

CT and MR Imagings of Semicircular Canal Aplasia¹

Chung Hee Shin, M.D., Hyun Sook Hong, M.D., Beom-Ha Yi, M.D., Jang-Gyu Cha, M.D.,
Seong-Jin Park, M.D., Dae-Ho Kim, M.D., Hae Kyung Lee, M.D., Shi-Chan Kim, M.D.²

Purpose: To evaluate the clinical, CT and MR imaging findings of semicircular canal (SCC) aplasia and to evaluate if a correlation exists between these findings and the associated anomalies or syndromes.

Materials and Methods: This study retrospectively reviewed the CT and MRI findings of five patients with SCC aplasia. The CT and MR findings were analyzed for SCC, direction of facial nerve canal, cochlea, vestibule, oval or round window, middle ear ossicles, and internal auditory canal (IAC).

Results: The subjects included three boys and two girls ranging in age from one to 120 months (mean age; 51 months). Four of the subjects had the CHARGE syndrome, and one had the Goldenhar syndrome. Moreover, four subjects had sensorineural hearing loss and one had combined hearing loss. The course of the facial nerve canal was abnormal in all five cases. Moreover, trapped cochlea and dysplastic modiolus were each observed in one case. Four subjects had atresia of the oval window; whereas ankylosis of the ossicles was present in three subjects. IAC stenosis was present in one patient with the CHARGE syndrome.

Conclusion: The aberrant course of the facial nerve canal, atresia of the oval window, and abnormal ossicles were frequently associated in patients with SCC aplasia. In addition, the Goldenhar and CHARGE syndromes were also commonly associated syndromes.

Index words : Ear, inner

Congenital, semicircular canals

Deafness

Tomography, X-ray computed

Magnetic resonance (MR)

¹Department of Radiology, Soonchunhyang University Bucheon Hospital

²Department of Otorhinolaryngology, Soonchunhyang University Bucheon Hospital

Received October 23, 2008 ; Accepted April 7, 2009

Address reprint requests to : Hyun Sook Hong, M.D., Department of Radiology, Soonchunhyang University Bucheon Hospital, 1174 Jung dong, Wonmi-gu, Bucheon-si, Gyeonggi-do 420-767, South Korea.

Tel. 82-32-621-5851-2 Fax. 82-32-621-5874 E-mail: hshong@schbc.ac.kr

The reported incidence of significant sensorineural hearing loss (SNHL) in young children varies from 1:1000 to 1:2000 depending on the populations studied. Approximately 20% of patients with congenital SNHL also have radiological anomalies of the inner ear (1-4). The most common radiologically detected inner ear anomalies include those associated with the cochlea, the SCC and the vestibular aqueduct. Many cases have abnormalities of more than one portion of the inner ear.

Semicircular canal (SCC) dysplasia is defined as a malformation with hypoplasia or aplasia of one or all of the semicircular canals. Dysplasia of the lateral SCC is a common inner ear malformation. Further, 40% of cochlear malformations are accompanied by dysplasia of the lateral SCC. Jackler *et al.* introduced a hypothesis for SCC aplasia that is based on an arrest of embryogenesis during the sixth week of gestation. During this time, the SCCs develop as evaginations of the vestibular appendage, and the associated anomalies of these structures are common and variable (5, 6). SCC aplasia has been reported in both syndromic and non-syndromic cases (5-11).

CT and MR imaging represent non-invasive modalities that can be used for the detection of ear malformations. These imaging modalities give complementary information; the CT depicts bony details whereas the MR shows the fluid filled inner ear and facial nerve. Recently, a heightened awareness of ear anomalies, combined with the greater sensitivity of CT and MR imaging has provided greater detection of these anomalies (2).

The objectives of this study are to review the clinical and radiological findings of SCC aplasia and to determine the associated anomalies and/or syndromes.

