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Case Report

Axonal Transport in Coats' Disease and Congenital Oculodentodigital Syndrome

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ABSTRACT

Introduction: Axoplasmic transport blockade has been demonstrated in acute experimental glaucoma in animals but has not been documented in childhood glaucoma. The purpose of this study was to detail findings and axoplasmic transport status in one eye of a two-year old male with Coat's disease using the two eyes from a 10-month-old female with Oculodentodigital Syndrome (ODDS) as controls for background axoplasmic transport. We document for the first-time axonal transport block in the lamina cribrosa (LC) in a blind and painful right eye associated with Coats' disease, (Case 1). Enucleation was done for suspected retinoblastoma without imaging. Histopathology demonstrated myriad macrophages migrated from the vitreous infiltrate into the anterior chamber and meshwork explaining the glaucoma. Two eyes from Case 2, diagnosed at autopsy as having congenital Oculodentodigital Syndrome (ODDS), with a history of surgically normalized intraocular pressures were included as controls for the Coats' case immunohistochemistry (IHC) and to report additional findings in this rare disorder.

Methods: We reviewed available history and performed histopathology on one enucleated Coats' globe and two additional autopsy eyes from an ODDS case with routine staining of paraffin-processed tissues and amyloid precursor protein (APP-A4) IHC, the only available IHC marker for orthograde axonal transport in human surgical or autopsy tissues. The two eyes from the 10-month-old infant with ODDS had normal IOPs following trabeculotomies at five months of age.

Results: In addition to typical pathology for Coats' the lamina cribrosa portion of the optic nerve from Case 1 demonstrated marked orthograde axonal transport block. Both eyes from Case 2 with ODDS had a history of congenital glaucoma treated medically before bilateral trabeculotomies at five months-age with normalization of IOPs. Descemet scrolls found OD were judged residual to goniotomy. Both globes from Case 2 demonstrated multiple anomalies previously reported in ODDS including microphthalmos, optic disc dysplasia OD, and features of persistent hyperplastic primary vitreous (PHPV) OU. The right eye also demonstrated spontaneous retinal detachment prompting an exam under general anaesthesia at 10-months age when, during induction, she expired with malignant tachycardia. Retinal ganglion cells (RCGs) labeled with APP-A4 OU, but optic nerve LC axons demonstrated only background accumulations of the IHC marker.

Conclusions: Orthograde axonal transport was interrupted in the LC of the Coats' nerve due to secondary inflammatory glaucoma. The angle remained open histologically in-spite-of-early neovascularization and pupillary margin ectropion. Block of orthograde axonal transport in the LC has been well established as the site of initial optic nerve damage in glaucoma in experimental studies and this report reinforces its importance to the pathophysiology of glaucoma optic nerve damage. The primary importance of the two ODDS globes for our report was as comparison controls for background APP-A4 staining in the Coats' case. Our additional findings in the ODDS eyes suggest delayed spontaneous retinal detachment, and lens epithelial abnormalities are additional anomalies to add to the list of known malformations in this rare disorder. Case 2 also serves as a caution in ODDS regarding vulnerability to combinations of medications when general anaesthesia is contemplated.

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