Case Report

Congenital Malformations of the Inner Ear: Case Series and Review of the Literature

Patorn Piromchai MD, MSc, FRCOT, FICS*, Pornthep Kasemsiri MD, FRCOT*,
Panida Thanawirattananit BNS, MA*, Kwanchanok Yimtae MD, FRCOT*

** Department of Otorhinolaryngology, Faculty of Medicine, Khon Kaen University, Khon Kaen, Thailand*

Patients with craniofacial anomalies often present to doctors due to their noticeable disfigurement and are routinely assessed by otolaryngologists for hearing evaluation. However, small percentage of craniofacial anomaly patients may present with delayed speech though they may not have initial obvious external deformation. The objective of case series is to identify the congenital inner ear malformation. The series of clinical presentation, physical examination, investigations, treatments and follow-up results were demonstrated followed by the discussion.

Keywords: *Hearing loss, Inner ear, Craniofacial anomaly*

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The undetected hearing loss in infants can significantly affect their speech and language acquisition ability. The congenital hearing loss is classified as genetic and non-genetic disorder[1,2]. Genetic causes account for fifty percent of these cases, and the remainder attributed to environmental factors[2]. This anomaly can manifest as conductive, sensorineural or mixed hearing loss[3].

The congenital craniofacial anomaly patients usually come to see the doctors due to their noticeable disfiguration. As part of their clinical evaluation, these patients are routinely referred to the otolaryngologists for hearing evaluation. It is aimed at early detection of underlying hearing problem and hearing rehabilitation early in their life to achieve normal speech and development.

The minor group of patients with syndromic congenital ear anomaly does not have the obvious external deformation and can present with the sensorineural or mixed hearing loss. Examples include Stickler syndrome[4-6], Pfeiffer syndrome[7], Saethre-Chotzen syndrome[8,9] and hemifacial microsomia[10]. Another group is the congenital inner ear anomaly patients with no associated visible craniofacial abnormalities[3]. As a result, these minority groups were left undetected or late detected.

Correspondence to:
Yimtae K, Department of Otorhinolaryngology, Faculty of Medicine, Khon Kaen University, Khon Kaen 40002, Thailand.
Phone: +66-86-8541515
E-mail: kwayim@gmail.com

The objective of the present study is to identify a subgroup of the congenital craniofacial anomaly patients that solely have inner ear malformation. The series of clinical presentation, physical examination, investigations, treatments and follow-up results were demonstrated followed by the discussion.

This study encouraged the multidisciplinary audience to undermine the imperative of universal hearing screening. The congenital inner ear malformation subgroup can be treated early to achieve a better hearing result in view of gaining normal level of speech, language ability and good quality of life.

Case Report

Case 1

An eight-year-old boy presented with fever was diagnosed with drug-resistant *Streptococcus pneumoniae* after the bacterial culture of cerebrospinal fluid. He was treated with 14 days course of intravenous antibiotics. After the resolution, hearing evaluation by an audiologist revealed the boy had right ear hearing loss. Interestingly, we learned from the interview that the boy was not aware of the onset of his right ear deafness.

One month later, the boy visited the community hospital for fever and malaise. The consulting doctor found drug-resistant *Streptococcus pneumoniae* in the cerebrospinal fluid (CSF) culture. For recurrent meningitis, the boy was sent for magnetic resonance imaging. The scans revealed inner ear

abnormality and Mondini dysplasia were suspected. The boy was given streptococcal vaccine and later referred to our hospital.

During the routine clinical examination, the boy had normal external ear and tympanic membrane. The audiogram showed a profound sensorineural hearing loss in the right ear. The computed tomography (CT) of the temporal bone has confirmed the incomplete partition type 1 (Fig. 1); however, the site of CSF leakage could not be located in the images.

After discussions with patient and his guardians, a surgical exploration of right mastoid cavity and middle ear was performed. The leakage site was located in the right lateral semicircular canal. The canal and the vestibular aqueduct were obliterated. The procedure was uneventful. The boy had been followed-up for one year with no recurrent meningitis. Later, he was referred to the local community hospital.

