INTRODUCTION

Gaucher’s Disease (GD) is an autosomal recessive disorder resulting from deficiency of glucocerebrosidase, a lysosomal enzyme, which results in accumulation of glucosylceramide in macrophages, also known as Gaucher cells. Symptoms include hepatosplenomegaly with associated anemia and neurological deficits. GD is classified into three categories, with Type III GD representing the chronic neuropathic form. Common symptoms include: cranial nerve palsies, ataxia, dementia, supranuclear ophthalmoplegia, and irreversible peripheral nerve damage, which may culminate in total sensorineural hearing loss in 10% of all GD cases. Minimal information exists regarding the management of hearing loss in GD patients. We present a rare case of successful cochlear implantation (CI) in a pediatric patient with Type III GD.

CASE PRESENTATION

Cochlear Implant Evaluation

A 16 year-old female with Type III GD presented with severe progressive auditory complaints. She had no history of meningitis, mastoiditis, chronic ear infections, or previous otologic surgeries. Hearing aids were worn for many years with success; however, a significant decline in functional benefit was documented. Audiometric testing showed a bilateral profound sensorineural hearing loss and extremely poor sentence discrimination (0% correct) as indicated by the AZ-BIO Sentence Test (Fig. 1). In addition, pure tone average before implantation was approximately 95 dB HL (Fig. 2). Based on these assessments and reported history, the patient was considered a candidate for cochlear implantation. A preoperative CT scan showed clear epitympanic and hypotympanic spaces bilaterally, well aerated mastoid aid cells on the right and opacified air cells on the left. In addition, the cochlea and semicircular canals were normal bilaterally.

Cochlear Implantation

A cortical mastoidectomy was performed revealing a significant amount of vascular granulation tissue which was carefully mobilized and debrided from the antrum (Fig. 3a). Anterior dissection approaching the middle ear cavity revealed significant edema of the mucosa completely covering the oval window, round window, stapes superstructure, and incudostapedial joint. With difficulty visualizing the normal landmarks, the case was aborted and biopsy was taken of the left mastoid tissue on suspicion that a cholesteatoma may be present. Initially, frozen section results indicated the tissue consisted of histiocytes (CD68+) outnumbering squamous cells, consistent with cholesteatoma. After further review, macrophage accumulation was deemed consistent with GD (Fig. 4a).

11 weeks after the initial operation, the patient was consented for right cochlear implantation. Similar presentation to the left ear was seen with thickened tissue all over the mastoid and antrum as well as thickened mucosa over the round window. Again, a biopsy was performed (Fig. 4b). A cochleostomy was performed and the placement of the implant was confirmed with expected impedances and neural stimulation. Appropriate positioning of the electrode was visualized on high penetrance transorbital x-ray. Nine months after successful right CI, the left CI was inserted (Fig. 3b). Neural Response Telemetry (NRT) on the days of surgical implantation for both right and left CIs showed neural response to stimulation across apical, medial, and basal electrodes.

DISCUSSION

Aside from this case report, there is no documentation of successful cochlear implantation in Type III GD patients. There is controversy whether cochlear implantation is appropriate for patients with peripheral neurological disorders, as the integrity of the auditory nerve may be compromised. Although auditory neuropathy may be present in association with GD, cochlear implantation may improve synchronicity of neural activity and stimulate sound awareness. Other studies reported improved electrically evoked brainstem responses and NRT after implantation in patients with similar neurological diseases. For pediatric patients with Type III GD, cochlear implantation may serve as effective management for improved auditory sensitivity, functionality, as well as quality of life. Further investigations are necessary to evaluate the means of aural rehabilitation for this rare population.

CONCLUSION

This case report serves to raise awareness of this rare condition and propose that patients with hearing loss secondary to GD may be candidates for cochlear implantation which may improve auditory sensitivity, sound awareness, and quality of life.

REFERENCES


TEXAS TECH UNIVERSITY HEALTH SCIENCES CENTER