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Case Report

Clinical Presentation of Congenital Cholesteatoma of the External Auditory Canal in Goldenhar Syndrome: A Case Series

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Abstract: Background: Goldenhar Syndrome, also called Oculoauricular Vertebral Dysplasia, includes abnormalities of the first and second branchial arches. Ear abnormalities are present in approximately 92% of those with Goldenhar Syndrome [1]. There is a wide spectrum for ear anomalies; they can range from preauricular tags and pits, microtia, external auditory canal (EAC) atresia, ossicular malformations, and facial nerve hypoplasia, which can lead to an array of clinical findings [5]. Hearing loss is an unfortunate consequence of these clinical findings, and often results in subsequent speech and language delays [4]. Despite the widely reported auricular and audiologic manifestations of this disease, a review of the literature revealed no detailed description or analysis of congenital canal cholesteatoma specifically in this patient population. Objective: This study aims to characterize congenital external auditory canal cholesteatoma as a feature of Goldenhar Syndrome. Method: A retrospective case series was conducted with patients seen at the UNC Craniofacial Center in Chapel Hill, NC. Inclusion criteria included patients with a diagnosis of Goldenhar Syndrome also found to have congenital cholesteatoma necessitating surgery. A total of three patients were identified who met inclusion criteria. Results: Three patients with Goldenhar Syndrome were identified who also had congenital external auditory canal cholesteatoma. All three patients underwent surgical management of their disease. Conclusion: Congenital external auditory canal cholesteatoma appears to be a feature of Goldenhar Syndrome not widely described in current literature. Though rare, it is a relevant disease process with significant clinical implications for both hearing as well as speech and language development. Awareness of this condition can thus help guide practitioners in the care of these patients.

Keywords: Goldenhar Syndrome, Congenital External Auditory Canal Cholesteatoma, Oculauriculovertbral Spectrum, Hemifacial Microsomia, Craniofacial Microsomia

1. Introduction

Goldenhar Syndrome, a congenital disorder also known as Oculoauricular Vertebral Dysplasia, is one of the most common craniofacial abnormalities with an estimated incidence of 1:3,500 to 1:5,600 [2]. It widely thought to due to abnormalities of the first and second branchial arches [1]. It has been associated with a variety of possible causes including environmental, teratogenic, vascular insufficiencies, and twin pregnancies [3]. Though known to

have an embryologic etiology, there have also been theories surrounding teratogenic exposures including thalidomide, primidone, anticonvulsants, retinoic acid, and smoking [4, 5]. Some studies suggest that placental abnormalities resulting in vascular insufficiencies (2nd trimester bleeding) have been linked to Goldenhar syndrome [6]. There have also been numerous chromosomal abnormalities associated with Goldenhar syndrome and reports of familial cases, suggesting a genetic component [7].

Craniofacial microsomia (CFM) is a complex congenital condition. It encompasses a wide phenotypic spectrum including malformations of the ear, facial nerve, kidneys, heart, mandible, and soft tissue structures of the face [8]. One unique challenge of CFM is that it lacks a single widely accepted definition, creating unintended discrepancies in published literature. It is important to gain understanding of the neurodevelopmental risks that CFM poses so that clinicians are aware of how to provide multidisciplinary care. Published literature illustrates that certain phenotypic patterns correlate with increased risks of neurobehavioral deficits. Patients with combined microtia and mandibular hypoplasia have increased risks of cognitive-academic problems [9]. The longitudinal cohort study Craniofacial microsomia: Longitudinal Outcomes in Children in pre-Kindergarten (CLOCK) aimed to investigate neurobehavioral outcomes and phenotypical patterns in infants and toddlers with CFM. The cohort characteristics in this study included patients with varying grades of ear canal atresia and auricular pits but did not demonstrate any patients with other auricular findings [10]. Given the association between certain phenotypic findings and neurobehavioral outcomes, it is imperative that clinicians have an in-depth understanding of the phenotypical abnormalities present in this patient population.

