Congenital intraorbital optic nerve cyst

Case report

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Congenital cystic lesions of the optic nerve are exceedingly rare; only one case is reported in the world literature. The authors describe a case of congenital simple glial cyst in the intraorbital portion of the optic nerve with a brief review of the literature and comment on its histogenesis. A 45-day-old male infant was admitted to the hospital because of progressive proptosis and hypotropia in the left eye, which had been present since birth. Magnetic resonance imaging of the left orbit revealed an ovoid, well-demarcated, homogeneous cystic mass in the intracranial retrobulbar area. The mass compressed the left eyeball with downward and lateral displacement. The wall of the cystic mass was very thin, and a needle puncture of the cyst released clear, colorless, watery fluid. The cystic wall was lined by loose astroglial nerve fibers with some scattered glial cells.

Case Report

History. This 45-day-old male infant was first examined in the Department of Neurosurgery because of progressive proptosis and hypotropia in his left eye, which had been present since birth. There was no history of perinatal infection or birth trauma. At birth, the infant’s body weight was 4.2 kg, his height was 47 cm, and his head circumference was 37 cm (within the 50th percentile).

Examination. The patient could fixate with either eye and both pupils were normal. He showed normal developmental milestones without any gross anomaly except for downward and lateral deviation of the left eyeball. The pupils were equal and round, with the left pupil reacting sluggishly to light and the right pupil reacting normally. Visual evoked potentials showed normal responses in the right eye (latency 80 msec and amplitude 3.43 μV when lights were presented using goggles; latency 169 msec and amplitude 2.81 μV when flashing lights were used) and no response in the left eye (all negative). There was increased resistance to retropulsion of the left eye. Bruit or pulsation was not noted. Eyeball motility was markedly compromised during upward gaze, and there was an afferent pupillary defect in the left eye. Funduscopic examination showed pronounced edema of the left optic disc. Neurological examination showing normal ranges, and pertinent laboratory findings excluded the possibility of infection, infestation, and any embryological maldevelopment.

Operation. The patient underwent left frontal craniotomy.
my with orbit unroofing. Careful dissection of the intraorbital structure exposed a light-tan, cystic optic nerve mass. The cystic mass originated from the retroglobal, distal end of the optic nerve and was confined to the intraorbital portion. The optic canal area was intact. The wall of the cystic mass was thin and continuous with the optic nerve itself. A needle puncture of the cyst released 3 ml of a clear, colorless, watery fluid having the macroscopic appearance of cerebrospinal fluid. The cystic wall was smooth and showed no evidence of vascular anomaly, solid tumor, or previous hemorrhage. Because there had been preoperative total blindness and there was some fear of an associated tumor process, the collapsed cystic mass was sectioned across the distal end of the optic nerve and proximally at its entry into the optic canal.

**Histological Examination.** Cytological examination of the aspirated fluid revealed no inflammatory or tumor cells. Microscopic examination of the operative specimen revealed a cystic lesion that was positioned off center in the optic nerve (Fig. 2). The cystic wall was lined by loose astroglial nerve fibers admixed with some scattered bland glial cells (Fig. 3). In these glial layers, we could not find any fibrovascular pial septa, which usually are found throughout the optic nerve. Prussian blue staining revealed no hemosiderin pigment deposition. Immunohistochemical staining for glial fibrillary acidic protein revealed strong positivity in the optic nerve itself and in the glial fibers surrounding the cyst. There was no arachnoidal layer or epithelial lining along the inner cystic wall, although the cyst was focally connected to the subarachnoid space. The remaining optic nerve showed slight atrophy of nerve-fiber bundles with focal minimal reactive gliosis and diffuse thickening of the fibrovascular pial septa. There was no evidence of neoplastic invasion or inflammatory cell infiltration.

**Postoperative Course.** The postoperative course was uneventful, and the patient has been well for 8 years following the operation.

**Discussion**

Cystic lesions of the optic nerve are extremely rare. To the authors’ knowledge, this is the second case report in which a congenital optic nerve cyst is described and the first account in which pathological findings and MR images are provided.

The diseases that can produce secondary optic nerve cysts include optic nerve glioma, neurofibromatosis, empty sella syndrome, hemangioma of leptomeningeal origin, and meningioma. Although there was no evidence of neoplastic infiltration by nerves or meninges in this case, it was very difficult to exclude the possibility of an arachnoid cyst and/or cystic glial tumors. Arachnoid cysts are leptomeningeal in origin and do not involve the nerve fibers of the optic nerve; however, they can compress the nerve fibers and cause them to atrophy. This case was different from previously reported cases of arachnoid cysts.
because the cystic component was mainly located in the optic nerve and the lining wall was gliomatous in nature. The absence of Rosenthal fibers or hypercellularity in the cystic wall differentiated our case from cystic tumors such as optic glioma and pilocytic astrocytoma. Finally, the findings in our case needed to be differentiated from heterotopic brain tissue in the orbit. Heterotopia of brain tissue is defined as the occurrence of normal neural tissue outside the cranial cavity or spinal canal without direct communication with the central nervous system. However, our case was different from such cases of heterotopic brain tissue because of the absence of neuronal cells, striated muscle fibers, and laminated calcific bodies. Thus this case was diagnosed as a simple glial cyst lined by astroglial fibers containing scattered hypocellular glial cells. We know this cyst was congenital in origin because proptosis and hypopitria in the patient’s left eye had been present since birth. We can therefore conclude that the lesion was a congenital simple optic nerve cyst. Similar cases have been reported in other sites, including brain parenchyma and cerebellum, providing limited pathological material. Previously described pathological findings and cystic contents have been very similar to those of the present case, with the only exception being location. Careful examination of serial sections through the optic nerve and cyst failed to disclose the cause of this cyst. Based on a review of previously reported brain cysts, we propose two hypotheses on the histogenesis of this cyst. The first is similar to that of simple cysts in brain parenchyma. The present cyst might have been formed originally by budding from the ventricular system (subarachnoid system in the optic nerve), with subsequent loss of ependymal cells due to stretching pressure effects as the cyst became distended with fluid. The second possibility is a congenital developmental anomaly of the optic nerve occurring during the embryonic stage. Invagination of the optic vesicle and stalk occurs at the 4.5-mm-sized embryonic stage with the formation of a fissure along their ventral aspects. It is presumed that this cyst developed during the invagination stage of the optic stalk.

Although the precise histogenesis of this case is highly speculative at the present time, it is noteworthy that such a benign cystic lesion of the optic nerve can be mistaken for a cystically changed optic nerve tumor. Clinicians should be aware of the possibility of a benign optic nerve cyst when they have eliminated the possibility of commonly encountered cystic lesions secondary to other diseases. Thus, when a neurosurgeon finds cystic lesions in the optic nerve, we recommend following simple aspiration of the cystic fluid with cytochemical examination of the evacuated fluid when there is no definite evidence of a tumor. These efforts may not only save the patient from a needless enucleation but more important, may dramatically improve the patient’s visual prognosis.

References


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