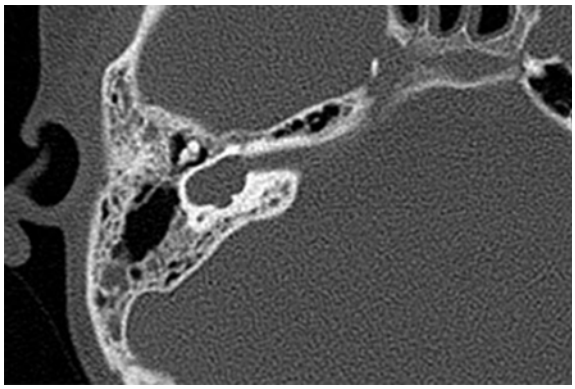

Fig. 1 The CT of right temporal bone revealed the replacement of a normal cochlea and vestibule with lobulated cavity.

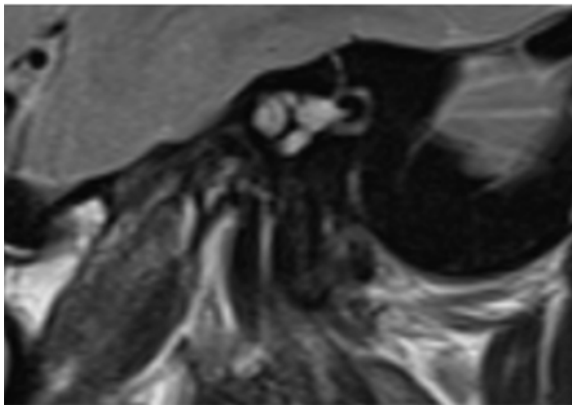

Fig. 2 The T2-weighted MRI oblique sagittal image of left temporal bone. CSF delineates the four nerves of the IAC. Cochlear nerve inferiorly to facial nerve was absent.

Case 2

An 11-year-old girl came to our hospital with the complaint of hearing difficulties in her left ear. Further discussions revealed that though she was aware of it she did not have an opportunity to consult a doctor. She did not experience any tinnitus, vertigo, otorrhea, pain or headache. Her vaccination history was complete, and she was excellent in learning at school. She had asthma for which she sometimes required hospitalization. The physical examinations were normal.

We sent her to an audiologist for the hearing test. The audiogram found profound left ear hearing loss with normal tympanogram in both ears. However, magnetic resonance imaging of the temporal bone revealed the small size of the left internal acoustic canal (IAC), absence of left cochlear nerve and the small size of left vestibular nerve. The right vestibulocochlear nerve and bilateral facial nerve were normal. The right cochlea and vestibule were also normal (Fig. 2).

This patient was given information about the cause of her hearing loss and how to manage her daily life activities. The possibility of the brain stem implant was also discussed in case of profound hearing loss in the other ear.

Case 3

A seven-year-old girl presented with a three-year history of progressive right ear, hearing loss. There were no associated complaints of otorrhoea, tinnitus, vertigo, facial palsy or head trauma. Otoscopic examination revealed normal findings. On audiometric assessment, pure tone audiogram showed right profound sensorineural hearing loss with a hearing threshold above 120 dB HL on pure tone average. The left ear showed a sensorineural hearing loss with hearing threshold 45 dB HL. Regarding speech audiometry, the right ear presented speech reception threshold above 110 dB HL. The discrimination cannot be tested due to her inability to follow a command. On the left ear, she had a speech reception threshold of 40 dB HL and discrimination level of 80%. Tympanogram revealed type A with absent acoustic reflex on both sides. CT scan revealed a common cavity of the vestibule and cochlea in the right inner ear. On the left inner ear, it has a complete separation of the vestibule and cochlea, but a small and incomplete turn of the cochlea. Cochlea hypoplasia was suggested on the left side (Fig. 3). Fortunately, she has residual serviceable hearing on

the left side. Therefore, she underwent rehabilitation with a hearing aid.

Case 4

A 13-year-old girl has a history of delayed speech and language development at the age of two-and-a-half years. Other developmental milestones were normal. There was no family history of speech or language disorders. Physical examination revealed diffuse enlargement of the thyroid gland. Otologic examinations were normal in both ears. For the hearing test, visual reinforcement audiometry responded to sound at 65-70 dB HL. Tympanometry showed type A on both sides. Wave five of the auditory brainstem response test could be identified down to 65 and 75 dB HL in the right and left ear, respectively. The threshold of auditory steady-state response test could be identified at 2.5 to 4 kHz.