Materials and Methods

We retrospectively reviewed five patients with SCC aplasia who presented with congenital hearing loss from 2001 to 2005. A high resolution CT of the temporal bone ($n=5$) was performed using contiguous 1-mm-thick axial sections made parallel to the infraorbital-meatal line or reconstructed coronal images reconstructed with a 512×512 matrix, a 1.0 mm collimation, and a 10 cm field of



Fig. 1. One-month-old boy with the CHARGE syndrome (A) The SCCs are not visualized at the level of the vestibule (arrowheads). Normal cochlea (white arrow) and modiolus (black arrow) are seen. (B) The left IAC (arrow) diameter is smaller than 2 mm, consistent with IAC stenosis. The lateral semicircular canal is not seen in this section at the level of the vestibule (arrowheads). (C) The modiolus is well defined with a low signal in the axial 3D T2-weighted FSE images with 1 mm slice thickness (black arrow). T2-weighted images showed high-signal labyrinthine fluid with low-signal bony detail of the osseous spiral lamina (white arrow) bisecting the cochlear turns.

view (GE HiSpeed CT/i Pro[®], Milwaukee, USA / Siemens Somatom sensation 16[®], Erlangen, Germany).

A temporal MR ($n=3$) was performed using a 1.5-T unit (GE 1.5T SIGNA[®], Milwaukee, WI., U.S.A.). The pulse sequences used were as follows: a routine brain axial T2-weighted images (T2-WI) and FLAIR, a temporal 3D FSE T2-WI with a 1 mm slice thickness, a temporal 3D FIESTA with three reconstructed axial, a coronal and both sagittal images, and a 1.4 mm slice thickness with a 0.7 mm interslice gap, and contrast enhanced T1-WI with 3 mm slice thickness. Both CT and MR scans were performed in two of the cases.

Image interpretation

Each of the CT and MR images were analyzed for aplasia of the SCC, direction of the facial nerve canal, patency of the cochlear aperture, presence of cochlear

anomalies, large vestibular aqueduct, atresia of the oval or round window, dysplasia or fusion of the middle ear ossicles, and the size of the internal auditory canal (IAC).

Aplasia of the SCC was diagnosed when the canals were completely absent and an isolated vestibule was present. The cochlear aperture was regarded as normal if the patent was visible on an MR, when available, or if trapped when a bony bar over the aperture was present. The vestibular aqueduct (VA) was considered enlarged when the diameter of the VA exceeded that of the posterior SCC or when the width of the VA at the midpoint between the common crus and its external aperture was larger than 1.5 mm. Atresia of the oval window was defined by a bony plate superimposed between the vestibule and middle ear. Dysplasia was noted if the ossicles demonstrated an abnormal morphology or size. The criteria for fusion were based on the proximity and

