Variables to consider when confronting a microphthalmos with a cyst: A case report

ABSTRACT

Microphthalmos with orbital cyst is a rare and severe non-hereditary ocular developmental anomaly. Pathologically it represents a failure in the closure of the embryonic fissure at the 7-14 mm stage of gestation, resulting in a congenital microphthalmia and the formation of a colobomatous cyst in different degrees. We present a patient with this condition to emphasize the complexity in the decision making process and review the various management strategies to obtain the best long term results.

KEY WORDS: microphthalmos, orbital cyst, aspiration, orbitotomy, Delleman syndrome

INTRODUCTION

The estimated prevalence rate of congenital microphthalmia is 1.8 per 10,000 births. When associated with colobomatous cyst, this condition is very rare. Currently, there is no agreement to the best approach for its management as this disorder can present in a variable way. We herein report a case of a patient with microphthalmos with orbital cyst and severe non-hereditary ocular developmental anomaly, discussing diagnosis and management.

CASE REPORT

A two-year old patient with Delleman Syndrome was referred to our service, presenting with progressive right proptosis. She had choanal atresia and a history of facial skin tags removed, but was confirmed not to have any brain malformations. After an extensive workup with her pediatrician, CHARGE syndrome (ocular coloboma, heart defects, choanal atresia, mental retardation, and genitourinary and ear anomalies) had been ruled out, and she was found to have a normal karyotype. Clinical examination showed severe right amblyopia with an eso-hyoptropia and marked proptosis. There was an evident infero-temporal iris-choroid-optic nerve coloboma affecting this eye (Figure 1a). MRI confirmed a right enlarged orbit with a microphthalmic globe and 3 cm multilocular intracranial cystic mass, displacing the orbital roof (Figure 1b).

Given the severity of the microphthalmos and the extension of the orbital cyst, a surgical approach was undertaken with cyst aspiration and excision, including the removal of the abnormal globe. A retrocaruncular medial orbitotomy, in conjunction with a Kronlein lateral orbitotomy, was chosen to allow for appropriate exposure of the large cystic lesion (Figure 2a). Following the removal of the cystic content and enucleation of the globe, the significantly enlarged orbital cavity was reconstructed with a 22ml silicone ocular implant wrapped in human donor...
sclera. Macroscopic evaluation of the removed specimen showed a microphthalmic globe with an iris coloboma at 7 o’clock and a cyst of 29x19x14.5mm inferiorly located, adjacent to the optic nerve (Figure 2b). Histopathological analysis revealed hypoplastic iris, dysplastic retina with lack of RPE, optic nerve hypoplasia and retrochoroidal coloboma.

**DISCUSSION**

Microphthalmos with orbital cyst is a relatively rare condition. It is typically unilateral, with no gender predominance. It develops due to failed closure of the ocular embryonic fissure during the 7mm to 20mm stage of development. Histopathological examination of the excised cyst shows an external layer of vascularized connective tissue with an inner lining of neuroglial cells. Progressive enlargement of the cyst is likely due to fluid production by the glial cells, though some hypothesize that glial cell proliferation or a connection with the subarachnoid space may contribute. Diagnosis is usually made in infancy. Patients may present with proptosis or lower lid swelling associated with a blue-tinged mass corresponding to the cyst. Though it is often diagnosed clinically, additional studies such as an orbital ultrasound, computed tomography (CT) and magnetic resonance imaging (MRI) are highly valuable. In addition, it is crucial to determine the visual potential of the microphthalmic eye. Electrophysiologic studies may be necessary. After diagnosing microphthalmos with orbital cyst, a team effort with a pediatrician should be coordinated to rule out any associated systemic abnormalities.

There is no consensus in the best management of this condition, as it varies in presentation. Observation is often recommended for mild microphthalmos and small orbital cyst. In cases of severe microphthalmos with small cyst, conservative treatment with a conformer can be used to allow for orbit development. Advanced cases with significant cystic volume and strong cosmetic concern may require surgical management.

The appropriate timing of surgery remains controversial, as the colobomatous cyst can induce orbital expansion. Some surgeons advocate conservative therapy with delay of surgical intervention until the orbit is of adequate size. For microphthalmos with an enlarging cyst, aspiration of the cystic fluid can be considered as a less invasive approach. Often, however, repeat aspirations are necessary due to fluid re-accumulation.

Surgical excision of the cystic component with preservation of the globe, should be reserved for those cases where the degree of microphthalmos is minimal, and the cyst is large enough to jeopardize patient’s outward appearance. Severe cases may require a more invasive surgical approach, with removal of the abnormal globe and multilocular cyst. It is important to emphasize that the potentially enlarged orbital cavity will necessitate an appropriate volume deficit reconstruction for the best long term results. Frequently, volume replacement with orbital implants or dermis fat grafts can achieve cosmetic and clinical success.

**REFERENCES**