A 10-Year-Old Female with Acute Onset of Superior Vision Loss

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Introduction:

A 10-year-old girl presented as a new patient with acute onset of superior vision loss in the right eye. She described a scotoma just superior to fixation in the right eye, with no changes in the left. The patient denied any prior history of vision problems or systemic illness. Her mother did report a vitreous hemorrhage at age 30, which was treated with laser. No other family history of vision problems was reported.

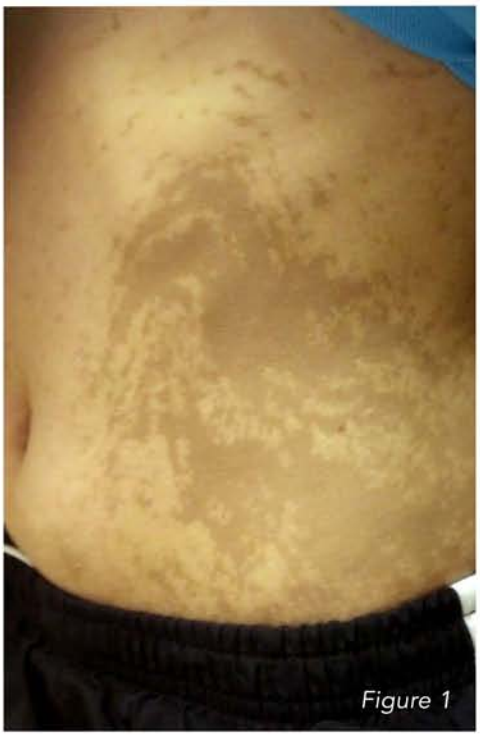


Figure 1

On exam, the patient was 20/25 OD and 20/20 OS. Pupils, confrontation visual fields, motility, ocular alignment and slit lamp exam were all normal. The patient did have a diffuse, swirling, hyper-pigmented rash covering her torso and extremities (Figure 1). She also had severe dental hypoplasia (Figure 2). Fundus exam showed a vitreous hemorrhage in her right eye inferior to the fovea with lipid exudation into the fovea and apparent neovascularization of the disc (Figure 3).

The posterior pole of the left eye was normal (not shown). There were no abnormalities found in the peripheral fundus on indirect ophthalmoscopy in either eye (not shown). Fluorescein angiography showed blockage from the vitreous hemorrhage, hyperfluorescence around the optic nerve consistent with neovascularization of the disc and some areas of apparent capillary dropout (Figure 4a-b). Peripheral shots were limited given patient cooperation. She was brought back to clinic a few days later for wide field angiography.

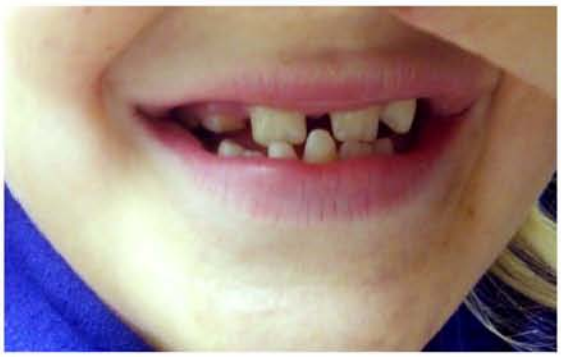


Figure 2

Photos of the posterior pole at the time were minimally changed (Figure 5). Fluorescein angiography showed significant peripheral vascular dropout in both eyes, with evidence of neovascularization of the disc and the periphery in the right eye (Figure 6a-c). Options were discussed with the patient and her mother including pan-retinal photocoagulation (PRP), anti-VEGF therapy and/or vitrectomy of the right eye with possible PRP to the left eye. After extensive discussion with the patient and her mother, the patient underwent PRP to both eyes.

Discussion:

Incontinentia pigmenti (IP) is a rare X-linked dominant disorder found only in females, as the mutation is lethal in males. It is a generalized ectodermal dysplasia that involves the eyes, hair, teeth and central nervous system.