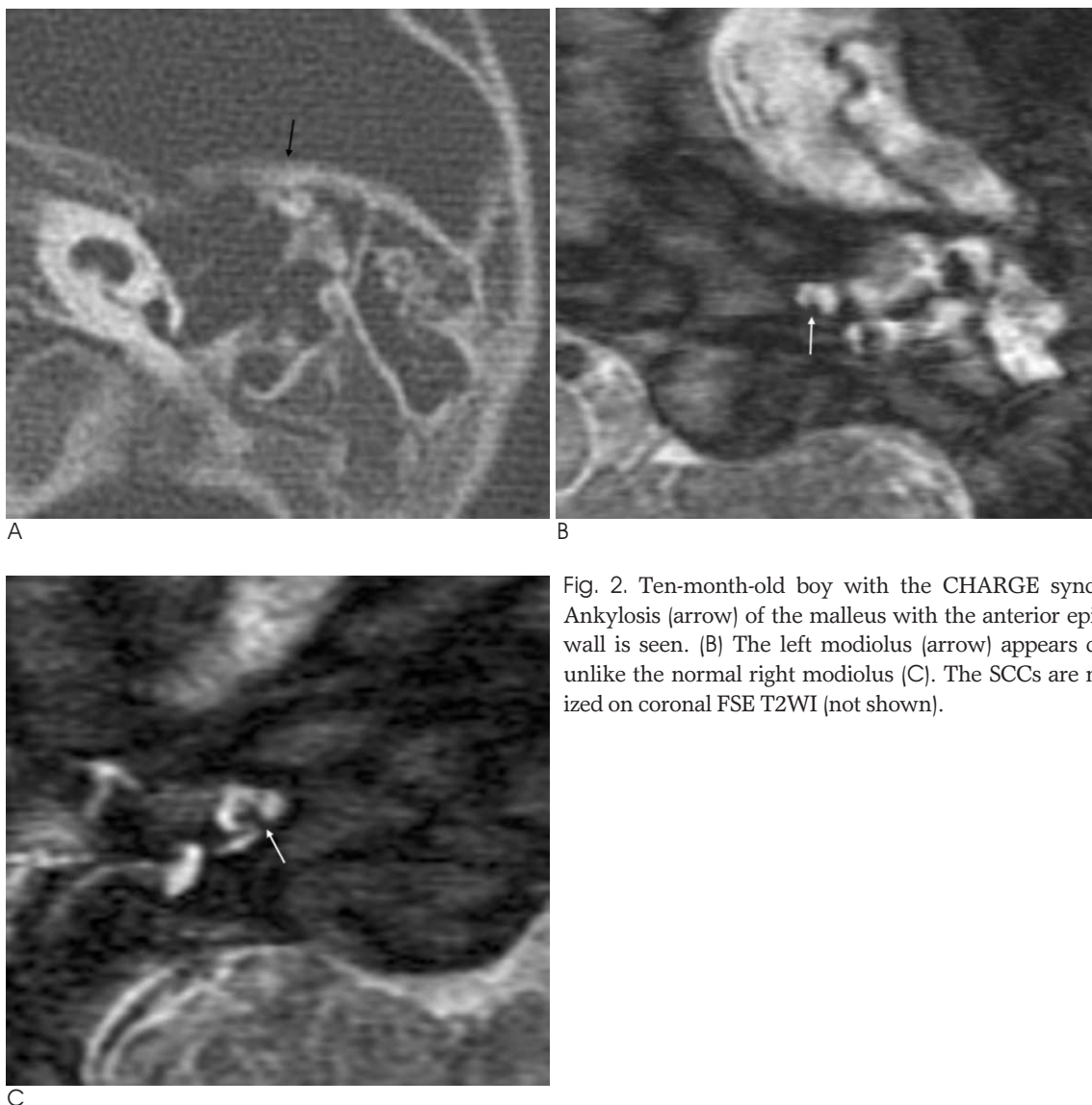


Fig. 2. Ten-month-old boy with the CHARGE syndrome (A) Ankylosis (arrow) of the malleus with the anterior epitympanic wall is seen. (B) The left modiolus (arrow) appears dysplastic, unlike the normal right modiolus (C). The SCCs are not visualized on coronal FSE T2WI (not shown).

appearance of ankylosis to the wall of the epitympanum or interossicular fusion. The IAC was considered stenotic when the size of the canal was less than 2 mm.

Results

The five patients included three boys and two girls. The patients' ages ranged from one to 120 months (mean age; 51 months). All patients had bilateral SCC

aplasia (Fig. 1A) that was presented as hearing loss. Associated anomalies were variable and included one case of GOLDENHAR syndrome and four cases of CHARGE syndrome. Four of the five patients had SNHL, whereas one had a combination of SNHL and conductive hearing loss. The clinical findings for the patients with recognized syndromes are summarized in Table 1.

The course of the facial nerve canal was abnormal in

Table 1. Clinical Information: Five Patients with Aplasia of the Semicircular Canals

No.	Age	Sex	Anomalies	Clinical Findings/Operation	Type of Hearing Loss	Modality
1	1 m	M	CHARGE syndrome Coloboma Growth retardation CHD	Polydactyly, SOM / not performed	SNHL	CT, MR
2	47 m	F	Goldenhar syndrome Cleft palate Facial palsy	Hearing aids / not performed	SNHL	CT
3	10 m	M	CHARGE syndrome Coloboma CHD Growth retardation Micropenis Cryptorchism	SOM /Cochlear implantation VII nerve exposed to promontory	SNHL	CT, MR
4	10 y	F	CHARGE syndrome Developmental delay	Short stature, / not performed	SNHL & CHL	CT
5	6 y	M	CHARGE syndrome CHD Developmental delay	Hearing aid /Cochlear implantation Bifurcation of tympanic segment	SNHL	CT, MR

m: month, y: year, F: female, M: male, SOM: serous otitis media
SNHL: sensorineural hearing loss, CHL: conductive hearing loss
CHD: congenital heart disease

