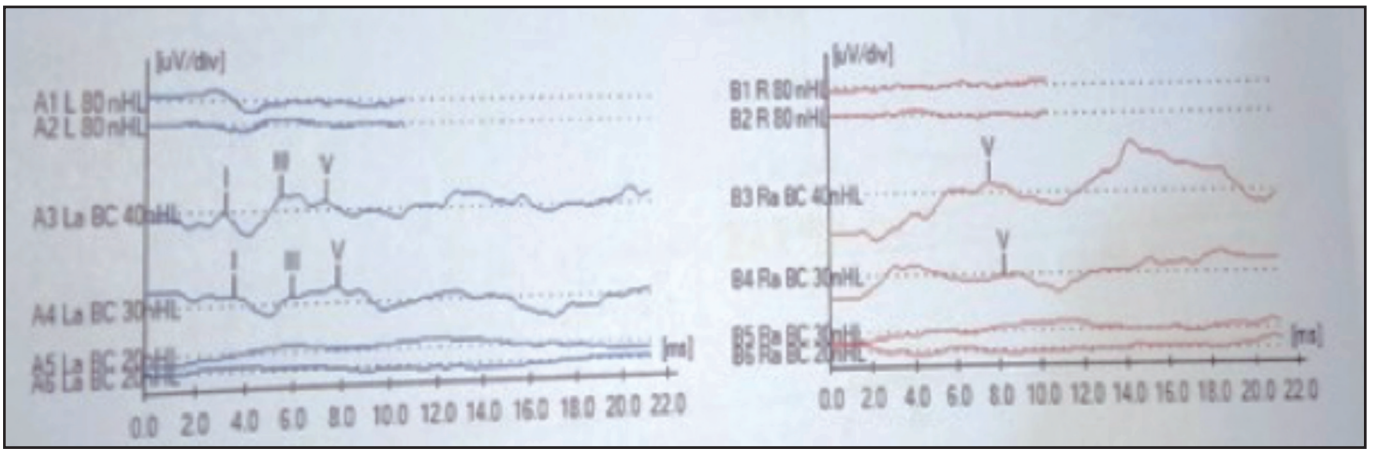

Fig. 2:

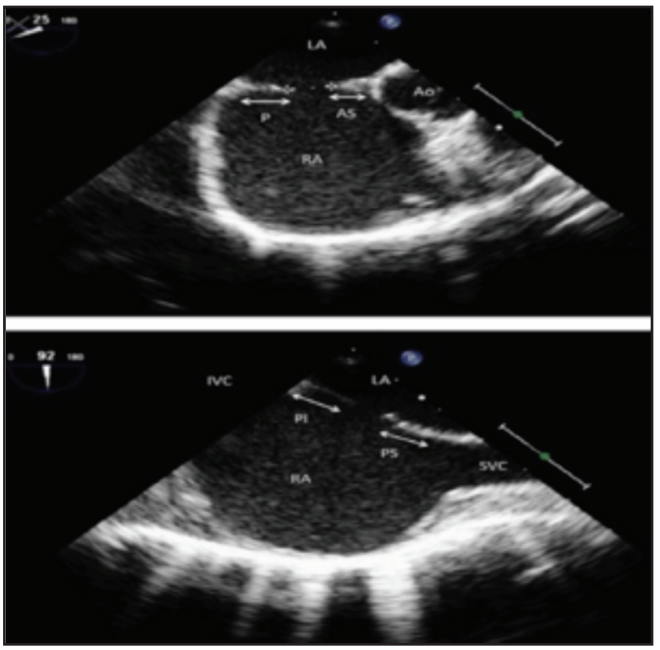

Fig. 3: Echocardiogram revealing an ASD and VSD found in the patient.

an abdominal x-ray examination (babygram with AP position). MRI imaging did not find any haemorrhage, infarction, nor intracranial lesion. However, simple contacts between the right anteroinferior cerebellar artery and V, VI, and VII right cranial nerve structures were found, albeit with no nerve compression.

The patient was diagnosed with GS based on deformities and abnormalities found on the face, ear, and heart, accompanied by global delayed development, left VII cranial nerve palsy, and severe bilateral conductive hearing loss. Therefore, by taking into account the clinical symptoms, physical examinations, and additional investigations such as the audiology examinations, the patient was confirmed to have GS, left anotia, second-degree right microtia, bilateral ear canal atresia, severe bilateral conductive hearing loss, secundum ASD and VSD.

