Venous Malformations of the Temporal Bone:
A common feature in CHARGE

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INTRODUCTION

• CHARGE (Coloboma of the eye, Heart defects, Atresia of the choanae, Retardation of growth and/or development, Genital and/or urinary abnormalities, and Ear abnormalities and deafness) is a phenotypically variable genetic disorder occurring in 1:10,000 births. It includes prominent otolaryngologic features including inner ear malformations.

• Deletions in the gene CHD7 on chromosome 8 is most commonly implicated.1,2 Such mutations are most often sporadic but may be an autosomal dominant condition.

• CHARGE is believed to result from abnormal differentiation of cephalic mesoderm (otic placode and first branchial arch), and neural crest cell migration.

• Classically, 4/7 criteria was needed to diagnose CHARGE, with proposed revisions using the presence of expanded major and minor criteria to help make the diagnosis.

• While inner ear malformation, (specifically semicircular canal aplasia and cochlear anomalies), are among the major criteria,3,5 some have proposed other temporal bone anomalies be considered as part of the diagnostic criteria.4

• As a result of their hearing impairment, many CHARGE patients are candidates for cochlear implantation for which they undergo preoperative CT of the temporal bone.

• While some authors have reviewed the challenges inherent in cochlear implantation in CHARGE related to anomalous cochleovestibular anatomy,3,6 venous malformations of the temporal bone have not previously been recognized as a feature of this syndrome.

• We sought to define the spectrum of venous abnormalities in CHARGE and their surgical implications in otology.

STUDY DESIGN

• Retrospective review of medical and radiology records from databases of patients with CHARGE from three tertiary care academic medical centers.

• Temporal bone CT of 20 patients with CHARGE were reviewed specifically to assess for venous malformations of the temporal bone. MRI was also viewed when available. Associated temporal bone abnormalities and clinical features were also noted.

RESULTS

• Venous malformations of the temporal bone were present in 9 of 20 (45%) patients.

• While some of these abnormalities are of little clinical importance, others may greatly impact surgical approaches to cochlear implantation.

• The most common abnormality was a trans-mastoid emissary vein (n=5) emanating from an occipital vein and draining to the sigmoid sinus which was sometimes bilateral (Fig. 1, 2, and 5).

• Enlarged emissary veins that normally involute during development were associated with a hypoplastic sigmoid venous sinus or jugular foramen (n=3).

• Other abnormalities included an aberrant petrosal sinus, venous lakes in proximity to the sigmoid sinus, posterior condylar canal veins (Fig. 2) and jugular bulb abnormalities including a high riding bulb obscuring the round window niche, jugular bulb diverticulum and a dehiscent jugular bulb.

• These venous anomalies were associated with other inner ear malformations in all cases, including absence of the semicircular canals and a variety of vestibulocochlear dysplasias.

• Venous canals were more easily identified on CT than MRI in cases where both were available for retrospective review.

• In some cases, the course of the aberrant vessel necessitated a change in the surgical approach such as altering the trans-mastoid approach or the placement of the cochleostomy in cochlear implant surgery.

DISCUSSION

• Venous malformations of the temporal bone are a common feature in CHARGE that have not previously been appreciated.

• While several case-series have reported isolated venous anomalies in CHARGE patients, none have previously focused on defining the spectrum of these specific abnormalities and its implications in otologic surgery which these patients undergo.

• In particular, collateral emissary veins of the temporal bone including posterior condylar veins, mastoid emissary veins connecting occipital or postauricular veins with the sigmoid sinus and retrosquamosal emissary veins of the temporal bone have been previously noted.6

• One report of patients with retrosquamosal sinuses included a 6-year old female with CHARGE. CT revealed inner ear anomalies and a hypoplastic jugular foramen noted on ipsilateral side of a transmastoid venous canal which joined the internal jugular vein.7 Others noted abnormalities in sigmoid sinus development and jugular bulb diverticulum.3,5,8

• Reports of skull base anomalies in CHARGE including abnormal basioccipital development and hypoplastic jugular foramen noted are suggestive of preceding developmental bony malformations leading to venous malformation, possibly as a compensatory response for required venous drainage.9,10

• Collateral emissary veins should be evaluated as part of the pre-operative assessment for cochlear implant surgery. CT of the temporal bone may reveal venous malformations that require pre-operative identification and possibly an altered surgical approach to mastoidectomy and/or cochleostomy as encountered in this series.

• Such abnormalities may not be mentioned in the official radiologic report, so otologic surgeons must look specifically for these findings.

CONCLUSIONS

• Venous abnormalities are a common feature in CHARGE patients and may be seen more often in association with other anatomic malformations of the inner ear. As such, they too constitute a spectrum of temporal bone abnormalities that may be included as minor criteria in making the diagnosis.

• The pattern of venous abnormality may suggest a correlation between the failure of skull base development or sigmoid sinus/jugular bulb to fully develop and persistence of emissary veins that normally involute.

• CHARGE patients undergoing cochlear implantation should be evaluated pre-operatively for venous malformations of the temporal bone in addition to anomalous vestibulocochlear anatomy.

• Identification and avoidance of these abnormal venous structures pre-operatively is critical to avoiding potentially catastrophic bleeding during otologic surgery or disrupting dependent drainage pathways leading to ischemia.

REFERENCES