The thyroid function test showed euthyroid, TSH 2.35 uIU/ml (normal range 0.54-3.7), FT4 0.88 pg/ml (normal range 0.7-1.8). CT scan revealed enlargement of the bilateral vestibular aqueducts, measuring diameter about 2.6 mm on the right and 2.5 mm on the left side. Associated enlargement of the bilateral upper vestibules was observed. Bilateral cochlea showed only 1.5 turns, normal basal turn with cystic apex (Fig. 4).

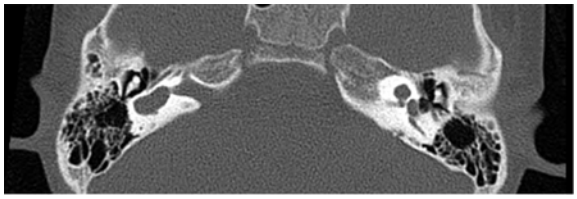

Fig. 3 The CT showed a cystic cavity on the right ear. On the left ear, the cochlea and vestibule were smaller than normal with incomplete turn of left cochlea.

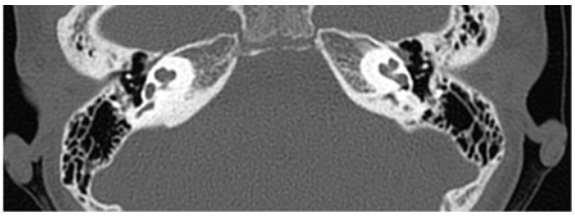

Fig. 4 The CT showed classic Mondini deformity. One and a half turn of cochlea were observed on both ears. The apical and middle turns coalesce to a cystic formation at apex. A dilated vestibule and enlarged vestibular aqueduct were noticed.

She underwent rehabilitation with a hearing aid. Unfortunately, she got a sudden sensorineural hearing loss (SSNHL) on the left side with 90 dB HL pure tone audiogram during rehabilitation. The cause of SSNHL could not be identified. Therefore, we treated her with pulse-methylprednisolone, but the result showed poor response. It developed into profound sensorineural hearing loss with 118 dB HL on the right side and 112 dB HL on the left side. She had some challenges in using hearing aids for communications. Therefore, cochlear implant was placed on her left ear. We performed a cochleostomy just anteroinferiorly to the round window. Subsequently, the multiple channels implant was inserted with all electrodes. There were no complications, including CSF gusher and facial nerve injury. She attended the hearing rehabilitation course after surgery. On her last visit with us, her speech reception threshold was 25 dB HL and the speech discrimination at presentation level of 50 dB HL was 96%.

Discussion

Case 1: incomplete partition type 1 with recurrent meningitis

The osseous labyrinth malformations are uncommon and counting for approximately 20% of the cases with congenital sensorineural hearing loss[11]. Jackler et al[11] has described these malformations on the basis of embryonic arrest during the development of the inner ear. The authors classified an osseous labyrinth malformation to four types including complete cochleovestibular aplasia (Michel deformity), common cavity, hypoplastic cochlea and incomplete partition (Mondini deformity).

Mondini was the first author who described the inner ear deformity. His classic presentation consisted of: 1) a cochlea of one-and-one-half turns instead of the normal two-and-one-half turns, comprising a normal basal turn and a cystic apex in place of the distal one-and-one-half turns; 2) an enlarged vestibule with normal semicircular canals; and 3) an enlarged vestibular aqueduct containing a dilated endolymphatic sac[12-14]. We called this deformity as classic Mondini dysplasia. The Mondini dysplasia was associated with the following syndromes: Pendred syndrome[15], CHARGE syndrome[16,17], Klippel-Feil syndrome[18], DiGeorge syndrome[19] and Wildervanck syndrome[20].

The latest classification system was proposed by Sennaroglu and Saatci[21]. Their concept relied on an embryogenic arrest at different stages of

development. The authors suggested that the cystic cochleovestibular anomaly was a severe form of incomplete partition (incomplete partition type 1; IP-I) while classic Mondini deformity was considered as incomplete partition type 2 (IP-II). The IP-I has a cystic dilated vestibule accompanied with the cystic and empty cochlea. This pathology represents a form of the common cavity that is one-step more classified.

Recurrent bacterial meningitis was defined as two or more episodes of meningitis that are separated by a period of convalescence and the complete resolution of all signs, symptoms and laboratory findings[22,23]. Recurrent bacterial meningitis is rare but has a high mortality and morbidity rate[23].

