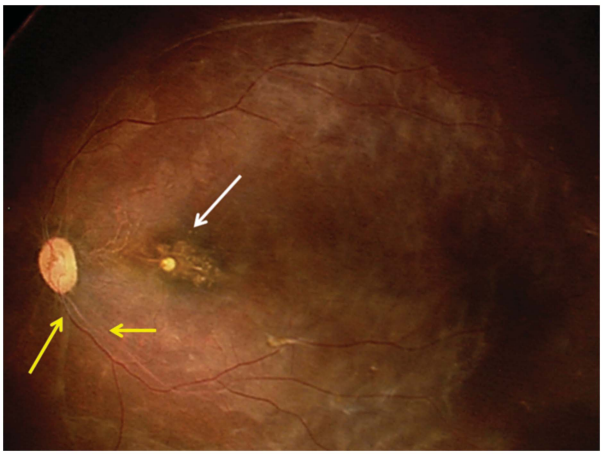

Figure 1 Fundus photograph of the left eye showing posterior segment involvement with sclerosed blood vessels and a pale optic disc (yellow arrows) and retinal infiltrates (white arrow).

absence of skin or systemic lesions. Consideration of this condition may prevent needless investigations for child protection.

Conflict of interest

The authors declare no conflict of interest.

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Sir,

Medulloepithelioma in DICER1 syndrome treated with resection

Medulloepithelioma is usually unilateral and arises from the nonpigmented epithelium of the ciliary body and rarely from the optic nerve. They generally occur in the first decade of life and present as a fleshy pink tan mass. We report here a familial cancer predisposition syndrome, which is not well documented in the ophthalmology literature.

Case report

A 16-year-old Caucasian female presented with blurred vision in her right eye. Her past medical history included ovarian Sertoli–Leydig tumor treated with resection, thyroid papillary tumor treated with thyroidectomy, pinealoblastoma treated with chemo-radiation, chronic renal insufficiency, and cystic disease of the kidneys and lungs. Her left eye was prephthisical secondary to anterior segment dysgenesis and had no light perception since early childhood. Visual acuity in the right eye was 20/60 and anterior segment examination showed posterior subcapsular and cortical cataractous changes. Fundus examination showed a white mass arising from the ciliary body (Figure 1a) with partial retinal detachment (Figure 1b), subretinal fibrous bands, and a giant retinal tear inferotemporally. Ultrasonography showed a densely hyper-reflective lesion of 8-mm thickness. Incisional biopsy via a lamellar scleral flap revealed cartilage suggestive of teratoid medulloepithelioma. The patient underwent transcleral excision of the residual ciliary body tumor with pars plana lensectomy, pars plana vitrectomy, partial retinectomy, silicone oil injection, and scleral buckling. Pathology showed scarce neuroepithelial cells with no malignant features and abundance of cartilaginous tissue consistent with teratoid cartilaginous medulloepithelioma of the ciliary body. At the second postoperative month, she had a visual acuity of 20/80 with aphakic glasses and her retina was attached. There was no evidence of recurrence 6 months postoperatively. Initial genetic testing was inconclusive for the *DICER1* gene mutation and the sample is being preserved for future study.

Comment

DICER1 gene (OMIM 601200) is a member of the ribonuclease III family and is inherited as an autosomal dominant trait with cytogenetic location 14q32.13.[1] The reported tumors with DICER1 syndrome are described in Table 1.[2] Priest *et al*[3] reported four patients with medulloepithelioma and pleuropulmonary blastoma, which possibly indicated DICER1 syndrome. Previous reports of local resection for medulloepithelioma have shown poor response with recurrence necessitating enucleation.[4,5] The role of chemotherapy and radiation therapy in patients with medulloepithelioma has not been well documented. Our patient showed no evidence of recurrent disease at last

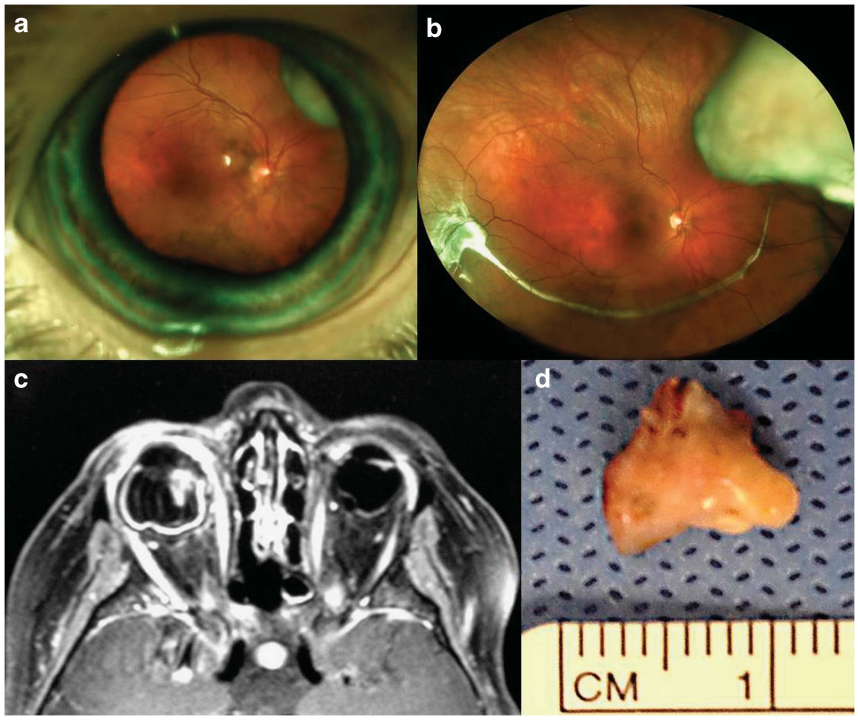

Figure 1 Fundus photograph showing a white nodular ciliary body mass superonasally (a) with subretinal fibrous bands and inferior retinal detachment (b). Axial MRI using T1-weighted image showed a bright signal intraocular mass (c). Gross histopathology revealed a triangular tissue with pearly surface and pigment on one side, suggesting ciliary body pigment epithelium (d).

Table 1 Characteristics of DICER1 syndrome

Tumor features	*Tumor features (possible)*
Cystic nephroma	Cervical cancer
Hamartomatous polyps in small intestine	Colon cancer
Ovarian Sertoli–Leydig cell tumor	Medulloepithelioma
Pleuropulmonary blastoma	Medulloblastoma
Thyroid hyperplasia/ goiter	Primitive neuroectodermal tumor (PNET)
	Rhabdomyosarcoma
	Seminoma
	Synovial sarcoma
	Thyroid adenoma
	Thyroid cancer, papillary
	Wilms' tumor (nephroblastoma)

follow-up 6 months following surgical resection, but is being followed up closely.

Conflict of interest

The authors declare no conflict of interest.

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