The Journal of International Advanced Otology

J Int Adv Otol 2022; 18(2): 183-187 • DOI: 10.5152/iao.2022.21279

Case Report

Active Middle Ear Implant in a Patient with Neurofibromatosis Type 1 and Multiple Calvarial Defects: A Case Report

Kei Kajihara[1], Akira Ganaha[1], Keiji Matsuda[2], Takeshi Nakamura[1], Tetsuya Tono[1]

[1]Department of Otorhinolaryngology-Head and Neck Surgery, University of Miyazaki, Miyazaki, Japan
[2]Matsuda ENT Miyazaki Surgi-Clinic, Miyazaki, Japan

ORCID IDs of the authors: A.G. 0000-0002-2800-8886; T.T. 0000-0001-7853-2179.

Cite this article as: Kajihara K, Ganaha A, Matsuda K, Nakamura T, Tono T. Active middle ear implant in a patient with neurofibromatosis type 1 and multiple calvarial defects: A case report. *J Int Adv Otol.* 2022;18(2):183-187.

Bony abnormalities, including sphenoid dysplasia and calvarial defects, are well recognized in patients with neurofibromatosis type 1. However, having multiple calvarial defects is rare. We present a case of a 35-year-old Japanese male patient who was referred to our hospital because of hearing loss. He was diagnosed with neurofibromatosis type 1 during early childhood. Otoscopic examination revealed a protrusion from the anterior wall of the external auditory canal that obstructed the external auditory canal. Computed tomography findings revealed multiple defects and an uneven skull surface. Large bony defects of the anterior wall of the external auditory canal were also identified bilaterally. Conductive hearing loss was caused by temporomandibular joint herniation that was obstructing the external auditory canal in both ears. An active middle ear implant was implanted in the right ear. A floating mass transducer was placed into the round window niche using a round window coupler. The active middle ear implant improved postoperative audiometric thresholds to approximately 35 dB across all frequencies. No complications occurred for up to 30 months after the operation. An active middle ear implant is a feasible and valuable option for patients with neurofibromatosis type 1 and conductive hearing loss due to multiple skull defects that result in temporomandibular joint herniation.

KEYWORDS: Neurofibromatosis type 1, multiple defects of skull, temporomandibular joint herniation, active middle ear implant

INTRODUCTION

Neurofibromatosis type 1 (NF1) is a progressive autosomal dominant disorder mainly characterized by café-au-lait macules, neurofibromas, axillary freckling, optic gliomas, Lisch nodules, and skeletal dysplasia.[1] Bony abnormalities, including scoliosis, sphenoid wing dysplasia, long-bone dysplasia, and osteoporosis, are well recognized in patients with NF1.[1] Sphenoid dysplasia is a common manifestation of calvarial defects,[2] but multiple calvarial defects are rare.[2-4] Temporomandibular joint (TMJ) herniation can be caused by defects in the anterior bony wall of the external auditory canal (EAC) and can result in clicking tinnitus, otorrhea, or conductive hearing loss.[5]

In this study, we present the surgical feasibility and clinical results of an NF1 patient with multiple calvarial defects and bilateral TMJ herniation who underwent an active middle ear implant (AMEI).

CASE PRESENTATION

A 35-year-old Japanese male patient was referred to our hospital because of hearing loss. He was diagnosed with NF1 during early childhood because he presented with café-au-lait spots, cutaneous neurofibroma, axillary freckling, and bony dysplasia. Otoscopic examination revealed a diffuse protrusion from the anterior wall of the EAC, which completely obstructed the EAC in both ears when the mouth was closed (Figure 1a and 1b). The obstruction was minimized when the mouth was fully opened with the retraction of the herniated TMJ anteriorly (Figure 1c and 1d). Preoperative pure-tone audiometry findings revealed a conductive hearing loss in both ears (Figure 2a and 2b). The hearing level fluctuated during mouth opening and closing movements (Figure 2a and 2b). The maximum Japanese monosyllable discrimination scores were 80% and 90% in the right and left ears (both 100 dB),

Corresponding author: Akira Ganaha, e-mail: ganaha.akira.t8@cc.miyazaki-u.ac.jp

Received: March 13, 2021 • **Accepted:** August 4, 2021
Available online at www.advancedotology.org

