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An incidental finding of intraocular choristoma in an enucleated microphthalmic globe: A histopathologic case report

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ABSTRACT

INTRODUCTION AND IMPORTANCE: Choristomas are benign growth of normal tissue in abnormal location and in the ophthalmic practice, they are more commonly found in the epibulbar region. Intraocular choristoma has been reported in different ocular structures but it is very rare especially in association with microphthalmos.

CASE PRESENTATION: We present a 13-month-old child with bilateral microphthalmia with the left side being more significantly smaller than the right that required enucleation for introducing a larger silicone implant. The histopathological examination revealed an intraocular choristoma consisting of chondroid and adipose tissue with surrounding fibrosis. Other areas in the globe were also underdeveloped and dysplastic including the optic nerve, which was replaced by dense wavy collagen fibers and fibrovascular tissue.

DISCUSSION: Even though choristomas are benign, they may be extensive interfering with visual development especially the ones involving the epibulbar area.

Systemic disease can have choristomas as an ocular feature such as in Goldenhar-Gorlin syndrome. Choristomas inside the eye are rare and they commonly involve the uveal tissue and the optic nerve head mostly in the form of ectopic glandular tissue and choroidal osseous choristoma. Our case is unique in its intraocular retrolental location, composition of chondroid tissue and fat, in addition to the fact that it was found within a microphthalmic globe with other interesting histopathological findings.

CONCLUSION: We report a case of an incidental finding of intraocular choristoma with associated microphthalmia, genetic testing may be useful for establishing a genetic etiology in such cases even in the absence of dysmorphic features.

1. Introduction

Choristomas are benign growth of normal tissue in an abnormal location [1,2]. There have been reports of epibulbar choristomas occurring as extensive lesions interfering with normal ocular development [2]. Some epibulbar dermoids may cover the visual axis and can severely affect vision if left untreated especially in pediatrics [3]. Choristomas can also be part of a systemic disease such as Goldenhar-Gorlin syndrome [4]. They may occur in non epibulbar locations such as the sclera, the eyelids, and extraocular muscles [5]. Furthermore, intraocular choristomas can rarely occur and have been reported before in the iris, ciliary body, choroid, retina and the optic nerve [6]. We report the rare occurrence of intraocular choristoma associated with a microphthalmic eye. This case report was prepared in accordance with the ethical standards and the Helsinki Declaration. No trial of new drugs or therapy is applicable in this case. Case reports do not require Ethical approval in our institution. However, a general written informed consent was taken from the guardian of the patient, which includes permission for anonymous use of information and photos for reporting. This case report has been prepared in line with the updated SCARE 2020 criteria [7].

2. Case presentation

A 13-months old full-term baby who was a product of uneventful pregnancy and was born by normal spontaneous vaginal delivery without perinatal complications, was referred to us by a neonatologist on the first day of birth because of an abnormal sunken left eye and absent red reflex in the right eye. There was no history of consanguinity and the parents had an older sibling who was completely healthy. External examination on the left side showed a small palpebral fissure with unidentifiable globe. The

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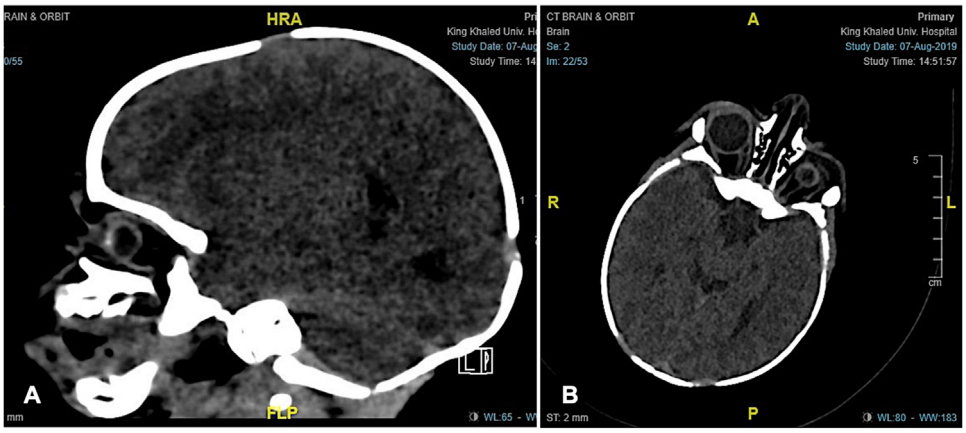

Fig. 1. **A**: A sagittal computerized tomography (CT) scan of the brain and orbits showing a very small left globe with anteriorly located opacification of the retrolental cartilaginous choristoma. **B**: An axial CT scan of the brain and orbits showing normal size right globe and microphthalmic left globe with thin connective tissue stalk where the optic nerve is expected to be present.

