Cochlear Abnormalities Associated with Dursun Syndrome

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ABSTRACT

Background:
G6PC3 deficiency causes severe congenital neutropenia type 4 and is associated with a wide phenotypic spectrum. Dursun syndrome represents a subset of these patients who have the triad of leukopenia, atrial septal defect, and primary pulmonary hypertension. To date approximately 57 patients with G6PC3 deficiency have been reported and only 3 with Dursun syndrome.

Study Design:
Case report.

Methods:
We describe a single novel case of a 3 year old male with Dursun syndrome who failed newborn hearing screening and was found to have bilateral profound sensorineural hearing loss (SNHL). MRI and CT imaging demonstrated bilateral cochlear malformations consistent with incomplete partition type 1.

Results:
This patient underwent successful placement of simultaneous bilateral cochlear implants and is progressing with auditory development.

Conclusions:
To our knowledge, this is the first report of cochlear abnormalities associated with G6PC3 deficiency and Dursun syndrome. If candidacy is confirmed, cochlear implantation is a viable option for auditory rehabilitation.

CASE REPORT

We report a single novel case of an ex-31 week, prematurely born, now 3 year old male with Dursun syndrome who failed newborn hearing screening. At 6 months of age ABR testing revealed bilateral profound sensorineural hearing loss (SNHL). Subsequent behavioral testing demonstrated similar findings, and he received no benefit from amplification.

MRI and CT temporal bone imaging were performed and demonstrated absence of the basal turn of the cochlea bilaterally, a bulbous middle turn, and absent partitioning of the apical turn. The modiolus was absent bilaterally, however a cochlear nerve was discernible. These findings are most consistent with a cystic cochlear vestibular anomaly (incomplete partition type 1).

The patient underwent successful simultaneous bilateral cochlear implants with the Nucleus 24RE devices and the 422 electrode.

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REFERENCES