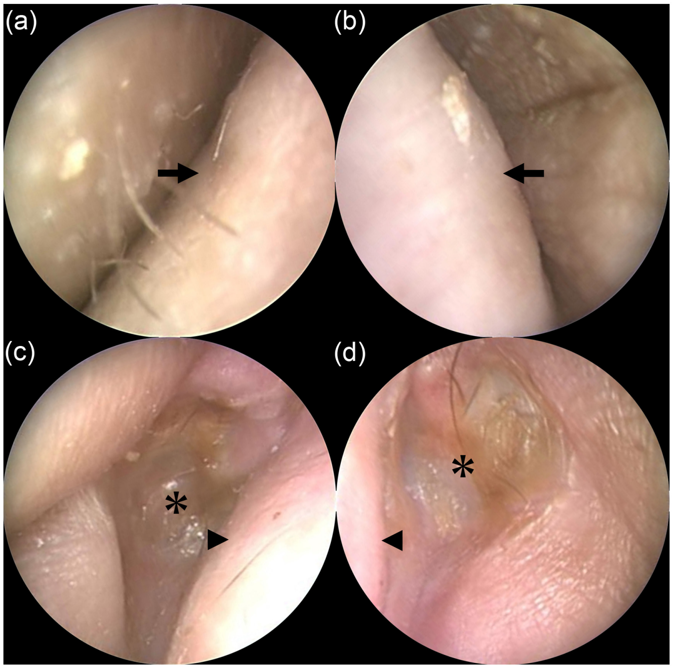

Figure 1. a-d. Preoperative otoscopic examination. Otoscopic examination in the (a) right and (b) left ear canals with the mouth closed. A diffuse protrusion of the anterior canal wall (black arrow) obstructs the canal completely. While the mouth is open, the anterior wall retracts anteriorly (black arrowhead) and the tympanic membrane (*) becomes visible in the (c) right and (d) left ears.

respectively (Figure 2c). High-resolution computed tomography (CT) scans showed multiple defects and an uneven skull surface (Figure 3A). A bony defect in the anterior wall of the EAC was identified, and the EAC in both ears had a soft-tissue lesion (Figure 4). Conductive hearing loss might have been caused by TMJ herniation due to the bony defect in the anterior wall of the EAC, leading to complete obstruction in the EAC of both ears. The patient could not use conventional hearing aids because of simultaneous ear canal wall and jaw movements. Magnetic resonance imaging findings revealed mild enlargement of the cisterna magna. The patient presented no neurological symptoms; therefore, treatment for the mild enlargement of the cisterna magna was deemed unnecessary. The patient also had no history of vertigo according to his self-reports.

An AMEI (Vibrant Soundbridge VORP 503; Med-El, Innsbruck, Austria) was implanted in the right ear because the patient noted that his hearing was better in the left ear than in the right ear. Moreover, there was a possibility that the audio processor could not be fixed on the skin using magnets because the neurofibroma had caused thickening of the skin behind the left ear. Because poor development of the mastoid cavity and narrow facial recess did not allow the standard round window approach via posterior tympanotomy (Figure 5A), the posterior bony canal wall was dissected to place the floating mass transducer on the round window membrane using a round window coupler (Figure 5B). An implant bed was created on the skull surface, and self-drilling screws were used to fix the implant. The posterior canal wall was reconstructed using conchal cartilage (Figure 5C) to prevent cable extrusion from the mastoid cavity. The cartilage was placed in the same position as the posterior canal wall to allow for cleaning of the ear canal after surgery. The AMEI audio processor (SAMBA; Med-El) was fitted 2 months after AMEI implantation. Postoperative audiometric thresholds improved to approximately 35 dB across all frequencies (Figure 2A). The patient enjoyed stable hearing improvements and sound quality with the AMEI, which were confirmed using audiological methods (Figure 2A-C). No complications occurred for up to 30 months after the operation.