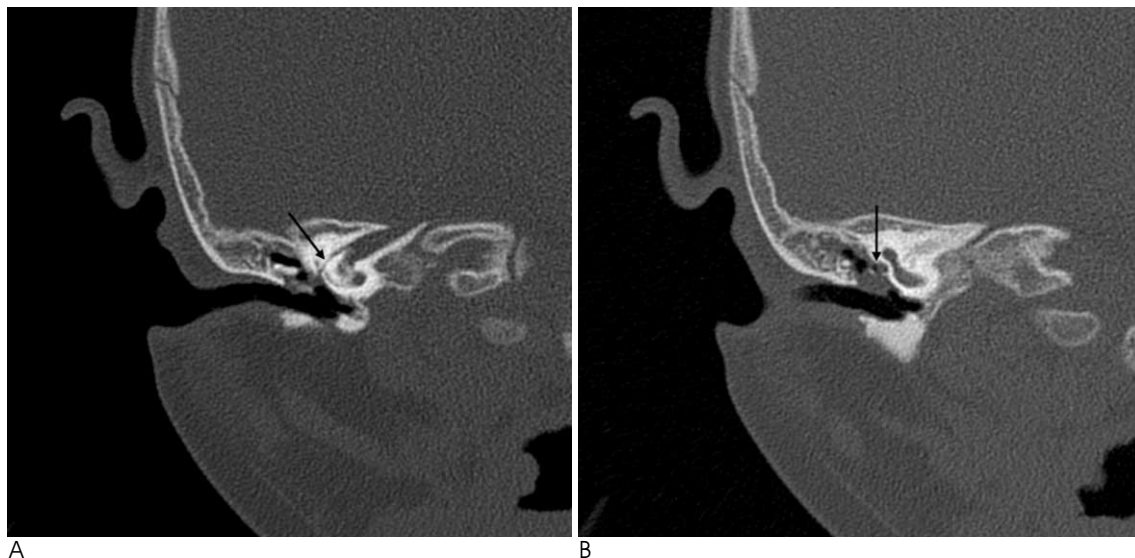


Fig. 3. Six-year-old boy with the CHARGE syndrome (A, B) The facial nerve canal (arrow) is situated over the promontory and slightly displaced medially.

all patients; in the inferomedial direction in three patients and in the medial direction in two (Fig. 3). A trapped cochlea (Fig. 4A) or dysplastic modiolus (Fig. 2B) was present in one patient for case each. The vestibular aqueduct could not be identified in any of the patients. Four of the five patients had atresia of the oval window (Fig. 4B, 5B), and could not be identified in one patient. Ankylosis of the ossicles (Fig. 2A, 5A) was noted in three cases. IAC stenosis (Fig. 1B) was present in a patient with the CHARGE syndrome. The middle ear cavity was small in one case and large in another case.

Discussion

Five to 15% of congenitally deaf individuals have radiological anomalies of the inner ear (1-4). The clinical manifestations and natural history of these deformities

are variable. The majority of the combined osseous and membranous malformations appear to develop from a premature arrest of the development of one or more components of the inner ear. As a general rule, the earlier the developmental arrest, the more severe the deformity and the more impaired the hearing (3). Whereas, some inner ear malformations involve only one portion of the inner ear, many of the patients examined in this study had a combination of anomalies involving more than one component. Between the fourth and fifth weeks of gestation, the spherical otocyst develops three buds that ultimately form the cochlea, SCCs, and the vestibular aqueduct. An inner ear malformation may be limited to one of these anlagen, may involve a combination of two of them, or may affect all three (3). The bony SCCs develop from the sixth week of gestation and are complete by week 22. The superior SCC is completed

