A 17-Year-Old Female with Poor Vision Since Childhood

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Introduction:

A 17 year old girl presented as a new patient complaining of poor vision in both eyes since childhood, with some decline in vision in the past few months. She was seen at a regular interval by optometry, who had prescribed glasses and soft contact lens. The patient presented with a diagnosis of amblyopia of the left eye, without any suggestive cause such as prior strabismus or visual pathway obstruction. On initial exam, best corrected vision was 20/40 OD and 20/60 OS. On external exam, the patient was noted to have very fair skin and hair color (Figure 1). The anterior segment exam was normal, besides lights colored irises and faint transillumination defects seen on retro-illumination (not shown). Fundus exam was remarkable for a poor foveal reflex in both eyes with a very blonde appearing fundus (Figure 2). Red free imaging showed very prominent choroidal vasculature with a very small or absent foveal avascular zone. This was confirmed on fluorescein angiography, where again there did not appear to be a foveal avascular zone (Figure 3). Optical coherence tomography (OCT) demonstrated an otherwise normal retinal architecture with an absent foveal depression in both eyes (Figure 4). A diagnosis of oculocutaneous albinism (OCA) was made given the absence of skin pigmentation and significant foveal hypoplasia.

Figure 1

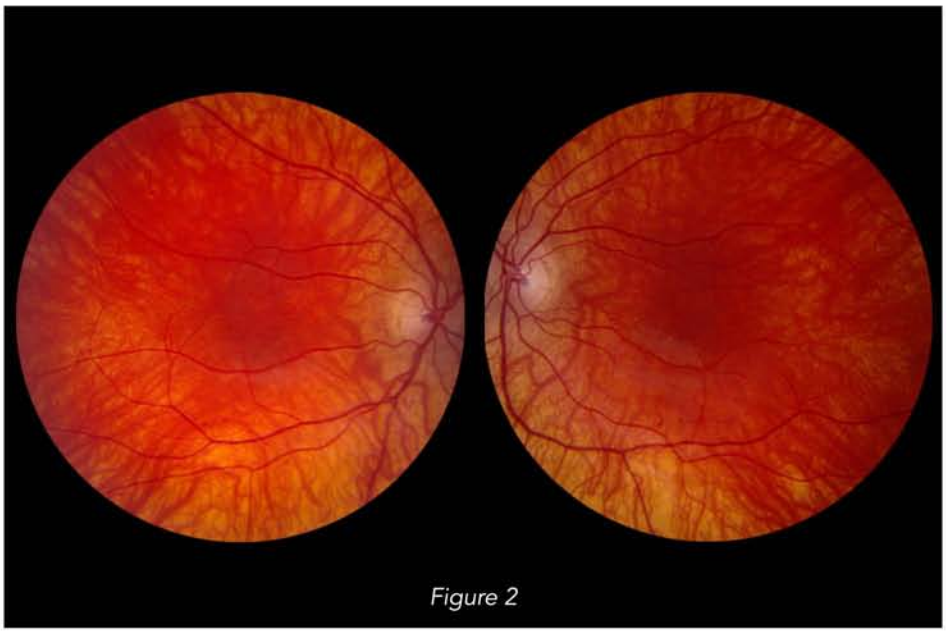

Figure 2

Discussion:

Albinism, a group of disorders characterized by errors in melanin production, is broken down into two major classifications. Ocular albinism (OA) occurs when only affecting the eyes and oculocutaneous albinism (OCA) is when the skin, eyes and hair all appear hypopigmented. Its prevalence worldwide is about 1/17,000. The classic ocular exam findings of both types of albinism include iris transillumination defects due to decreased iris pigmentation, foveal aplasia or hypoplasia and reduced or absent pigment in the retinal pigment epithelial layer giving the appearance of a blonde fundus. Visual acuity can be variable, ranging from about 20/40 to about 20/200. Patients often present due to poor central acuity, nystagmus and photophobia. The severity of visual impairment is often proportional to the degree of foveal hypoplasia and pigmentation. In severe cases, nystagmus can present as early as 2-3 months of age.

Other findings typical of albinism include high refractive errors and significant rates of strabismus. Patients with albinism often have an abnormally high rate of crossing fibers in the optic

chiasm compared to normal individuals. This often limits stereopsis and can lead to strabismus. This finding can be deduced by asymmetric monocular visually evoked potentials.