DISCUSSION

Skeletal dysplasia has been reported in approximately 50% of patients with NF1.[3] The most common skeletal dysplasias are scoliosis, the absence of the greater sphenoid wing, tibial pseudarthrosis, short stature, spinal meningocele, and macrocephaly.[2] Multiple calvarial defects in patients with NF1 are rare.[2-4] An anterior bony defect of the EAC with multiple calvarial defects has not been previously reported in patients with NF1. The anterior bony defect of the EAC led to TMJ herniation in our patient. Temporomandibular joint herniation can occur in various conditions such as necrotizing external otitis, temporal bone trauma, rheumatoid arthritis of the

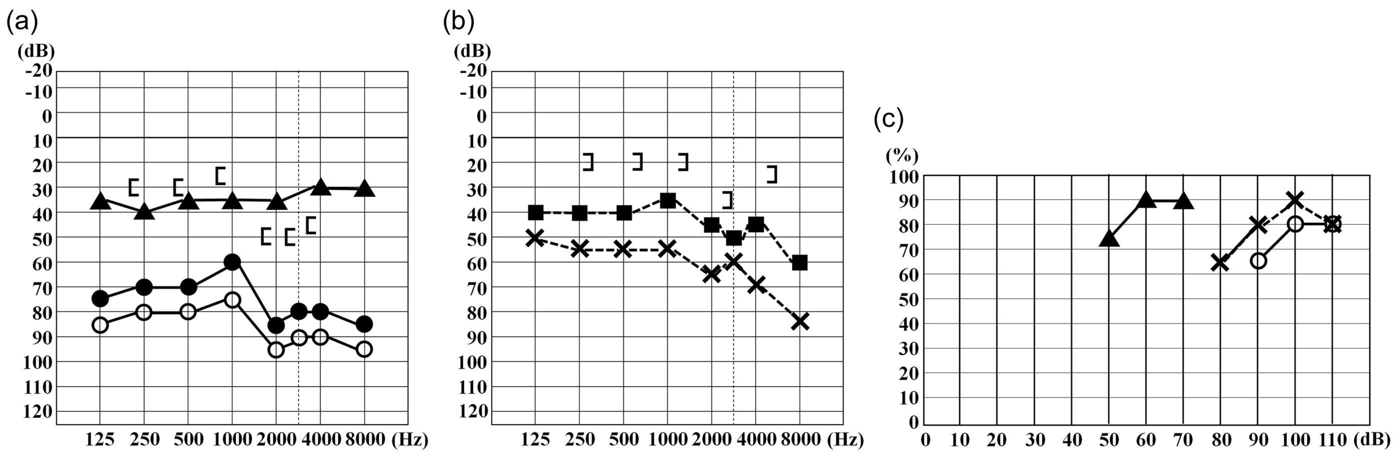

Figure 2. a-c. Preoperative and postoperative pure-tone and speech audiograms. The preoperative pure-tone audiograms of the (a) right and (b) left ears. (a) The filled circles and triangles indicate hearing when opening the mouth and postoperative air conduction aided by the AMEI, respectively. (c) The unaided preoperative speech audiograms of both ears are also presented. (b) The black squares indicate the hearing level during mouth opening. (c) The black triangles indicate the postoperative speech audiogram aided with AMEI. Uncolored circles, right ear; X, left ear; AMEI, active middle ear implant.