We present a case of an 8-year-old boy with unilateral hearing loss and recurrent bacterial meningitis. This case did not have any visible craniofacial abnormalities. During the investigations for recurrent meningitis, the cystic formation in the cochlea and vestibular dilatation, consistent with the incomplete partition of the inner ear, were detected by the imaging study of the temporal bone.

This patient was scanned by MRI as the first imaging test. We believed that the pediatrician who ordered this test might want to investigate additional intracranial lesions that can be addressed by an MRI. However, the CT of the temporal bone is the imaging of choice for inner ear malformation.

The linkage of recurrent meningitis and incomplete partition of the inner ear was reported in this study. This report was consistent with other literature. Inner ear malformation can cause spontaneous CSF fistulae and meningitis[12,24]. Communication between the middle ear and CSF spaces may result in bacterial translocation, with the most common infecting organism being *Streptococcus pneumoniae*[25-27]. This organism was also found in our cases.

The treatment options included the CSF leakage repair. This can be achieved by transmastoid, middle fossa craniotomy or combined approaches[28]. The fistula could be repaired by the autologous tissue such as fascia, fat, bone and bone dust depending on the size of it. Mastoid obliteration is frequently done after the fistula repair. There was a variation using partial obliteration technique[29] and the variation depended on the surgeon's preferences. The cochlear implant is the treatment of choice for hearing loss for this particular scenario. The hearing aid can be used if the patient has residual hearing[30,31] and the hearing protections to preserve the residual hearing is routinely recommended.

Case 2: cochlear nerve deficiency

Cochlear nerve deficiency (CND) is referred to clinical diagnosis of profound hearing loss associated with imaging study which reveal no visible cochlear nerve[32-34]. An auditory brainstem response test would confirm diagnosis CND with cochlear microphonic in the absence of neural waves[35].

Govaerts et al[36] proposed the classification of hypoplasia and aplasia of the cochleaovestibular nerve on imaging as follows: type 1 affected cochleovestibular nerve in which the labyrinth may be normal or dysplastic and the internal auditory canal is stenotic; type 2a affected cochlear branch with labyrinth dysplasia, in which the labyrinth dysplasia ranges from a minor dysplasia, like in case 3, to a common cavity; type 2b affected cochlear branch with normal labyrinth; and type 3 affected vestibular branch as an isolated aplasia.

Labyrinthine development commences at approximately the third week of gestation with the formation of the otic placode that will become the otic vesicle. At the seventh week, the spiral organ of Corti develops from the cochlear duct, with fibers from the spiral ganglia forming the cochlear nerve. Simultaneous development of the ampulla with vestibular ganglia results in formation of the vestibular branches of the vestibulocochlear nerve[37].

At approximately the ninth week of gestation, the mesenchyme surrounding the otic vesicle begins to chondrify and will form the otic capsule that ossifies. The bony labyrinth is routinely identified on CT scans as the hyperdense bone surrounding the inner ear structures. The IAC is formed by inhibition of cartilage formation at the medial aspect of the otic vesicle. This inhibition requires the presence of the vestibulocochlear nerve. In the absence of the nerve, a canal will not be formed[38]. Not only does the presence of the vestibulocochlear nerve allow the formation of the IAC, but survival and promotion of the nerve seem to require the presence of a growth factor from the otic vesicle[39].

This study reported a case with the absence of left cochlear nerve diagnosed by MRI. The absent of cochlear nerve is an absolute contraindication for cochlear implant[40] but there was a dilemma for cochlear nerve deficiency. Much literature found that the cochlear implantation may help the patients to develop speech understanding and production, but other literature came up with negative results[41-44]. The benefits from the

cochlear implant were extremely variable, ranging from sporadic cases in which open set speech perception and acquisition of a spoken language were achieved, to most cases in which only an improved access to environmental sound was developed[42]. Further studies are needed to answer this dilemma.

Case 3: common cavity and hypoplasia cochlea malformation with progressive hearing loss

A common cavity deformity is a deformed inner ear in which the cochlea and vestibule are confluent forming a common rudimentary cystic cavity[45]. This patient had a congenital common cavity of the right cochlea and hypoplasia of the left cochlea. She presented with the progressive hearing loss in her right ear for three years. This is atypical for the common cavity cochlea malformation, which is a congenital onset. We are aware that the progressive hearing loss may not cause by the common cavity itself. There was, however, no infection or ototoxic drugs history of this patient. We also conducted the blood test for the syphilis and autoimmune diseases with negative results.