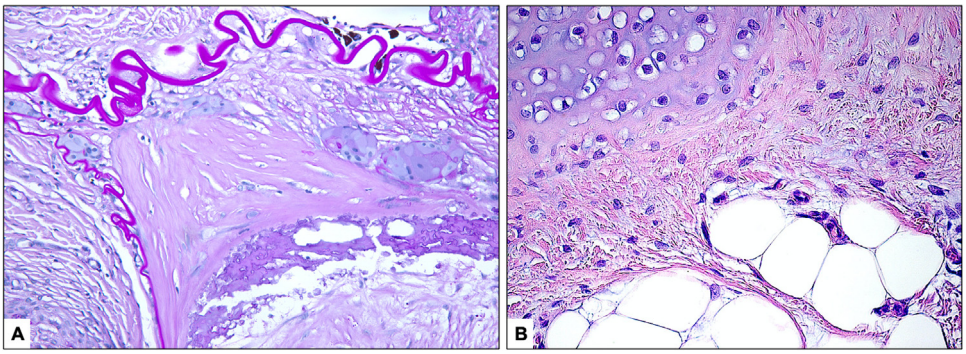

Fig. 2. **A**: Histopathological photo of a calcific lens within the microphthalmic globe showing wrinkled lens capsule and bladder cells (Original magnification ×200 Periodic acid Schiff). **B**: Histopathological photo of the area of retrolental intraocular choristoma consisting of cartilage and fat (Original magnification ×400 Hematoxylin and eosin).

right eye showed a hazy small cornea measuring 6 mm diameter, and evidence of sclero-cornea with narrow palpebral fissure. The baby did not have any dysmorphic features. Computed tomography (CT) revealed a microphthalmic left eye with mild peri-septal thickening and opacification. A thin connective tissue stalk was seen behind the very small globe with no identifiable definite optic nerve tissue (Fig. 1a & b). The right eye was of an average size with an optic disc coloboma. At 12 months of age, he underwent a left silicone orbital implant [14 mm] to enhance the orbital development and expansion by an experienced oculoplastic surgeon. 1 month later he presented to the emergency room with an exposed implant, for which it was decided to go ahead with enucleation of the left globe with a size 16 mm silicone orbital implant. This decision was discussed with the parents, who agreed on the procedure to improve the cosmetic appearance. The globe grossly measured 12 × 10 × 7 mm with an opaque mass anteriorly obscuring the cornea and a 3 mm connective tissue stalk posteriorly where the optic nerve was expected to be located. The histological sections showed a replacement of the corneal epithelium by a fibrovascular mass, which was partially lined by non-keratinizing stratified squamous epithelium with no skin appendages. Bowman's layer was totally absent. The corneal stroma consisted of connective tissue lamellae with loss of the normal stromal lamellar architecture representing a "sclero-cornea" associated with stromal neovascularization especially posteriorly. A peripheral portion of intact Descemet's membrane with few endothelial cells was observed. The anterior chamber angle was completely obliterated on 180 degrees of the globe, while shallow and underdeveloped on the other side. The adjacent iris and ciliary body tissue were disorganized. A retro-corneal folded wrinkled Periodic acid Schiff (PAS)-positive membrane representing a lens capsule was present with anterior subcapsular fibrosis and calcification (Fig. 2a). Furthermore, a retrolental immature chondroid tissue with areas of mature cartilage and a focus of adipose tissue were present with surrounding fibrosis within the posterior cavity of the globe (Fig. 2b). Dysplastic retinal tissue was identified posteriorly with total retinal detachment, subretinal fluid and evidence of retinal gliosis. The choroid showed dilated vascular channels and overlying intact retinal pigmented epithelium with small focal areas of hypertrophy. The connective tissue stalk observed posteriorly consisted of dense wavy collagen fibers and fibrovascular tissue. No definite optic nerve tissue was seen even with deeper sections at multiple levels. The patient did well after the surgery. The last follow up 6 months later showed clean well-healed socket with the cosmetically acceptable prosthesis in place. The parents were satisfied with the surgical outcome.

3. Discussion

Microphthalmia is defined as a small disorganized eye. Kim et al. in 2005 have nicely summarized reported cases of choristomas inside the eye. Most of these cases involved the uveal tissue (iris, ciliary body, and choroid). Most of these consisted of ectopic lacrimal gland followed by osseous choristoma in the choroid [6]. It is rare for microphthalmos and choristoma to occur concurrently [8]. There were two cases of corneal choristomas associated with microphthalmos in which they were also associated with retinal choristomas [9]. Our case is unique because of the location of the choristoma behind the lens, its composition of cartilage and fat, and

the associated severe microphthalmos with probable optic nerve aplasia.

Imaging is helpful in evaluating ocular malformation in the presence of a choristomas. Ultrasonography, computerized tomography (CT) imaging, and magnetic resonance imaging can be used to determine the extent of the choristoma and help in the guidance towards the most appropriate surgical approach [9]. The result of CT in our case has corelated well to our clinical and histopathological findings.