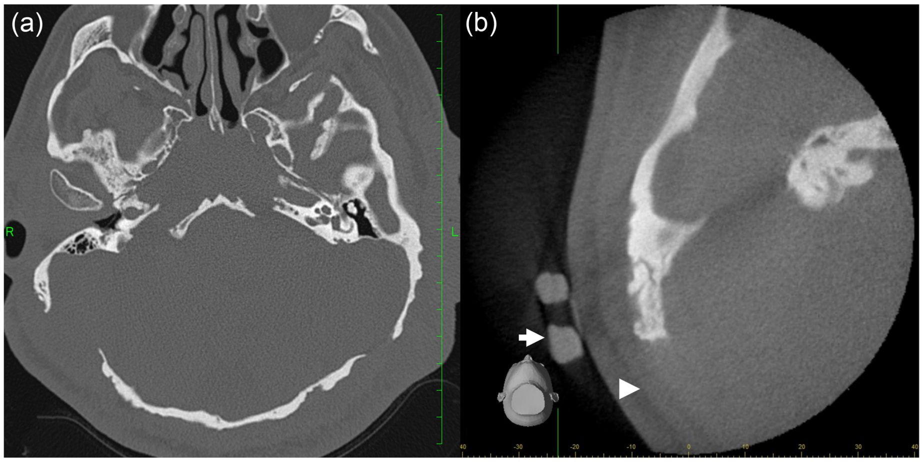

Figure 3. a,b. High-resolution computed tomography image of the skull. (a) Multiple calvarial defects with uneven skull surface can be identified in the axial computed tomography image of the skull. (b) Axial computed tomography findings of the right ear. White arrow indicates a doughnut-shaped radiation impermeable marker at the assumed implant sight of the bone-anchored hearing aid. White arrowhead indicates the bone defect of the skull. The assumed implant site of the bone-anchored hearing aid was near the bone defect of the skull with skull irregularities.

TMJ, and otologic surgery.[6] Our patient had no history of diseases that could cause TMJ herniation without NF1. Moreover, TMJ herniation in our patient obstructed the EAC, leading to conductive hearing loss in both ears.

Neurofibromatosis type 1 rarely causes conductive hearing loss when the neurofibroma invades the ear canal.[7,8] Conductive hearing loss caused by an anterior bony defect of the EAC with TMJ herniation has not been previously reported in patients with NF1. The treatment possibilities for our patient included surgical EAC reconstruction, adaptation of hearing aids by bone conduction, or implantable hearing aid adaptation.

The 2 most common approaches to correct TMJ herniations are the transcanal approach, based on the anterior wall exposure of the tympanic bone, and the preauricular approach, based on TMJ exposure.[9] In both approaches, a graft is needed to obliterate the defect in the anterior canal wall. Surgical reconstruction of the anterior bony EAC using cartilage, polyethylene, and titanium meshes as graft materials have been described as treatment options for TMJ herniations.[10] Autologous grafts such as tragal cartilage may undergo a degree of resorption, which may lead to recurrence, whereas synthetic materials present a risk of suppuration and extraction.[10]

The materials for the anterior canal wall reconstruction were placed between the anterior canal bone and TMJ capsule.[11] The inserted material was supported by the remaining anterior canal wall. The conventional reconstruction method was inapplicable in our patient because the overall anterior canal wall had

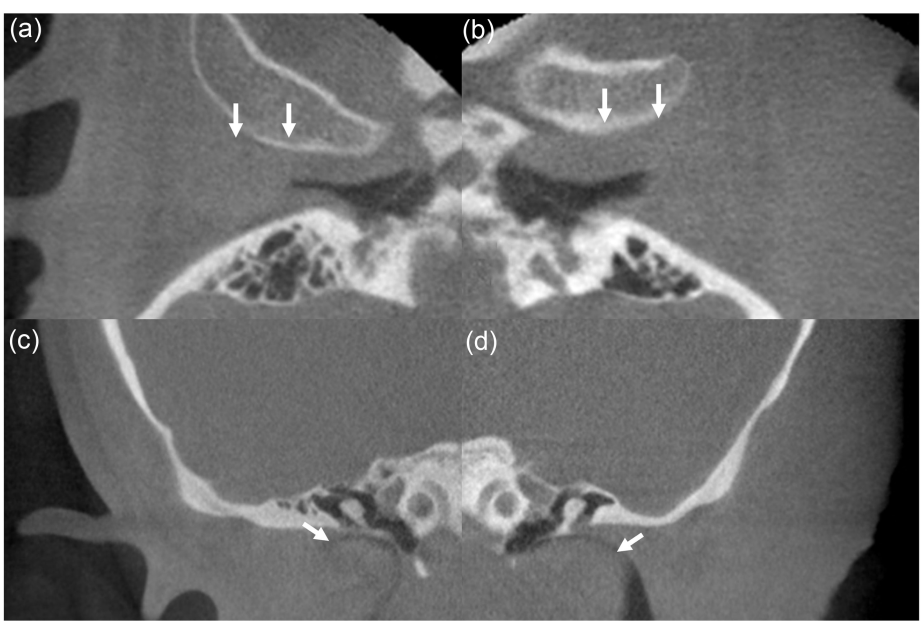

Figure 4. a-d. Computed tomography images of the external auditory canal. (a and b) Axial and (c and d) coronal computed tomography images of the external auditory canal in the (a and c) right and (b and d) left ears. The computed tomography images show the entire bony defects of the anterior external auditory canal wall, leading to external auditory canal obstruction with the soft tissue (white arrow), which is caused by temporomandibular joint herniation in both ears.