DISCUSSIONS

The incidence of GS ranges from 1 in 3,500 to 5,600 live births and is found in 1 out of 1000 children with congenital hearing loss. There is a slight male preponderance with a ratio of 3:2 compared to that of females. Eighty-five percent of this syndrome presents unilaterally, while the remaining

10-33% occur bilaterally.[1,2] The exact aetiology of GS has not been yet fully elucidated, but it is found to be inherited in an autosomal dominant and/or recessive manner. This disease is thought to be a multifactorial condition involving interaction between genetic and environmental factors. Infectious features such as positive results of anti-CMV (cytomegalovirus) IgG, anti-rubella IgG, and anti-HSV (herpes simplex virus) IgG are thought to be the most probable cause of GS. Nevertheless, other factors such as alcohol consumption, smoking, and the use of vasoactive drugs (retinoic acid, tamoxifen, diabetes medications) during pregnancy could also contribute as precipitating factors.[4,5]

Typical clinical manifestations of GS include hemifacial microsomia, facial asymmetry, preauricular skin tag, epibulbar dermoid, and vertebral anomalies. Other craniofacial abnormalities include microsomia and outer ear, mandible, temporal bone, zygoma, outer ear, facial nerve, bones, and surrounding tissues malformation. Alternatively, the term 'oculo-auriculo-vertebral dysplasia is often used to depict this syndrome due to the accompaniment with epibulbar dermoid, auricle malformation, and fistule, alongside vertebral anomalies.[4-6]

In our case, abnormalities involving multiple organs such as eyes, ears, heart, and vertebrae were found. In addition, neurological examination revealed a VII cranial nerve palsy affecting the left side of the face, along with global delayed development.

Ear abnormalities that were found in this patient include left anotia and second-degree right microtia. It is worth noted that microtia is one of the most common abnormalities found in this syndrome, comprising 90% of all ear malformation in Goldenhar syndrome.[4,6] Other than that, the patient also had severe bilateral conductive hearing loss. The ear malformation found in this patient contributes to causing conductive hearing loss, which hindered auditory development before finally impacting speech development.[6,7]

Abdominal x-ray examination (babygram in AP position) revealed a suspicion towards T8-10 vertebral body deformities. The deformities that present in the thoracal segment contribute to the postural instability of the patient. This head-neck postural abnormality happened due to the typical craniofacial deformities of Goldenhar syndrome.[4] Poor postural control of the head, neck, and thoracal segment of the spine will negatively impact the breathing, speaking, and swallowing function. These postural problems are thus negatively affecting quality of life of the patient in the long run.[4,8]

It has been thoroughly researched and shown that postural stability and sensorimotor integration process play a vital role in the functional maturation of a child's speaking and swallowing abilities. Thus, concerns should be placed more upon the role of postural control and sensorimotor maturity.[8] Postural coordination exercises and interventions comprising postural modification have been extensively examined and proven to be correlated to a better upper airway sensorimotor integrity and ability. This, in turn, will support better sucking-breathing-swallowing cycle coordination alongside clearer and stronger speech articulation. In this case, it is also worth remembering that our patient also suffered from facial nerve palsy, adding further issues to what was an already compound problem. Many studies have reported that children with postural control failure had a higher risk of aspiration during feeding, due to the uncoordinated and non-integrated upper airway sensorimotor function. It will pose significant danger if not properly intervened.[8,9] Hence, collaboration with fellow clinicians from the medical rehabilitation division is needed to modify and exercise postural control therapy. The patient is also experiencing delayed motor development, making the importance of physiotherapy under the guidance of medical rehabilitation experts more prominent, besides the hearing aid use and therapy speech itself.

The parents have received information regarding treatment plans—specific to hearing problems—in which a conventional bone-conduction hearing aid, called bone-anchored hearing aid (BAHA), will be used. BAHA is chosen as it is a good option for hearing habilitation in bilateral microtia-atresia cases and can improve the quality of life of the patients significantly in the long run. Besides, BAHAs are relatively accessible in terms of availability and pricing.[5-7,10] The parents were also educated regarding speech therapy plans after hearing aid usage and the possibility of surgical intervention. One of the intervention choices is reconstruction surgery which will address the patient's microtia or ear malformation. It will only be done when the patient has reached the appropriate age.[4,6]

Hence, a solid collaboration between fellow clinicians in the field of ENT, medical rehabilitation, and paediatric neurology is of the utmost importance in ensuring the best possible development in children with GS.

CONCLUSIONS

Goldenhar syndrome is a congenital disease with a variety of risk factors and multi-organ involvement. The hearing disorder is one of the culprits behind the delayed speech and hearing development in patients. Therefore, an accurate diagnosis coupled with comprehensive multidisciplinary management is needed to achieve the best results for the patients.

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