Saudi Journal of Ophthalmology (2018) 32, 79–82

Case Report

Orbital tumor associated with a microphthalmic eye and colobomatous cleft: Pilocytic astrocytoma (glioma) or massive retinal gliosis

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Abstract

A 11-year-old boy with congenital microphthalmos of the right eye presented with gradual protrusion of his ocular prosthesis. MRI showed an orbital mass adjacent to the microphthalmic eye. After removal of the eye and the orbital soft tissue mass a gliotic mass, resembling a pilocytic astrocytoma WHO grade 1 (glioma) was diagnosed. Through a colobomatous cleft in the eye the tumour spread in the orbit. There were no clinical signs of neurofibromatosis 1. This case showed a very rare association between a microphthalmic and colobomatous eye and pilocytic astrocytoma, grade 1. However a far advanced and infiltrative massive retinal gliosis cannot be definitively excluded as differential diagnosis.

Keywords: Orbit, Pilocytic astrocytoma, Glioma, Massive retinal gliosis, Congenital microphthalmos, Colobomatous cleft

https://doi.org/10.1016/j.sjopt.2018.02.011

Introduction

This case report highlights the unique presentation of protusion of an ocular prosthesis as the sign of an infiltrating unknown gliotic tumour. This underscores the need of orbital imaging with new ocular prosthesis problems and the importance of a specialised histopathologic examination of all resected tissue.

Case report

An 11-year-old boy was referred with a 2-year history of progressive painless protrusion of his right ocular prosthesis (Fig. 1). At birth, he was diagnosed with clinical anophthalmos of the right eye and a small cleft of the soft palate. Specific questioning did not reveal a family history of eye problems and other congenital disorders. On clinical examination the anophthalmic socket displayed a soft, pink subconjunctival mass and a shallow inferior fornix. The midface had a normal symmetrical appearance. There were no ocular or systemic clinical signs of neurofibromatosis type I (NF1). The examination of the left eye gave normal results.

Magnetic resonance imaging (MRI) of the orbit showed a microphthalmic eye with identifiable extraocular muscles and optic nerve. (Fig. 2) There was a well-defined, multilobulated orbital mass located anteroinferior to the microphthalmic eye. Adjacent to the orbital mass, there was bowing

Received 28 November 2017; received in revised form 11 February 2018; accepted 11 February 2018; available online 9 March 2018.

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Peer review under responsibility of Saudi Ophthalmological Society, King Saud University

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Fig. 1. (Top) Clinical photograph of the patient with protruding prosthesis at the right side. (Bottom) Same patient, without prosthesis. A pink subconjunctival mass was visible.

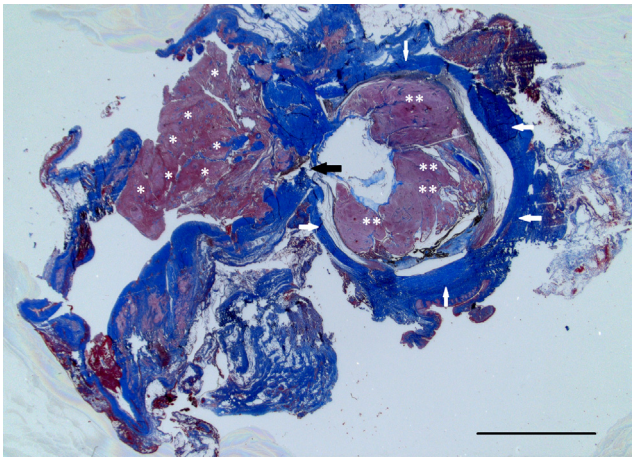

Fig. 3. Overview of biopsy specimen with orbital gliotic tumour (*) and retinal tumour (**) in red color; sclera (white arrow) and fibrous tissue in blue color; coloboma (black arrow); pigment epithelium remnants in black. (Masson's Trichrome stain, bar = 5 mm).

of the medial orbital wall without sinus invasion. Previous radiologic scans taken at birth were not available. Via anterior orbitotomy through conjunctival incision, a large, soft orbital mass without capsule and strongly adherent to the microphthalm eye was removed en-bloc with the eye. A spherical acrylic ball covered with donor sclera was implanted and the shallow inferior fornix was restored with deepening sutures.