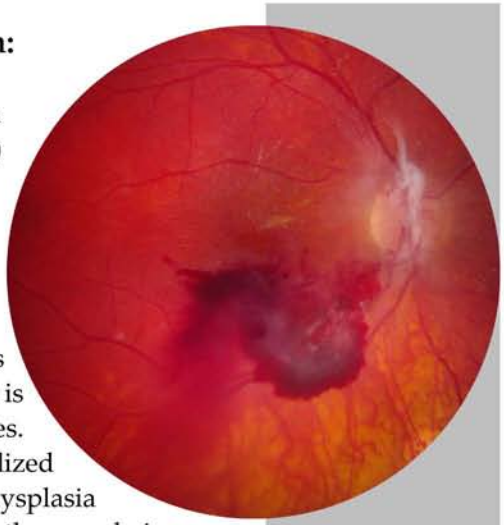
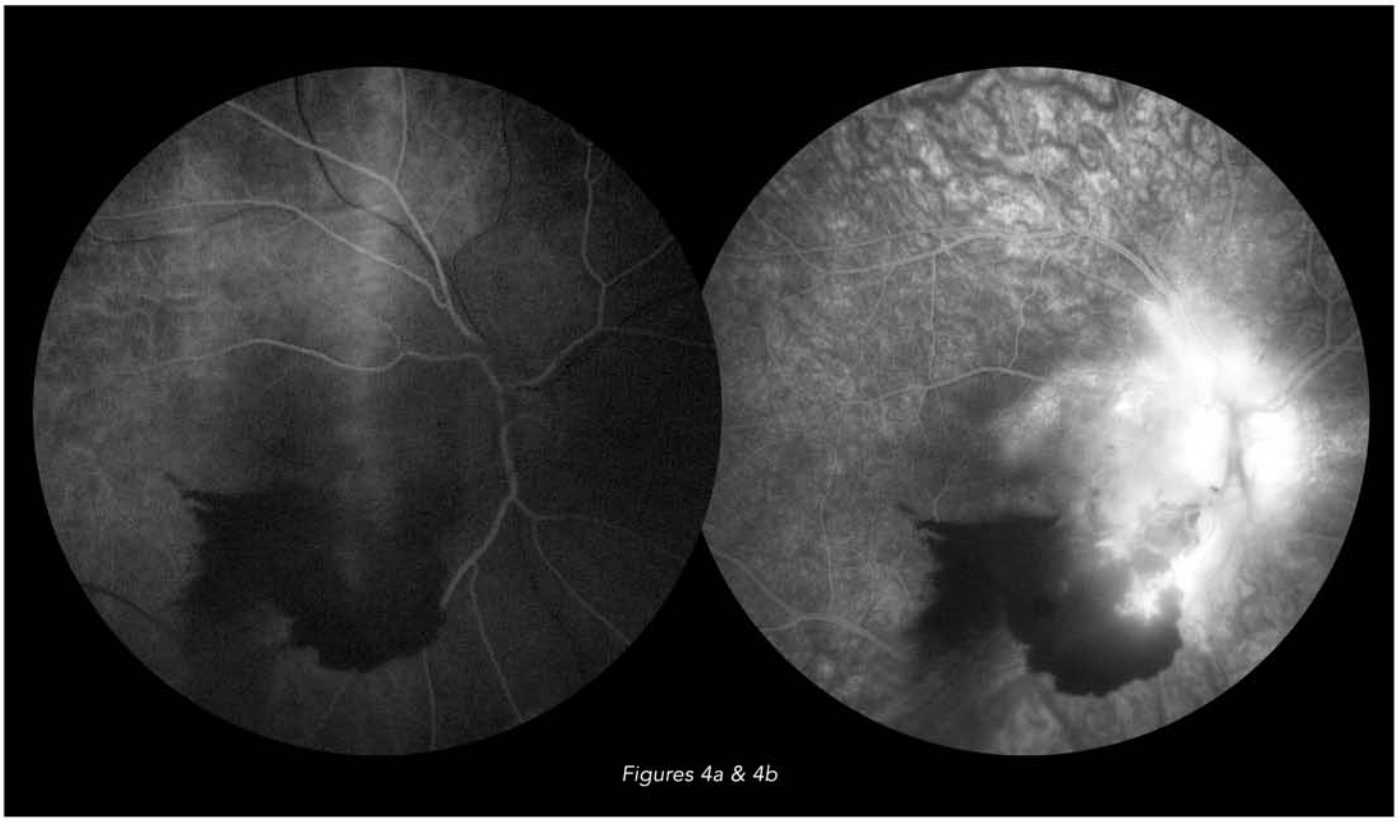


