Sialidosis (mucolipidosis type I) is a rare inherited lysosomal storage disease characterized by deficiency of α-N-acetylmuramidase (sialidase) in leukocytes and cultured fibroblasts. Results in intracellular storage of excess sialyloligosaccharides and is histologically observed as abnormal vacuolization of various cell types. Two major phenotypes of mucolipidosis exist: type I or the cherry red macula (congenital form) and type II. Sialidosis has an autosomal recessive pattern of inheritance, and the gene has been localized on chromosome 6p21.2 It occurs in 1 of every 2,200,000 live births. Patients with type II disease have somatic changes, characterized by coarse facies, hepatomegaly, bony changes of dysostosis multiplex, and developmental delay. Patients with type II disease usually die within the first 2 years (congenital form) or by the second decade (infantile form) of life. By contrast, patients with type I disease are less severely affected and typically develop symptoms of myoclonic epilepsy, visual problems, and ataxia in the second or the third decade of life. Macular cherry red spots are always present and, therefore, sialidosis should be included in the differential diagnosis of a cherry red macula in this clinical setting. Diagnostic evaluation for a patient with a cherry red spot in the macula not due to arterial occlusion should include a genetic history and an appropriate laboratory workup to confirm the underlying cause.

Conclusions. We herein present a rare case of a 14-year-old boy with mucolipidosis type I who had cherry red spots in the maculae of both eyes. The patient had minimal symptoms of myoclonic epilepsy, and visual acuity correctable to 20/20 OU. Of interest in this case are the fluorescein angiogram and optical coherence tomography findings. The fluorescein angiogram shows blocked fluorescence surrounding the fovea throughout the angiogram (eFigure 1). The optical coherence tomography shows increased reflectivity in the glio-neuronal cell layer corresponding to the blocked fluorescence (eFigure 2). Although pathological confirmation is lacking in this case, a previous autopsy report of a patient with mucolipidosis type I demonstrated diffuse intracytoplasmic accumulation of lipofuscin-like pigment in the cerebral neurons. This leads us to speculate that the ophthalmoscopic changes observed in our patient are due to an accumulation of sialyloligosaccharides in the inner layers of the retina.

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Macular Retinal Detachment in Hallermann-Streiff Syndrome

Hallermann-Streiff syndrome (also known as oculomandibulofacial syndrome) is a rare syndrome chiefly comprising facial and ocular abnormal...
malities. Cataract and microphthalmos are the most apparent ocular features, but retinal abnormalities may be the primary cause of poor vision.

**Report of a Case.** A 1-week-old girl of European descent was initially seen with bilateral cataracts. She was small for her 35-week gestational age. She exhibited microcephaly with birdlike facies, natal teeth, sparse fine hair, and long hyperextensible fingers consistent with features of Hallermann-Streiff syndrome (Figure 1).

Ophthalmologic examination revealed bilateral microphthalmia, horizontal corneal diameters of 6 mm, and bilateral visually significant cataracts preventing view of the ocular fundi. Cataract extractions were performed in this patient at 5 and 6 weeks of age. Indirect ophthalmoscopy revealed bilateral macular serous retinal detachments with horizontal retinal folds through the macula, as well as peripapillary and peripheral retinal pigment epithelial mottling (Figure 2). Treatment of uveal effusion syndrome by means of scleral surgery was considered but was declined by the parents.

**Comment.** Hallermann-Streiff syndrome was first described by Charles Aubry in 1893. Franois analyzed 22 cases and described 7 essential signs for what he regarded as a new syndrome linked to the various hereditary ectodermal dysplasias. These signs are (1) dyscephalia and birdlike facies, (2) dental abnormalities, (3) proportionate short stature, (4) atrophy of skin (especially on the nose), (5) hypotrichosis, (6) bilateral microphthalmos, and (7) cataract. Most cases of Hallermann-Streiff syndrome are sporadic. Autosomal dominant inheritance has been suggested in some cases.

The retinal changes we observed were strikingly similar to the original drawing of fundus abnormalities in this syndrome by Franois. To our knowledge, there is no fundus photograph of a patient with Hallermann-Streiff syndrome in the English-language literature. A fluorescein angiogram (without red-free or color photography) of an exceptional case diagnosed at 11 years of age showed multiple areas of choroidal leakage. Uveal effusion in Hallermann-Streiff syndrome may not be uncommon if these findings of chorioretinal pigmentary changes, multiple areas of choroidal leakage, and serious retinal detachment represent different manifestations of uveal effusion syndrome in these patients. Indeed, a histopathologic case series of 8 eyes with uveal effusion included a case of Hallermann-Streiff syndrome that demonstrated abnormal scleral collagens.

Sclerectomy has been advocated for primary uveal effusion syndrome, but its efficacy in congenital uveal effusion in microphthalmic eyes is unknown. Early detection and management of retinal detachment in other patients having Hallermann-Streiff syndrome offer the chance of improved visual function.

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Resolution of Exudative Retinal Detachment From Retinal Astrocytoma Following Photodynamic Therapy

An 18-year-old woman with a visual acuity of 20/70 OD from an exudative macular retinal astrocytoma confirmed by needle biopsy was treated with photodynamic therapy (PDT). Subsequent resolution of subretinal fluid and intraretinal edema led to improvement in vision during 6 months.

Acquired retinal astrocytoma is a benign intraocular tumor typically located in the macular or juxtapapillary region.1,2 Despite its benign cytology, progressive growth, exudation, and secondary retinal detachment, acquired retinal astrocytoma can lead to poor visual acuity or enucleation.1,2 Current therapies include laser photocoagulation, plaque radiotherapy, external beam radiotherapy, and enucleation. In this report, we describe a patient with retinal astrocytoma who showed resolution of macular edema and exudation following PDT.

Report of a Case. An 18-year-old woman had an asymptomatic retinal mass with exudative retinopathy in her right eye. Visual acuity was 20/20 OU. She had no history of tuberous sclerosis complex or neurofibromatosis. The left eye had normal vision. In the temporal macular region of the right eye, there was an amelanotic retinal tumor with slight intrinsic grey pigmentation measuring 6.0 mm in diameter and 3.0 mm thick. There were slightly dilated feeding and draining vessels. Optical coherence tomography showed shallow subretinal fluid and macular edema. The clinical diagnosis was acquired retinal astrocytoma rule-out pigment epithelial adenoma. Transvitreal fine-needle aspiration biopsy through the pars plana revealed amelanotic bland spindle and stellate cells with cytoplasmic processes without mitoses.

Immunohistochemistry showed a strong reaction to glial fibrillary acidic protein and a negative reaction to melanoma-specific antigen (HMB-45), suggesting the diagnosis of astrocytoma. Observation was advised.

Nine months later, the exudation had increased, but visual acuity was stable. Subsequently, visual acuity decreased from macular exudation, edema, and subretinal fluid to 20/25 OD (at 15 months) and 20/60 OD (at 16 months). Argon la-