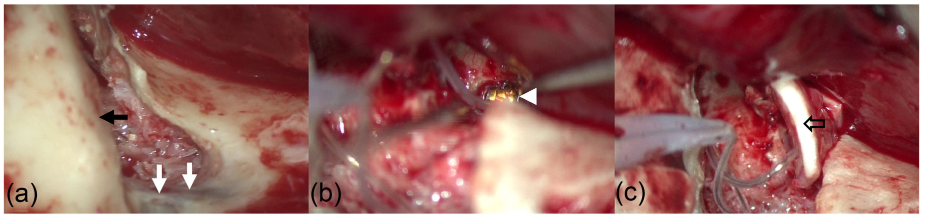

Figure 5. a-c. Intraoperative photographs of the AMEI. (a) The transmastoid procedure is restricted by the low-lying tegmen (black arrow) and the anteriorly situated sigmoid sinus (white arrow) in the right ear. (b) The posterior canal wall of the external auditory canal is partly removed to visualize the round window. The floating mass transducer with a round window coupler (white arrowhead) is placed into the round window niche. (c) The canal wall is reconstructed using a piece of conchal cartilage (blank arrow) to protect the cable from extrusion. AMEI, active middle ear implant.

disappeared, and no supporting structure to insert the material for reconstruction was found (Figure 3).

The conventional hearing aid was not useful in our patient because the fluctuating hearing level led to an unstable effect on the hearing aid. Bone conduction hearing aids were considered an option for treating conductive hearing loss in our patient. However, the devices were not accepted by our patient because of the pressure on the head, sweating, and cosmetic stigma. The bone-anchored hearing aid (BAHA; Cochlear Ltd, Sydney, Australia) is mainly used in cases of conductive and mixed hearing loss when conventional hearing aids do not provide sufficient access to sound or are contraindicated because of anatomical or chronic middle ear conditions. The BAHA was not a feasible option for our patient because CT findings showed an uneven skull surface with irregular thickness and multiple calvarial defects (Figure 3A). A temporal bone CT examination was performed with a radiation impermeable marker at the implant site before surgery. CT revealed that the assumed implant site was near the bone defect of the skull with skull irregularities (Figure 3B). Bony dysplasia is a contraindication for BAHA regarding implant stability with osseointegration. In addition, a BAHA was not accepted by our patient because of cosmetic stigma.

An AMEI is a partially implantable hearing system for patients with conductive and mixed hearing loss. It is implanted in the middle ear and mechanically vibrates the inner or middle ear structures. Active middle ear implants have been used successfully in patients with conductive or mixed hearing loss due to EAC obstruction (i.e., aural atresia). An AMEI was implanted in our patient that avoided the site of the calvarial defect. Because of the poor development of the mastoid cavity and narrow facial recess, the posterior canal wall was dissected to visualize the round window. Cable exposure is the most common cause of revision surgery, occurring in 24%-33% of cases of AMEI implantation after open cavity surgery.[12,13] The cartilage sheet placement over the exposed cable area was effective in preventing cable extrusion.[13] The blind sac closure of the canal was not performed to preserve the future option of using a hearing aid. No cable extrusion or other issues were observed in our patient after canal reconstruction using cartilage during the 30-month follow-up period. The 2 AMEI magnets caused scanning artifacts that consisted of 2 artifact centers. The cerebral hemisphere of the implant side could not be assessed because of the blackening and distortion in MRI caused by the AMEI.[14,15] A cranial CT examination was performed instead of MRI where needed. The AMEI demonstrated a stable effect regardless of the conditions of the patient's mouth, and he was impressed by the improvement in sound quality.

CONCLUSION

Active middle ear implant implantation is a feasible and valuable option for patients with NF1 who have multiple skull defects and TMJ herniation.

Ethics Committee Approval: Investigations were performed according to the Declaration of Helsinki. Our research protocol was approved by the Ethics Review Boards of the University of Miyazaki (registration number: O-0702).