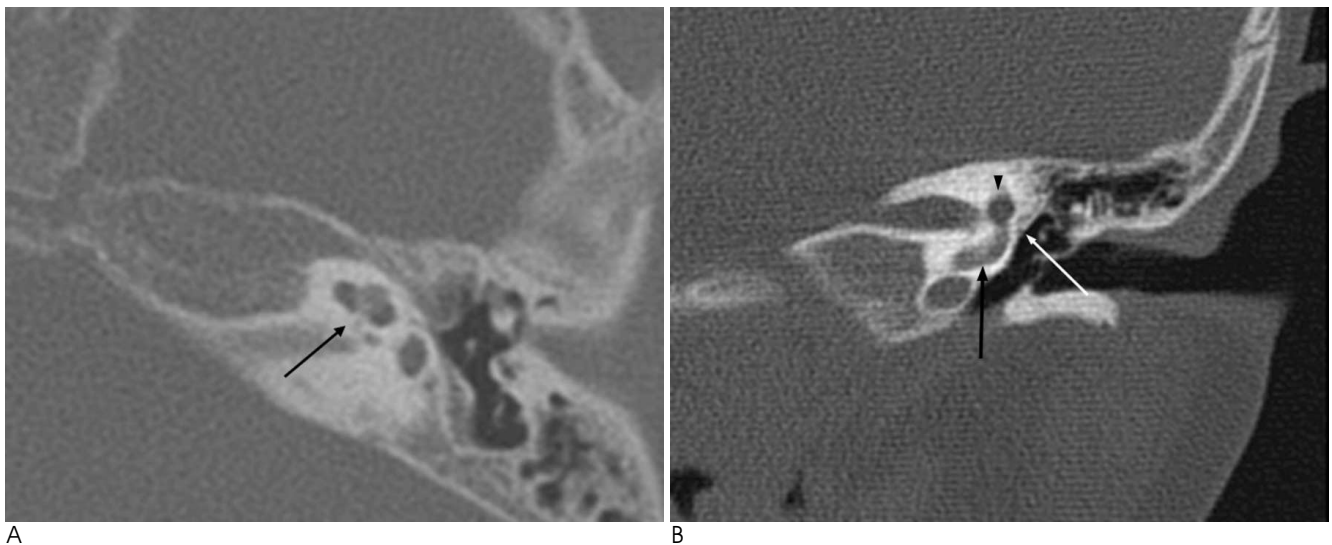


Fig. 4. Forty seven-month-old girl with the Goldenhar syndrome (A) On axial CT, a trapped left cochlea (arrow) is noted at the level of the cochlea. (B) Coronal image shows oval window atresia on the left (white arrow). The basal turn of the cochlea (black arrow) and vestibule (arrowhead) are normal.

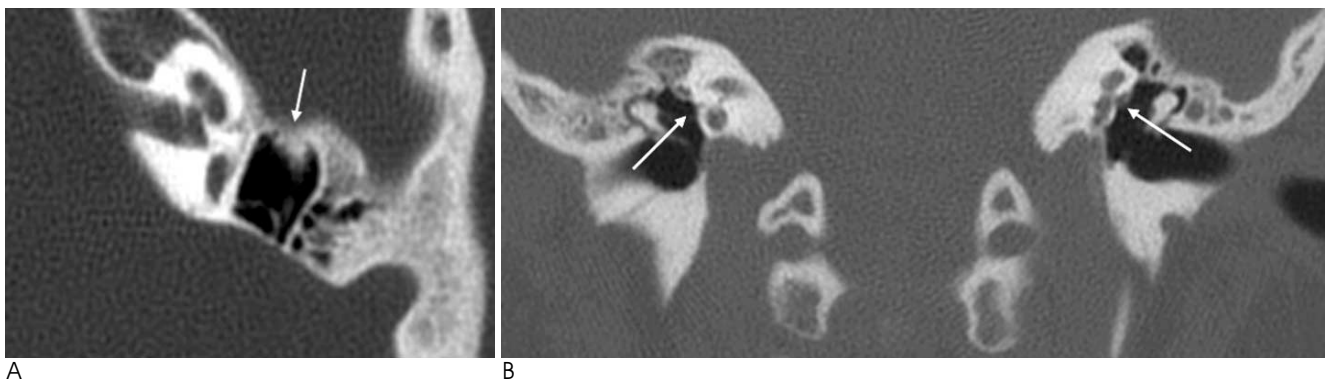


Fig. 5. Ten-year-old girl with the CHARGE syndrome (A) An axial CT demonstrates ankylosis of the malleus with the anterior epitympanic wall (arrow). (B) Coronal CT scan shows atresia of the oval windows bilaterally (arrows).

first, followed by the posterior, and finally the lateral portions. These are the body's organs for balance as well as detecting acceleration in the three perpendicular planes.

