
Characteristic Cochlear Hypoplasia in Patients with Walker-Warburg Syndrome: A Radiologic Study of the Inner Ear in α-Dystroglycan-Related Muscular Disorders

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Abstract

Background and purpose: Walker-Warburg syndrome, muscle-eye-brain disease, and Fukuyama congenital muscular dystrophy are α-dystroglycan-related muscular disorders associated with brain malformations and eye abnormalities in which no structural inner ear abnormality has been described radiologically. We collected patients from 6 tertiary pediatric hospitals and reported the radiologic features and frequency of inner ear dysplasias.

Materials and methods: Patients previously diagnosed clinicoradiologically with Walker-Warburg syndrome, muscle-eye-brain disease, or Fukuyama congenital muscular dystrophy were included. We recorded the pathogenic variant, when available. Brain MR imaging and/or CT findings were reviewed in consensus, and inner ear anomalies were classified according to previous description in the literature. We then correlated the clinicoradiologic phenotype with the inner ear phenotype.

Results: Thirteen patients fulfilled the criteria for the Walker-Warburg syndrome phenotype, 8 for muscle-eye-brain disease, and 3 for Fukuyama congenital muscular dystrophy. A dysplastic cochlea was demonstrated in 17/24. The most frequent finding was a pronounced cochlear hypoplasia type 4 with a very small anteriorly offset turn beyond the normal-appearing basal turn (12/13 patients with Walker-Warburg syndrome and 1/11 with muscle-eye-brain disease or Fukuyama congenital muscular dystrophy). Two of 8 patients with muscle-eye-brain disease, 1/3 with Fukuyama congenital muscular dystrophy, and 1/13 with Walker-Warburg syndrome showed a less severe cochlear hypoplasia type 4. The remaining patients without Walker-Warburg syndrome were healthy. The vestibule and lateral semicircular canals of all patients were normal. Cranial nerve VIII was present in all patients with diagnostic MR imaging.

Conclusions: Most patients with the severe α-dystroglycanopathy Walker-Warburg syndrome
phenotype have a highly characteristic cochlear hypoplasia type 4. Patients with the milder variants, muscle-eye-brain disease and Fukuyama congenital muscular dystrophy, more frequently have a normal cochlea or milder forms of hypoplasia.

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