Informed Consent: Informed consent was obtained from the patient involved in the study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – A.G., K.M.; Design – K.K., A.G.; Supervision – A.G., T.T.; Data Collection and/or Processing – K.K., A.G.; Analysis and/or Interpretation - A.G.; Literature Search – K.K., A.G.; Writing Manuscript - K.K.; Critical Review - A.G.

Acknowledgments: The authors would like to thank all the individuals who participated in this study. They are grateful to audiologist S Shimoara from the Miyazaki University and T. Yuji of University of Miyazaki for their assistance in this study.

Declaration of Interests: The authors have no conflict of interest to declare.

Funding: The authors declared that this study has received no financial support.

REFERENCES

1. Boyd KP, Korf BR, Theos A. Neurofibromatosis type 1. *J Am Acad Dermatol*. 2009;61(1):1-14; quiz 15. [CrossRef]
2. Arrington DK, Danehy AR, Peleggi A, Proctor MR, Irons MB, Ullrich NJ. Calvarial defects and skeletal dysplasia in patients with neurofibromatosis type 1. *J Neurosurg Pediatr*. 2013;11(4):410-416. [CrossRef]
3. Kankane VK, Jaiswal G, Gupta TK. Congenital extra calvarial plexiform neurofibroma in occipito-cervical region with occipital bone defect with neurofibromatosis type 1 and segmental neurofibromatosis: case report and review of literature. *J Pediatr Neurosci*. 2016;11(4):295-297. [CrossRef]
4. Mislow JM, Proctor MR, McNeely PD, Greene AK, Rogers GF. Calvarial defects associated with neurofibromatosis type 1: report of two cases. *J Neurosurg*. 2007;106(suppl 6):484-489. [CrossRef]
5. Moriyama M, Kodama S, Suzuki M. Spontaneous temporomandibular joint herniation into the external auditory canal: a case report and review of the literature. *Laryngoscope*. 2005;115(12):2174-2177. [CrossRef]
6. Park YH, Kim HJ, Park MH. Temporomandibular joint herniation into the external auditory canal. *Laryngoscope*. 2010;120(11):2284-2288. [CrossRef]

7. McKennan KX. Neurofibromatosis type I: a rare case resulting in conductive hearing loss. *Otolaryngol Head Neck Surg*. 1991;104(6):868-872. [CrossRef]
8. Abbarah T, Abbarah MA. Neurofibromatosis type 1 causing conductive hearing loss. *Ear Nose Throat J*. 2009;88(5):912. [CrossRef]
9. Ertugrul S, Keskin NK. Relationship of age to foramen of Huschke and investigation of the development of spontaneous temporomandibular joint herniation. *Int J Oral Maxillofac Surg*. 2019;48(4):534-539. [CrossRef]
10. O'Brien DC, Purpura KR, Cassis AM. Bilateral spontaneous temporomandibular joint herniation: a case report and literature review. *Ear Nose Throat J*. 2018;97(9):E23-E27. [CrossRef]
11. Tan NC, Wilson A, Buckland J. Herniation of the temporomandibular joint into the external auditory meatus secondary to benign necrotising otitis externa. *Br J Oral Maxillofac Surg*. 2009;47(2):135-137. [CrossRef]
12. Lassaletta L, Calvino M, Sánchez-Cuadrado I, Pérez-Mora RM, Muñoz E, Gavilán J. Pros and cons of round window vibroplasty in open cavities: audiological, surgical, and quality of life outcomes. *Otol Neurotol*. 2015;36(6):944-952. [CrossRef]
13. Barbara M, Volpini L, Covelli E, Romeo M, Filippi C, Monini S. Complications after round window vibroplasty. *Eur Arch Otorhinolaryngol*. 2019;276(6):1601-1605. [CrossRef]
14. Ezerarslan H, Ataç GK, Keskin S, Beriat GK, Kocatürk S. Temporal bone posterior wall defect, a new clinic entity? *J Int Adv Otol*. 2015;11(1):77-80. [CrossRef]
15. Todt I, Mittmann P, Ernst A, Mutze S, Rademacher G. In vivo experiences with magnetic resonance imaging scans in Vibrant Soundbridge type 503 implantees. *J Laryngol Otol*. 2018;132(5):401-403. [CrossRef]