Auricular abnormalities, specifically, are present in approximately 92% of Goldenhar syndrome patients [1]. These anomalies can range from preauricular tags and pits, microtia, external auditory canal (EAC) atresia, ossicular malformations, and facial nerve hypoplasia, which can lead to an array of clinical findings [5]. Of the utmost clinical importance is the potential for significant hearing loss with subsequent speech and language delays [4]. Hearing loss, of any kind, has been reported to be as high as 85% in Goldenhar syndrome patients [1]. Despite the widely reported auricular and audiologic manifestations of this disease, a review of the literature revealed no detailed description or analysis of congenital canal cholesteatoma specifically in this patient population. Congenital external auditory canal cholesteatoma is a rare occurrence and its pathophysiology is thought to be similar to that of acquired cholesteatomas [11, 12]. It develops from epithelial squamous cells and presents as an occupying mass on the floor of the EAC. Patients typically present at the mean age of four and a half years and symptoms usually include pain and otorrhea [11, 12]. Characteristically, congenital cholesteatomas occur most frequently in the middle ear though when they occur in the EAC, the cholesteatoma causes gradual expansion of the canal secondary to bony erosion by accumulated desquamated debris [12]. Definitive management entails surgical resection [13]. Overall, the incidence of congenital cholesteatomas occurring in the EAC is rare and not well documented in Goldenhar syndrome patients. Thus, the primary objective of this study is to illustrate and describe three separate patients with Goldenhar Syndrome who presented with congenital external auditory canal cholesteatoma.

2. Materials and Methods

This is a case series with retrospective chart review of three separate Goldenhar syndrome patients who presented with congenital canal cholesteatoma necessitating surgery.

3. Results and Discussion

The study includes 3 patients (Female=2, Male=1) who were all diagnosed with Goldenhar syndrome and found to have congenital external auditory canal cholesteatoma. All three patients had evidence of hemifacial microsomia with microtia and atresia on the left in 2 patients and on the right in 1 patient.

The first patient is a 4-year-old female with Goldenhar syndrome with associated Grade 2 microtia and atresia on the right. Her presenting symptom was a chronic history of persistent otorrhea from her atretic ear canal despite medical therapies including antibiotic and steroid ear drops and an oral course of antibiotics. A computerized tomography (CT) scan was performed which demonstrated complete opacification of the right external auditory canal with extension into the middle ear cavity and mastoid with evidence of ossicular erosion (See Figures 1 and 2). She was taken to the operating room for a right atresiaplasty and canal wall up tympanomastoidectomy with removal of the cholesteatoma. She follows up regularly for repeat otologic examinations and is doing well with her bilateral hearing aids. She has not had any signs of disease recurrence.

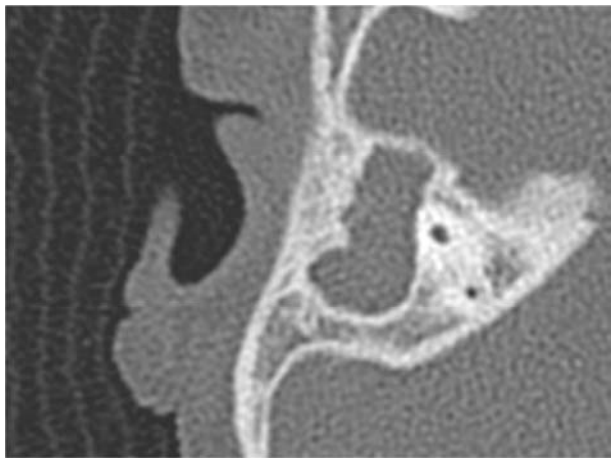

Figure 1. *Axial CT Right Temporal Bone. Left external auditory canal (EAC), middle ear cavity, and mastoid, are completely opacified with soft-tissue. Findings are compatible with a congenital cholesteatoma.*