Diagnosis of albinism is largely clinical. There should be a high level suspicion for the disorder in individuals with minimal skin and ocular pigmentation with subnormal visual acuity. These patients should all undergo a full ophthalmic examination, including checking for refractive error, ocular alignment, presence of transillumination defects and careful analysis of the fundus. OCT is useful to better evaluate the foveal contour. Definitive diagnosis, however, requires genetic evaluation.

OCA is actually a group of 4 autosomal recessive conditions with similar presentation. OCA1 is due to a mutation in the tyrosinase gene. Depending on whether the gene is completely inactive (OCA1A) or minimally active (OCA1B), the patients may have variable degrees of pigment and may even tan. These patients often present with the poorest visually acuity. OCA2 is caused by a mutation in the OCA2 gene, which helps tyrosinase function. These individuals usually have minimal pigmentation at birth, which increases over time. This is the most prevalent type worldwide, especially in African populations. OCA3 is caused by a mutation in the tyrosinase-related protein 1 gene. These individuals often present with red or reddish brown hair. Most patients are of African descent. Visual abnormalities are often mild in this variant. Finally, OCA4 is caused by mutations in the membrane-associated transported protein gene. They have a similar phenotype to those individuals with OCA2.

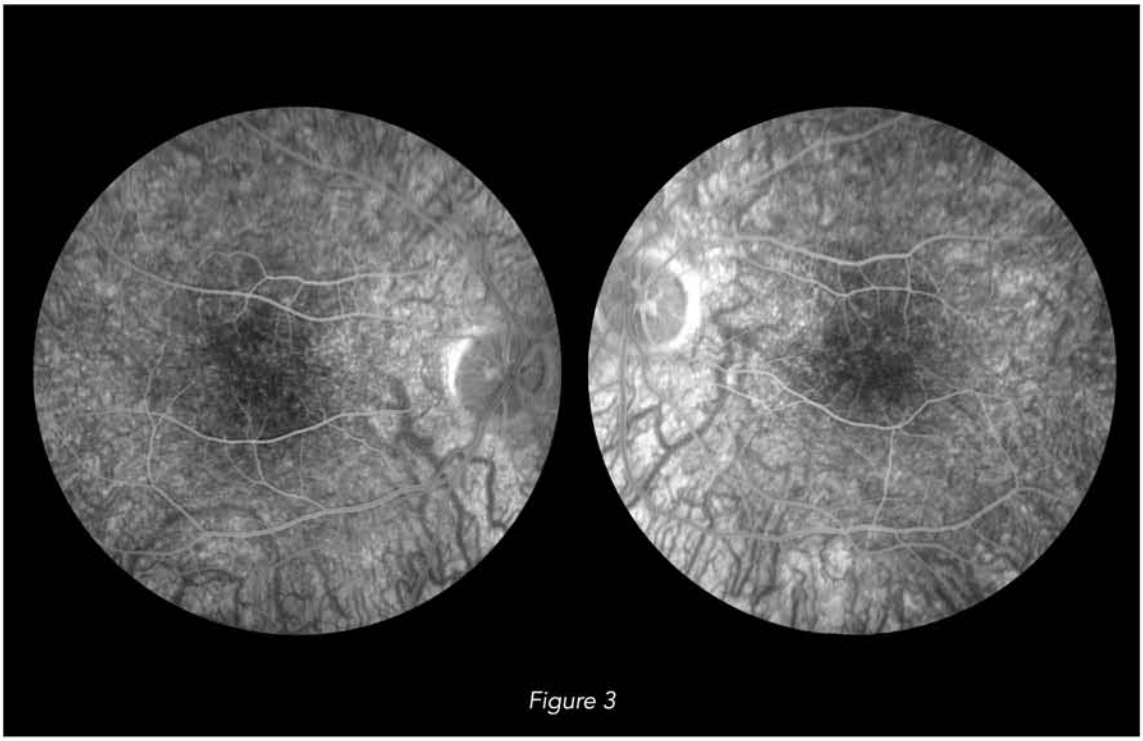

Figure 3

Unlike OCA, OA is often X-linked, while autosomal recessive forms have been described. These individuals have decreased pigmentation of the eyes only, with normal appearing skin.

Albinism can be part of a larger systemic disorder, including two lethal autosomal recessive variants. Hermansky-Pudlak syndrome is characterized by poor platelet function, leading to bleeding abnormalities, interstitial lung fibrosis, granulomatous colitis and severe immunological deficiencies from neutropenia and lack of killer T cells. This syndrome is very rare, except in people of Puerto Rican decent, where it affects about 1/1,800 individuals. Chediak-Higashi syndrome is a disorder of white blood cell dysfunction, leading to an increased susceptibility to bacterial infections.