Admiraal *et al.* (6) introduced the acronym CHARGE for the association of the anomalies: coloboma (C), heart defects (H), atresia choanae (A), retardation of growth and/or development (R), genital hypoplasia (G), ear anomalies (E), and/or hearing impairment or deafness. The diagnosis of the CHARGE association is generally based on the presence of at least four of the six cardinal features, provided that these include coloboma and/or choanal atresia. Several investigators have described various ear anomalies associated with the CHARGE syndrome (4, 5). The ossicular abnormalities in the CHARGE syndrome (generally of the long process of the incus and the stapes) appear to result primarily from the abnormal development of the second branchial arch (6–8). SCC aplasia is one of the characteristic findings of the CHARGE syndrome.

Goldenhar's syndrome is a clinically heterogeneous disorder. In its most typical presentation, it is characterized by hemifacial microsomia in association with other anomalies, including vertebral defects and epibulbar dermoids. In addition, bilateral involvement of the craniofaces is known to occur; however, one side is usually more severely affected than the other. Cardiac, renal, anal, and CNS malformations have been reported to occur in patients with Goldenhar's syndrome. Marc *et al.* described a patient with Goldenhar's syndrome (oculoauriculo-vertebral dysplasia) and unilateral aplasia of all semicircular canals (9). Oculoauriculo-vertebral, VATER, and CHARGE might be related by a common pathogenetic mechanism resulting in disturbed neural crest development (7). Semicircular canal dysplasia can also be found in isolation or in conjunction with other syndromes including Waardenburg's syndrome and the Noonan syndrome (5, 10–11, 13). Posterior SCC is selectively affected in the Alagille syndrome, and the superior SCC is abnormal in cases with prenatal thalidomide exposure (5, 11, 12, 14, 15).

It is possible that secondary to the aplasia of the SCC, the surrounding structures become displaced. Some reports suggest that facial palsy may be a reliable predictor of SNHL (8). In one of our cases with a cochlear implant, the facial nerve was situated over the promontory and slightly displaced medially (Fig. 5). It is essential for the radiologist to accurately describe the entire course of the facial nerve canal, especially if a cochlear implantation

procedure is going to be performed, to minimize the possibility of iatrogenic facial nerve damage. Atresia of the oval or round window and hypoplastic cochleae can be problematic for cochlear implants (12).

CT scanning provides information about the size of the tympanic cavity, presence and nature of ossicular anomalies, presence of atresia of the oval and round windows, and an anomalous course of the facial nerve canal. The MRI is preferred for the evaluation of the presence of fluid within the membranous labyrinth prior to cochlear implantation, and shows the cochlear morphology associated with deficient septation and malformations of the modiolus, in addition to the size of the cochlear division of the vestibulocochlear nerve. The MRI and CT are complementary techniques.

Fibrous or calcified obliteration of the SCC in cases of labyrinthine ossificans is differentiated by the presence of SCC aplasia. In patients with labyrinthine ossificans, a history of SNHL, after an episode of meningitis, as well as a normal internal auditory canal and the normal course of the facial nerve canal can help with the diagnosis. The cochlea is also obliterated with involvement of the SCC in cases of labyrinthine ossificans.

The course of the facial nerve canal was abnormal in all cases. Four of the five patients had atresia of the oval window. Ankylosis of the ossicles was seen in three cases. A trapped cochlea or dysplastic modiolus was each present in one case. IAC stenosis was present in one patient. Therefore, the results of this study suggest that all patients with SCC aplasia be evaluated with high resolution CT or MR imaging to determine whether an abnormal facial nerve course and cochlear aperture is present, especially if cochlear implantation is planned. Absence of the cochlear nerve would be a contraindication to cochlear implantation.

In conclusion, atresia of the oval window, abnormality of the ossicles, and an aberrant course of the facial nerve canal were frequently associated in patients with SCC aplasia. In addition, the Goldenhar and CHARGE syndromes were commonly associated syndromes in these patients.