Figure 3



Figures 4a & 4b

Many different systemic findings are seen in IP. The skin findings often appear shortly after birth, both on the torso and extremities. They usually first appear as bullous, erythematous lesions that progress to a verrucous appearance and may then further develop into the flat, pigmented macules arranged in whorls, as seen in our patient. In some patients, the pigmentation may fade later in life. Many central nervous system abnormalities have been observed including seizures, mental retardation and rapidly progressive neonatal cerebral ischemia. Other systemic findings include alopecia and dental hypoplasia, both seen in our patient.

Numerous ocular abnormalities have been described including cataract, high refractive error, nystagmus, corneal opacities, thin sclera, strabismus, optic atrophy and conjunctival pigmentation. In regards to the fundus, there are a number of presentations including retinal vascular anomalies (including arteriovenous malformations), vascular dilation, peripheral retinal nonperfusion with or without neovascularization, retinal detachments, chorioretinal atrophy, macular dragging and foveal hypoplasia (1-2). Findings can be unilateral or bilateral. Ocular findings can be seen at birth, with rapid progression to detachment and phthisis in some as early as 3-4 months, while showing up later in life in other patients.

The differential for incontinentia pigmenti includes other pediatric vascular disorders including retinopathy of prematurity and familial exudative vitreoretinopathy. Although there is no consensus on treatment, photocoagulation to the avascular retina is generally recommended, especially with the development of retinal vascular proliferation, to prevent detachment of the retina and vitreous hemorrhage. Outcomes are variable.

