Abstract

INTRODUCTION
CHARGE syndrome is a rare genetic disorder with a spectrum of developmental abnormalities. The diagnostic criteria include various CNS malformations, tracheoesophageal fistula, cleft palate, and characteristic facial dysmorphisms. Temporal bone findings are sensitive in the identification of the CHARGE patient.

OBJECTIVE
The purpose of this study is to review radiological findings of temporal bone anatomy in CHARGE patients diagnosed at our institution, as compared with previous publications.

METHOD
IRB approval was obtained. A retrospective chart review of all patients with an ICD-9 code of 759.89, corresponding to CHARGE syndrome, at a pediatric tertiary referral center from 2000-2010.

RESULTS
A total of 810 patients were identified. A total of 14 patients had a genetic diagnosis of CHARGE. CT temporal bone scans were available for 8 patients, resulting in 16 temporal bones that were analyzed. Ossicular chain abnormalities were noted in 87.5% of temporal bones, aplasia of the oval window in 94%, and some degree of semicircular canal aplasia in 100%. In addition, 81% had an enlarged emissary vein and 87.5% had an aberrant course of the facial nerve.

CONCLUSION
Patients with a diagnosis of CHARGE syndrome often have significant hearing and vestibular deficits. Understanding their temporal bone anatomy will help facilitate auditory and vestibular rehabilitation. Characterizing the anatomic abnormalities expected for these children provides a guide for preoperative surgical planning of any otologic intervention. Additionally, the findings can serve as useful adjuncts for children who are being evaluated for CHARGE syndrome.

Objective
To report the radiological findings of temporal bone anatomy in CHARGE patients diagnosed at our institution.

We will further define the role of computed tomography in the diagnosis, therapeutic evaluation, and preoperative planning in CHARGE patients.

Methods & Materials
IRB approval was obtained for a retrospective chart review. A medical records search was performed for all patients encountered with an ICD-9 code of 759.89, corresponding to CHARGE syndrome, who were evaluated at our institution from 2000-2009.

Inclusion criterion was a diagnosis of CHARGE syndrome by the Department of Medical Genetics.

Fourteen patients were identified who met this criterion. CT temporal bone scans were available on the imaging system for 7 patients, and one hard copy scan was obtained following request of the identified patient.

Each ear was evaluated. Measurements of all structures were obtained using the available calipers of the imaging system.

Structures were deemed aplastic when there was a complete absence of development. Hypoplasia was noted in cases of an incomplete development. Dysplasia denotes an aberration in development. Furthermore, in characterization of cochlear anatomy, we have chosen to use Mondini deformity to denote a hypoplastic cochlea, with an incomplete partition of the apical turn. The course of the facial nerve was scrutinized and compared with expected norms of anatomy.

Discussion
We have provided a complete and thorough description of the anatomic anomalies of the temporal bone in CHARGE syndrome patients. Our results are comparable to other studies (see below) with similar sample sizes.

By characterizing the anatomic abnormalities encountered in these patients, we hope to provide a guide for surgical planning for cochlear implantation or otologic intervention. Abnormal anatomy should not preclude surgical intervention in these patients, but modification in surgical technique may be required.

Finally, these findings can serve as useful adjuncts to the assessment of patients who are being evaluated for CHARGE syndrome.

Conclusions

- Radiographic abnormalities of the temporal bone are sensitive and specific findings in CHARGE syndrome.
- Our findings corroborate and further the body of literature for this syndrome.
- Children with CHARGE syndrome will inevitably be evaluated by otolaryngologists who must be cognizant of their unique anatomy to aid in diagnosis and surgical planning.

References