Cochlear implantation may have a role in bilateral common cavity malformation[45,46]. This patient was not a candidate for cochlear implantation because she had residual hearing (moderate level of hearing loss) on her left ear, which can help with the hearing aids.

Case 4: Pendred's syndrome with Mondini's dysplasia

Pendred syndrome is one of syndromic deafness diseases. It has a problem related to thyroid hormone synthesis resulting in decreased thyroid hormone. The most important clinical symptom is bilateral congenital sensorineural hearing loss. However, the degree of hearing loss is variable among individual patients and it may present at birth or appear gradually[47]. Pendred syndrome usually has a vestibular aqueduct enlargement with or without an inner ear anomaly. A typical inner ear anomaly for Pendred syndrome is a classic Mondini deformity.

This case presented with the thyroid goiter, which is a classical finding of Pendred syndrome. The goiter results from the disorder of iodide organification due to mutation of the SLC26A4 gene code for the Pendrin protein. In the early phase, the goiter enlarges diffusely and subsequently develops into a multiple nodular goiter. Additionally, thyroid goiter in Pendred syndrome has been reported that may develop to thyroid cancer[48]. Manifestations of goiter can be varying in the degrees. However, obvious clinical manifestations usually presented during adolescent, between the ages of 20 and 30 years[49]. Most of the patients presented with euthyroid goiter[47]. A perchlorate discharge test has been introduced to diagnose Pendred syndrome, but it is not a specific diagnostic test[49]. The gold standard test is genetic testing to detect PDS mutations. However, it is not feasible to perform in most institutions. Therefore, clinical manifestations and radiologic examinations are considered the best modalities to diagnose Pendred syndrome in clinical practice.

This case was treated successfully with the cochlear implant. Kontorinis et al reported the outcomes of cochlear implantation in Pendred syndrome[31]. They studied in five cochlear implant recipients with four unilateral and one bilateral implantation. All of them enjoyed a high level of open-set speech recognition with satisfactory hearing outcomes.

Conclusion

We identified a subgroup of the congenital craniofacial anomaly patients that solely had inner ear malformations. Our case series would help to increase the awareness of this condition for the multidisciplinary congenital, craniofacial anomaly care team.

What is already known on this topic ?

The congenital craniofacial anomaly may not present with a visible external anomaly.

An inner ear malformation was associated with multiple syndromes.

What this study adds ?

This subgroup of congenital craniofacial, anomaly patients usually comes to see the doctor too late to get the benefits from a cochlear implant and hearing therapy.

The authors recommend universal hearing screening.

The CT scan of the temporal bone is the imaging of choice in children presenting with meningitis following acute otitis media, or sudden or undetermined period of sensorineural hearing loss.

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Potential conflicts of interest

None.

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ผู้ป่วยหูชั้นในผิดปกติแต่กำเนิด: การศึกษาในผู้ป่วยหลายรายและมีการทบทวนวรรณกรรม

ภาธร ภิรมย์ไชย, พรเทพ เกษมศิริ, พนิดา ธนาวิรัตนานิจ, ขวัญชนก ยิ้มแต้

ผู้ป่วยที่มีความผิดปกติของกะโหลกศีรษะและใบหน้าแต่กำเนิดมักถูกส่งต่อหรือมาพบแพทย์เร็ว เนื่องจากความผิดปกติส่วนใหญ่นั้นมองเห็นได้ชัด ทำให้ได้รับการตรวจหูและประเมินการได้ยินแต่เนิ่นๆ ทุกราย แต่ยังมีผู้ป่วยส่วนน้อยซึ่งความผิดปกติภายนอกไม่ชัดเจนหรือไม่มีความผิดปกติของหูที่มองเห็นจากภายนอก มักมาพบแพทย์ช้าหรืออาจมาพบแพทย์ด้วยปัญหาอื่น เช่น พูดช้าเป็นต้น มักจะเสียโอกาสในการตรวจการได้ยิน การศึกษานี้มีวัตถุประสงค์เพื่อรวบรวมกลุ่มผู้ป่วยที่มีความผิดปกติเฉพาะหูชั้นใน โดยได้เสนออาการและการตรวจร่างกายทางคลินิก ผลการตรวจทางห้องปฏิบัติการ การรักษาและผลการรักษา รวมถึงวิจารณ์โดยอิงหลักฐานเชิงประจักษ์