Different approaches have been recommended regarding the management of epibulbar intraocular choristomas. Age is an important factor to consider as the bony orbit reaches 90% of its adult size by the age of 5 years [1]. The main aim is to achieve a good cosmetic outcome as vision is almost always poor when they occur extensively [2]. Whether it is evisceration, enucleation, or subtotal exenteration depending on the nature of its growth [1,7,8]. Although, simple surgical excision of the tumor is an acceptable approach as the residual eyeball may help in stimulating orbit expansion in congenital cases [1]. The main goal in our case was to stimulate orbital bony growth, therefore, a larger implant following enucleation was attempted and has resulted in a satisfactory outcome. Microphthalmos is mostly sporadic, but Mandilion inheritance in the form of autosomal dominant, autosomal recessive and x linked have been described. Mutations in genes SOX2, OTX2and BMP4 represent an autosomal dominant inheritance while PAX6, STRA6 and FOXE3 represent a recessive form of inheritance [10]. As genetic study was not performed in our case, it is unclear whether there is a genetic cause and further studies might establish an association.

In conclusion, we report a case of intraocular choristoma consisting of chondroid and adipose tissue in association with a microphthalmic globe. Most of the previously reported cases of choristoma in the eye are uveal ectopic lacrimal gland tissue or choroidal osseous choristomas. To the best of our knowledge, it is exceedingly rare to have the simultaneous occurrence of microphthalmia and an intraocular choristoma since this is the 3rd reported case in the English-written literature. Genetic testing in such cases might be useful to establish any common genetic etiology.

Declaration of Competing Interest

The authors have no conflict of interest related to this case report.

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Ethical approval

This case report was prepared in accordance with the ethical standards and the Helsinki Declaration. No trial of new drugs or therapy is applicable in this case. Case reports do not require HEC approval in our institution, however the General informed consent includes patient's guardian(s) approval for use of relevant clinical and surgical information in an anonymous way for the purpose of publication.

Consent

A General informed consent from the patient's guardian is taken and includes approval for use of relevant clinical and surgical information in an anonymous way for the purpose of publication.

Author's contribution

First and second authors: Review of chart, literature review and drafting of the case report

Third author: Histopathology of the case along with images, manuscript critical review and the corresponding author

Senior author: Surgical care of the patient and manuscript review before submission.

Registration of research studies

Not applicable.

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References

[1] T.-Y. Huang, Y.-J. Tsai, H.-Y. Tan, L. Ma, Managing epibulbar choristoma with microphthalmos, J. Pediatr. Ophthalmol. Strabismus 45 (3) (2008) 172–173.

[2] Z. Hou, B.S. Korn, J. Ding, D. Li, Management of extensive epibulbar choristoma associated with microphthalmos: a rare clinical entity, JAMA Ophthalmol. [Internet] 132 (6) (2014) 753–755, http://dx.doi.org/10.1001/jamaophthalmol.2014.148, Jun 1.

[3] G. García de Oteyza, A.M. García-Albisua, K.A. Vázquez-Romo, J. Betancourt, M.B. Sandner, V. Sánchez-Huerta, Anterior lamellar keratoplasty for the management of a central dermoid tumour in a baby, Arch. la Soc. Española Oftalmol. (English Ed) [Internet] 95 (1) (2020) 48–51, Available from: http://www.sciencedirect.com/science/article/pii/S2173579419301951.

[4] P. Stergiopoulos, B. Link, G.O.H. Naumann, B. Seitz, Solid corneal dermoids and subconjunctival lipodermoids: impact of differentiated surgical therapy on the functional long-term outcome, Cornea 28 (July (6)) (2009) 644–651.

[5] A.M. Mansour, J.C. Barber, R.D. Reinecke, F.M. Wang, Ocular choristomas, Surv. Ophthalmol. [Internet] 33 (5) (1989) 339–358, Available from: http://www.sciencedirect.com/science/article/pii/0039625789900118.

[6] B.H. Kim, B.A. Henderson, Intraocular choristoma, Semin. Ophthalmol. [Internet] 20 (4) (2005) 223–229, http://dx.doi.org/10.1080/08820530500354052, Jan 1.

[7] R.A. Agha, T. Franchi, C. Sohrabi, G. Mathew, A. Kerwan, for the SCARE Group, The SCARE 2020 guideline: updating consensus Surgical CAse REport (SCARE) guidelines, Int. J. Surg. 84 (2020) 226–230.

[8] R.J. Casey, A. Garner, Epibulbar choristoma and microphthalmia: a report of two cases, Br. J. Ophthalmol. 75 (April (4)) (1991) 247–250.

[9] T. Murata, T. Ishibashi, Y. Ohnishi, H. Inomata, Corneal choristoma with microphthalmos, Arch Ophthalmol. [Internet] 109 (8) (1991) 1130–1133, http://dx.doi.org/10.1001/archopht.1991.01080080090037, Aug 1.

[10] T. Bardakjian, A. Weiss, A. Schneider, Microphthalmia/Anophthalmia/Coloboma Spectrum – Retired Chapter, for Historical Reference Only Genetic Counseling, 2020, pp. 1–12.