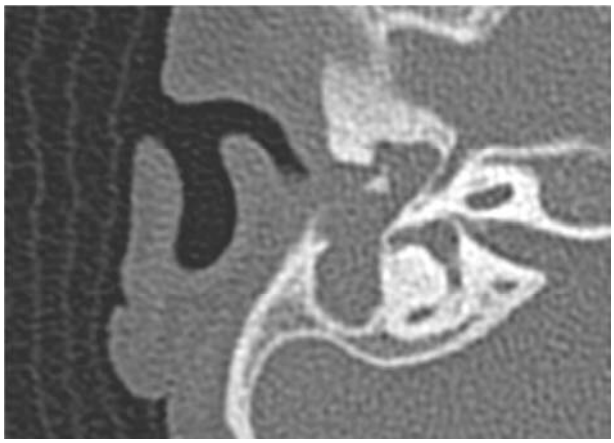

Figure 2. *Axial CT Right Temporal Bone with evidence of ossicular erosion. There is a small portion of the malleus and incus remaining.*

Our next case involves a 7-year-old male with Goldenhar Syndrome and associated Grade 2 microtia and atresia on the left side. He presented with a recurrent left post-auricular abscess that was drained multiple times at an outside facility and required multiple rounds of antibiotics. He ultimately developed a chronic draining post-auricular cutaneous fistula prompting imaging which included a magnetic resonance imaging (MRI) scan. Imaging confirmed a T2 avid expansile mass within the right ear canal (See Figure 3). An audiogram performed showed a 30dB conductive hearing loss across all frequencies. He was taken to the operating room for a canal wall up tympanomastoidectomy and excision of the fistula tract. Operative findings included a congenital canal cholesteatoma with a small amount of disease medial to the atresia plate which was removed. No extension of disease into the mastoid cavity was appreciated. He has been followed for 3 years post-operatively and has had no signs of disease recurrence. He is currently being evaluated for a Bone Anchored Hearing Aid (BAHA).

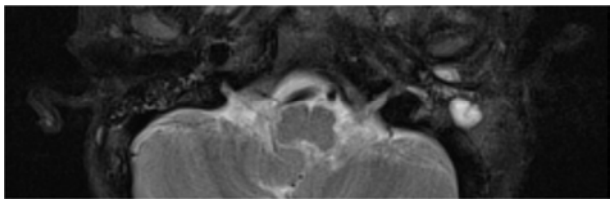

Figure 3. *Axial T2-Weighted MRI with evidence of an expansile mass located in the right external ear canal.*

The last case involves a 7-year-old female with Goldenhar syndrome and left-sided Grade 2 microtia with atresia characterized by a pin point ear canal. She presented with one year of recurrent otorrhea refractory to antibiotic and steroid otic drops. A CT and MRI were performed which demonstrated opacification of the left EAC with a T2 avid mass with restricted diffusion consistent with a cholesteatoma (See Figures 4 and 5). She was taken to the operating room for removal of external ear canal cholesteatoma via atresiaplasty approach and underwent simultaneous placement of a left-sided BAHA. She has been followed for one year postoperatively and has had no disease recurrence.

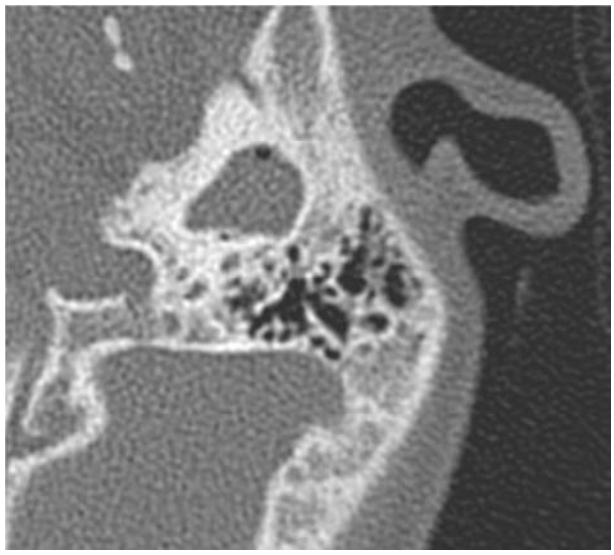

Figure 4. *Axial CT Left Temporal Bone with evidence of a dysmorphic and expanded left external auditory canal (EAC) with near complete opacification. The scan demonstrates that the soft tissue portion of the left EAC is imperforate. The soft tissue invasion into the middle ear is consistent with a congenital cholesteatoma.*

