A Successful Treatment of Endoscopic Third Ventriculostomy with Choroid Plexus Cauterization for Hydrocephalus in Walker-Warburg Syndrome.

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Abstract

Walker-Warburg syndrome (WWS) is a rare autosomal recessive congenital muscular dystrophy with brain malformations and ocular abnormalities that falls under the wider phenotypic spectrum of the dystroglycanopathies. Mutations in a number of genes including POMT1, POMT2, POMGNT1, POMGNT2, FKTN, FKRP, LARGE, and ISPD are known to cause alpha dystroglycan-related muscular dystrophy. Mutations in these genes result in a broad phenotypic spectrum ranging from the severe WWS to a mild congenital muscular dystrophy with no brain involvement. WWS is fatal to most patients early in life with mean survival of 9 months. The most common brain finding is cobblestone lissencephaly with the vast majority of patients (97%) also having ventricular dilation with or without hydrocephalus. Surgical treatment has not been frequently detailed. This report describes our successful treatment of a patient with WWS and hydrocephalus with Endoscopic Third Ventriculostomy (ETV) with choroid plexus cauterization (CPC). Fourteen months following treatment, a follow-up MRI CSF flow study demonstrated robust CSF flow through floor of third ventricle from interpeduncular cistern to lateral ventricle.

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