References

1. Mafee MF, Selis JE, Yannias DA, Valvassori GE, Pruzansky S, Applebaum EL, *et al.* Congenital sensorineural hearing loss. *Radiology* 1984;150:427-434
2. Swartz JD. Sensorineural hearing deficit: a systematic approach based on imaging findings. *Radiographics* 1996;16:561-574
3. Brookhouser PE. *Sensorineural hearing loss in children*. In Cummings CW, Harker LA. *Otolaryngology - Head and Neck*

- Surgery*, 2nd ed. St. Louis: Mosby, 1993:3080-3102
4. Lane JI, Lindell EP, Witte RJ, DeLone DR, Driscoll CL. Middle and Inner Ear: Improved Depiction with Multiplanar Reconstruction of Volumetric CT Data. *Radiographics* 2006;26:115-124
 5. Satar B, Mukherji SK, Telian SA. Congenital Aplasia of the Semicircular Canals. *Otol Neurotol* 2003;24:437-446
 6. Admiraal RJ, Joosten FB, Huygen PL. Temporal bone CT findings in the CHARGE association. *Int J Pediatr Otorhinolaryngol* 1998;45:151-162
 7. Sanlaville D, Verloes A. CHARGE syndrome: an update. *Eur J Hum Genet* 2007;15:389-399
 8. Morimoto AK, Wiggins RH 3rd, Hudgins PA, Hedlund GL, Hamilton B, Mukherji SK, et al. Absent semicircular canals in CHARGE syndrome: radiologic spectrum of findings. *AJNR Am J Neuroradiol* 2006;27:1663-1671
 9. Lemmerling MM, Vanzieleghem BD, Mortier GR, Dhooge IJ, Kunnen MF. Unilateral semicircular canal aplasia in Goldenhar's syndrome. *AJNR Am J Neuroradiol* 2000;21:1334-1336
 10. Oysu C, Oysu A, Aslan I, Tinaz M. Temporal bone imaging findings in Waardenburg's syndrome. *Int J Pediatr Otorhinolaryngol* 2001;58:215-221
 11. Koch BL, Goold A, Egelhoff J, Benton C. Partial absence of posterior semicircular canal in Alagille syndrome: CT Findings. *Pediatr Radiol* 2006;36:977-979
 12. Witte RJ, Lane JI, Driscoll CL, Lundy LB, Bernstein MA, Kotsenas AL, et al. Pediatric and adult cochlear implantation. *Radiographics* 2003;23:1185-1200
 13. Parnes LS, Chernoff WG. Bilateral semicircular canal aplasia with near-normal cochlear development. two case reports. *Ann Otol Rhinol Laryngol* 1990;99:957-959
 14. Phelps PD. Congenital lesions of the inner ear, demonstrated by tomography. *Arch Otolaryngol* 1974;100:11-18
 15. Okuno T, Takahashi H, Shibahara Y, Hashida Y, Sando I. Temporal bone histopathologic findings in Alagille's syndrome. *Arch Otolaryngol Head Neck Surg* 1990;116:217-220

세반고리관 무형성증의 CT와 MRI 소견¹

¹순천향대학교 부천병원 영상의학과

²순천향대학교 부천병원 이비인후과

신충희 · 홍현숙 · 이범하 · 차장규 · 박성진 · 김대호 · 이해경 · 김시찬²

목적: 세반고리관 무형성증의 임상 및 CT와 MRI 소견을 분석하고, 동반된 기형이나 증후군을 알아보려고 하였다.

대상과 방법: 세반고리관 무형성증을 보인 5예의 CT 및 MR 소견을 후향적으로 분석하였다. 세반고리관, 안면 신경관, 와우, 전정, 난원창, 정원창, 이소골, 내이도의 이상을 CT와 MRI 소견을 중심으로 분석하였다.

결과: 남자 3예, 여자 2예이고 평균 연령은 51개월이었다. 4예는 CHARGE, 1예는 Goldenhar 증후군을 진단받았다. 4예는 감각신경 난청이었고, 1예는 혼합성 난청이었다. 안면 신경관은 5예에서 주행의 이상을 보였다. 단힌 와우와 와우축의 이형성은 각각 1예에서 보였다. 4예에서 난원창 폐쇄가 동반되었다. 3예에서 이소골의 이상이 동반되었다. CHARGE 증후군에 동반된 1예는 내이도 협착이 동반되었다.

결론: 세반고리관 무형성증 환자들에서 안면 신경관의 주행이상, 난원창 폐쇄, 이소골 기형 등이 흔히 동반되었으며, Goldenhar 증후군과 CHARGE 증후군이 흔한 동반 기형이었다.