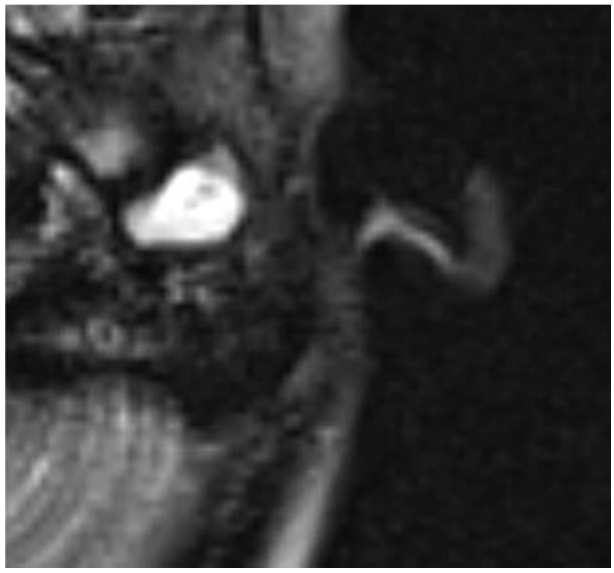

Figure 5. *Axial T2 MR Imaging of the Left temporal bone. Avid mass invading the middle ear space with hyperintensity. The mass is consistent in location and size as the CT left temporal bone in Figure 4. Weighted diffusion image (not shown) demonstrated restricted diffusion, suggestive of a congenital cholesteatoma.*

Thus, presenting symptoms for a congenital cholesteatoma of the external auditory canal included persistent otorrhea refractory to antibiotic and steroid otic drops and, in one case, a persistently draining post-auricular cutaneous fistula. The average age at the time of surgery for this patient population was 6 years-old.

All patients underwent surgery with extensive removal of cholesteatoma centered in the EAC with middle ear and mastoid involvement in only one case. Additional procedures performed at the time of cholesteatoma removal included placement of a bone-anchored hearing aid in one case for

hearing rehabilitation. These three cases stress the importance of understanding the various clinical features of Goldenhar syndrome so that providers maintain the appropriate clinical suspicion should a patient present with persistent otalgia and otorrhea despite multiple medical therapies. Though a rare entity, congenital external auditory canal cholesteatoma has a multitude of clinical manifestations and consequences that greatly impact both the patients and their families.

4. Conclusion

Congenital cholesteatoma of the external auditory canal, though rare, represents a relevant disease process with real clinical implications in patients with Goldenhar Syndrome. The presence of congenital cholesteatoma can have lasting impacts on hearing, developmental milestones, and social maturity. As clinicians, it is important to understand these implications so that appropriate screening and diagnostic workup is pursued. Craniofacial disorders present with a variety of multidisciplinary challenges and require a holistic approach to care. Current literature should reflect the full range and spectrum of clinical phenotypes in patients with craniofacial disorders so that multidisciplinary teams can appropriately care for these patients. The presence of congenital cholesteatoma of the EAC in Goldenhar Syndrome is an example of a clinical implication that is not widely recognized. This paper identifies and describes congenital cholesteatoma of the EAC in patients with Goldenhar Syndrome, requiring surgical management. Should a patient with Goldenhar Syndrome present with persistent otalgia and otorrhea despite multiple medical therapies, congenital cholesteatoma should be considered. Awareness of this condition can thus help guide practitioners in the care of these patients, particularly when deciding whether early radiographic imaging may be helpful should clinical concerns arise.

Author Contributions

S. H. and A. D conceived of the present idea and identified the subjects for the study. S. H. and A. P. wrote the manuscript with support from A. D. and B. O. The surgical relevance and implications were provided by B. O. and A. D. The project was supervised by A. D. and B. O.

Conflict of Interest

There are no conflicts of interest. This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

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