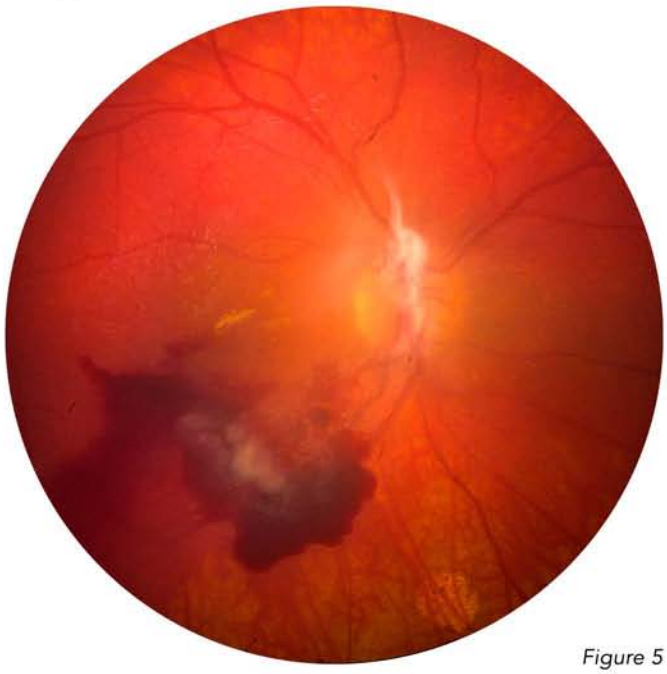
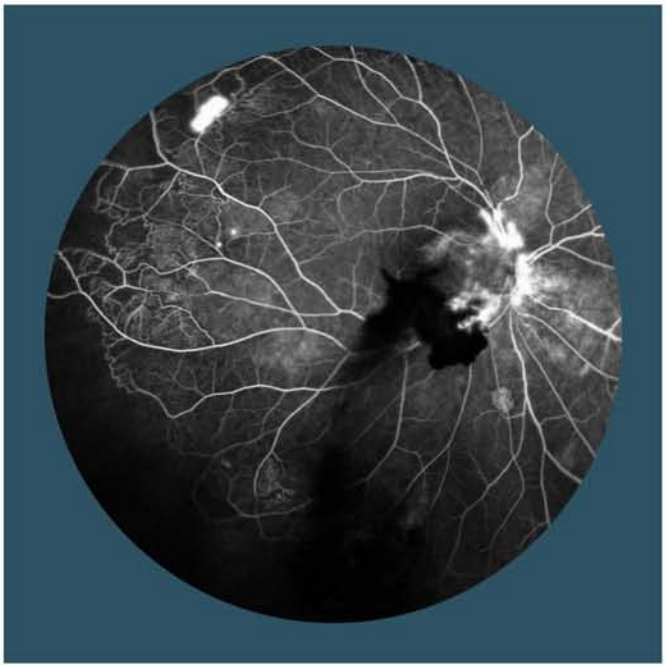
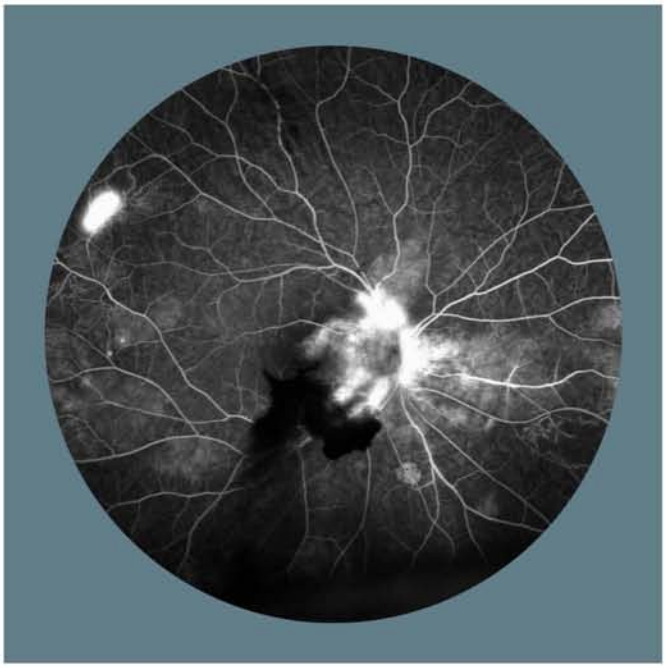
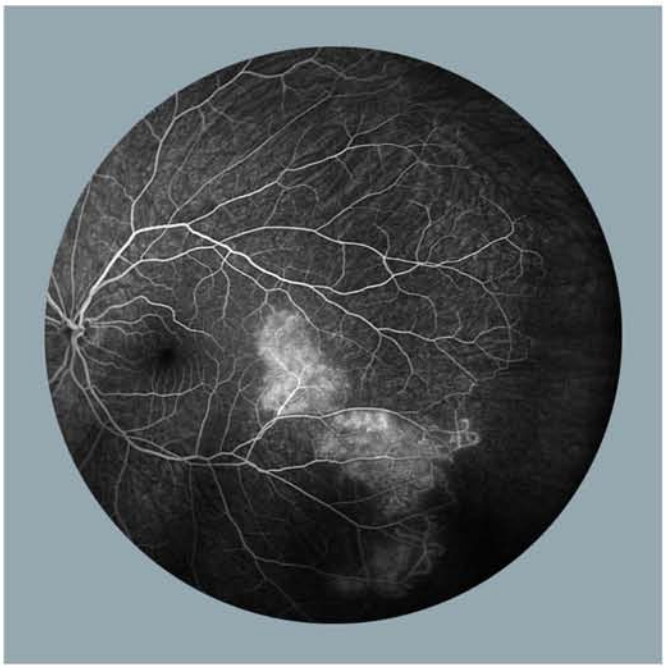


Figure 5





The cause for the disorder is not totally known. About 80% of patients have a mutation in the inhibitor of the kappa light polypeptide gene enhancer, kinase gamma, IKBKG. Mutations of this gene lead to activation of eotaxin, an eosinophil chemokine. Vaso-occlusion occurs from accumulation of eosinophils in and around blood vessels, leading to the vascular abnormalities seen in IP (3).



Figures 6a, 6b, 6c

References:

1) Holmstöm G, Thorén K. Ocular manifestations of incontinentia pigmenti. Acta Ophthalmol Scand. 2000. 78(3):348-53.

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3) Smahi A, Courtois G, Vabres P, et al. Genomic rearrangement in NEMO impairs NF-kappaB activation and is a cause of incontinentia pigmenti. The International Incontinentia Pigmenti (IP) Consortium.Nature.2000.405(6785):466-72.