Currently, there is no known treatment for albinism. Patients are often co-managed with optometry and low vision services. Individuals are often fitted with either tinted or photochromic lens to reduce photophobia. Many patients require the use of low vision aids such as magnifiers and closed circuit televisions. Treatment also requires the aid of a pediatric ophthalmologist to manage patching therapy and possible muscle surgery for strabismus and nystagmus. All patients with albinism should, of course, be advised to use judicious sun protection given increased risk of skin burns and malignancy.

References:

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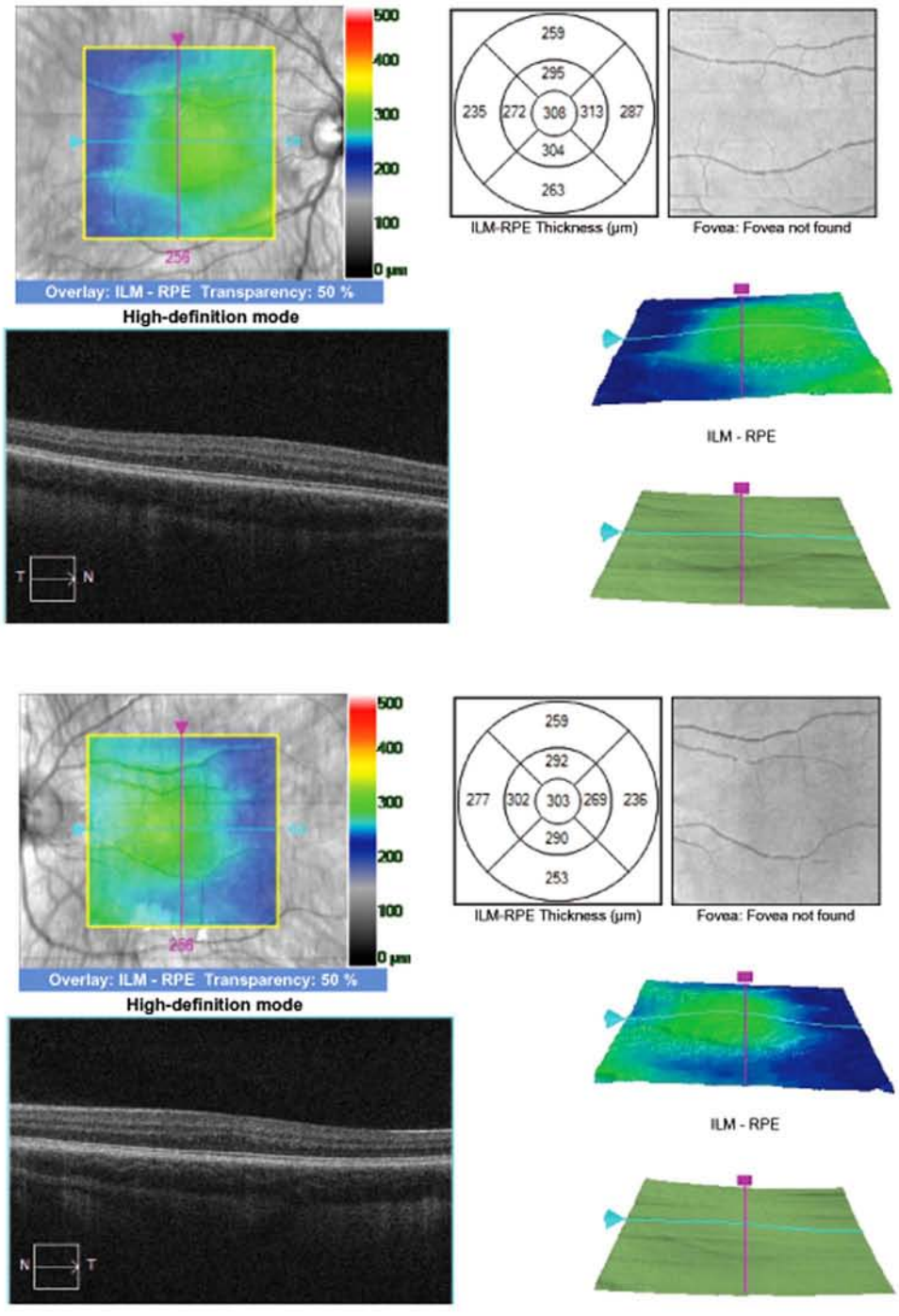

Figure 4

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