On pathologic examination, the specimen consisted of a soft tissue mass and a small rudimentary eyeball with a diameter of 10 mm with a colobomatous opening at the posterior sclera. (Fig. 3) The cornea lacked Bowman's layer and the Descemet's layer was reduced to irregular endothelial cells. The anterior chamber lacked a pupil, lens and trabecular meshwork, and the posterior chamber was absent. The iris displayed normal dilator myofibers and a pigmented posterior surface with irregular ingrowths of clumps of pigmented epithelial cells. The ciliary body and retinal pigmented epithelium were normally developed, and the choroid was thickened. Overall, the content of the eye was filled with sheets of spindle shaped cells with fibrillar eosinophilic cytoplasm and long oval, large nuclei, focally atypical with inhomogeneous distribution of chromatin and large nucleoli (Fig. 4). Rosenthal fibers were present. The rudimentary optic nerve was partially replaced by bundles of collagen fibers. Examination of the soft tissue orbital mass, which was adherent to the eyeball, revealed eosinophylic spindle shaped cells similar to those in the eyeball. There was abundant calcification, and old hemorrhages and hyalinized blood vessel walls were found.

The retinal and orbital tumor cells stained homogeneously strongly positive for glial fibrillary acidic protein (GFAP) (Fig. 4). MIB-1 (Ki67-proliferation index or cell-cycling marker) was positive in approximately 1–4% of tumor cells. There were no immunoreactivity for p53 and mutated IDH1 (isocitrate dehydrogenase 1). Genetic analysis after DNA extraction did not show BRAF-mutations. The histological findings were in compliance with pilocytic astrocyoma WHO grade I originating from the retina, but, because of negative

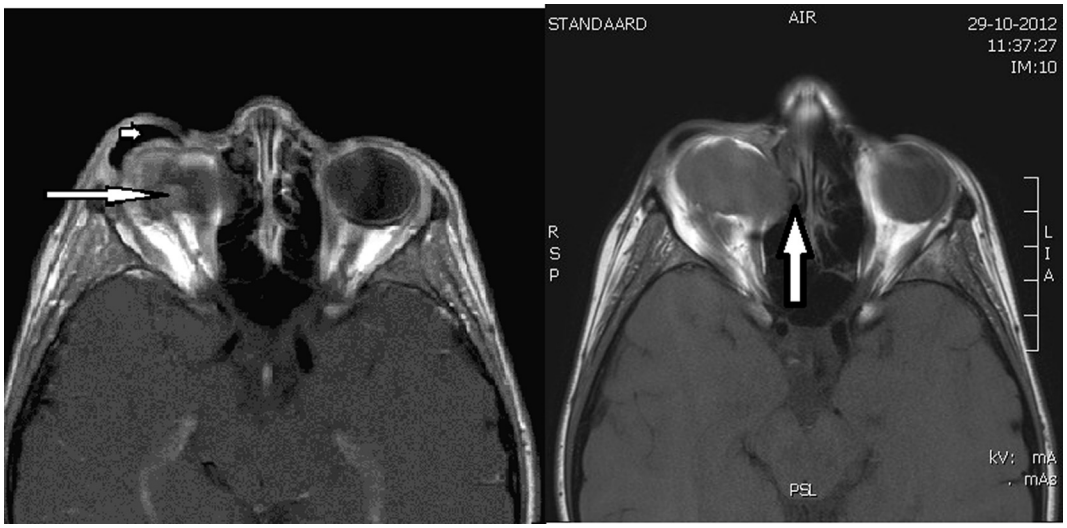

Fig. 2. MRI scan of the orbit, axial views. (Left) Microphthalmic eye (long arrow), with preserved lacrimal gland, horizontal rectus muscles and optic nerve structures. Notice the ocular prosthesis. (short arrow). (Right). Well-delineated, multilobular orbital mass in the microphthalmic socket. Notice the bowing of the medial orbital wall (arrow). There is limited image quality owing to orthodontic braces.