# Foveal Hypoplasia in Oculocutaneous Albinism and the Role of OCT

**Abstract**

We describe a case of a 16 year old girl with Oculocutaneous albinism (OCA) who presented to us with photophobia and blurring of vision. She showed typical features of OCA, blonde hair, refractive error, iris transillumination and foveal hypoplasia. Our report highlights the role of macular OCT in diagnosis of foveal hypoplasia which is an essential feature of OCA.

**Keywords:** Oculocutaneous; Albinism; Foveal hypoplasia

Case Report

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## Introduction

Oculocutaneous albinism (OCA), is a group of autosomal recessive disorders of melanin biosynthesis that are characterized by congenital hypo pigmentation of ocular and cutaneous tissues. Clinical manifestations may include reduced visual acuity, refractive errors, colour vision defects, photophobia, nystagmus, iris transillumination, reduced retinal pigment, foveal hypoplasia and abnormal chiasmal decussation [1,2]. Earlier onset of nystagmus correlates with degree of foveal hypoplasia. History of easy bruising or recurrent infections in patients with Hermansky-Pudlak syndrome and Chediak-Higashi syndrome, respectively.

## Case Presentation

A 16 year old girl presented to us at the Aberystwyth Eye Centre, Hywel Dda University Health Board, with photophobia and blurred distance vision. She was not sure how long this was but put it down to six months.

She had no problem with near vision. She was a simple myope. She did not tan when exposed to sun. There was nothing of significance in her past ocular history. She was a full term normally delivered child with a normal birth weight. She was not on any medications and did not have a significant family history.

Her examination showed.

i. Visual acuity: Right Eye 6/6-3 (No improvement with pin hole), and Left Eye 6/9+ (Pin hole 6/6-2). Her near vision was Right and Left N5.

ii. Refraction: Right-1.00 Diopter Spherical 6/6 Left-1.00 Diopter Spherical 6/6. No RAPD, Colour vision with Ishihara's Colour Vision Test Plates was Right 12/12 and Left 12/12. There was a small esophoria with good recovery, 55" arc stereoacuity, full ocular motility and No Proptosis / Synkinesis or Nystagmus.

iii. Slit lamp examination: RIGHT, LEFT Iris transillumination (Figure 1). She was blonde, did not tan and showed mild photosensitivity. Fundus examination revealed bilateral foveal hypoplasia (Figure 2,3). An OCT of the macula was done and this confirmed bilateral foveal hypoplasia (Figure 4-6).

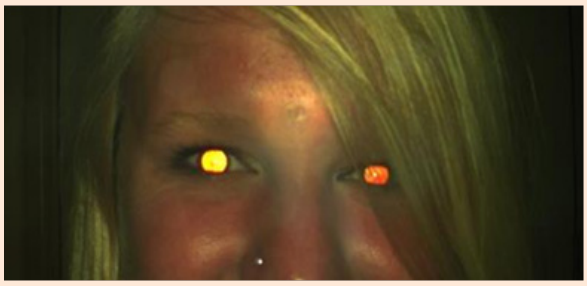

**Figure 1:** Photograph showing iris transillumination.

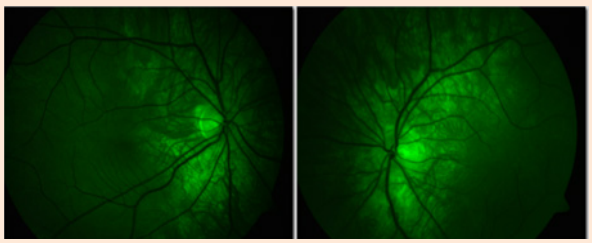

**Figure 2:** Red Free fundus Photographs of the Right and Left eye showing Foveal hypoplasia.

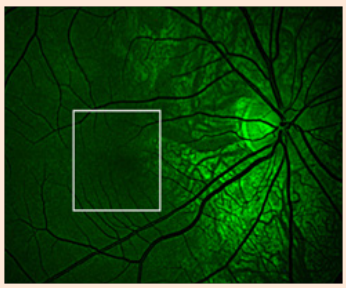

**Figure 3:** Foveal hypoplasia under higher magnification.

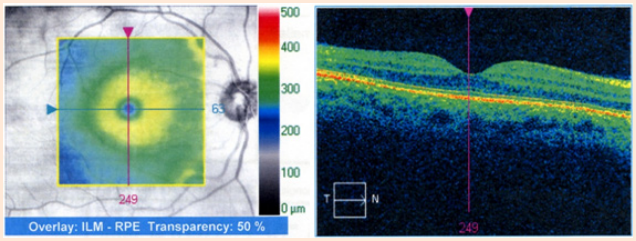


**Figure 4:** OCT Macula in a Normal Eye.

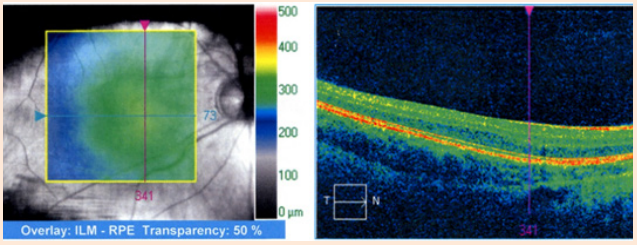


**Figure 5:** OCT Macula Right Eye.

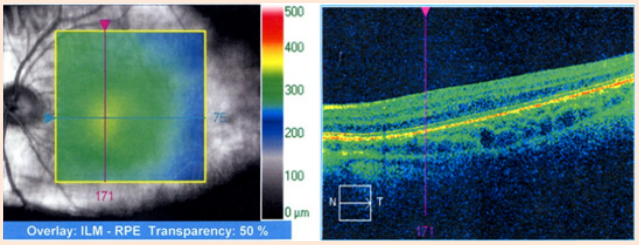


**Figure 6:** OCT Macula Left Eye.

## Discussion

Our patient presented with photophobia and blurring for six months, however the history was not reliable. She had a BCVA of 6/6 in each eye, the visual acuity did not improve further. Given the morphological appearance of the foveae on clinical examination and on OCT, the blurring was likely due to foveal hypoplasia.

Foveal hypoplasia has been graded by some according to the OCT appearance. At one end was Grade 1 with features of shallow foveal pit, presence of outer nuclear layer widening, presence of outer segment lengthening and at the other end was grade 4 with absence of outer nuclear layer widening. There was significant difference in visual acuity. Grade 1 was associated with the best visual acuity whereas grades 2, 3, and 4 were associated with progressively poorer acuity [3].

The Macular OCT of our patient showed the following features:

i. Absence of foveal pit.

ii. Absence of the small elevation of the inner segment/outer segment junction at the fovea.

iii. Presence of multiple inner retinal layers at the centre of the fovea.

These findings have also been described in studies by various colleagues [4-6]. She was advised to wear photo chromatic / dark glasses whenever she found photophobia bothersome. We also referred her to the Institute of Medical genetics where she was given genetic counselling. While the role of OCT has been utilized in the diagnosis and management of conditions such as Diabetic macular edema, Age related macular degeneration, Stargardt's disease [7,8] and Retinitis Pigmentosa [9], in recent years there has been a lot of interest on the role of OCT in Albinism [5,10].

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