

# Foveal avascular zone in oculocutaneous albinism

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## SUMMARY

Optical coherence tomography angiography imaging in two patients with oculocutaneous albinism, one with severe nystagmus, showed persistence of both the superficial and the deep retinal capillary plexus adding another vascular feature to the foveal hypoplasia.

## BACKGROUND

Oculocutaneous albinism (OCA) is a group of autosomal recessive disorders of melanin biosynthesis with a wide clinical spectrum: most severe in OCA1A type from complete lack of melanin production throughout life, to the milder forms OCA1B, OCA2, OCA3 and OCA4 where some pigment accumulates over time.<sup>1</sup> The most common involved genes include *TYR*, *OCA2*, *TYRP1* and *MATP*.<sup>1</sup> A hallmark of albinism is foveal hypoplasia shown by OCT, with retention of the inner retinal layers in the fovea and a mean foveal thickness of 300 µm as opposed to 150 µm in a normal individual.<sup>2</sup> Little is known on the status of the superficial and deep vascular plexus of the fovea in OCA. We present the optical coherence tomography angiography (OCTA) findings in two cases of OCA (figure 1). The study received Institutional Review Board approval.

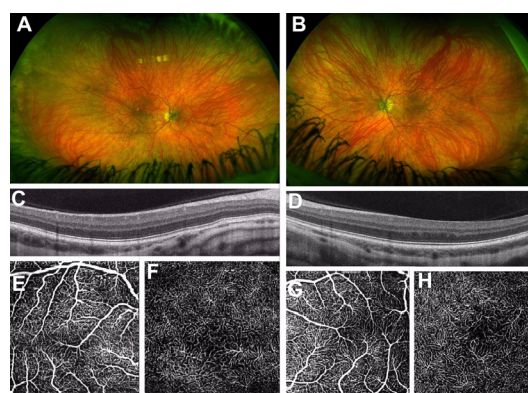
## CASE PRESENTATION

### Case 1

This 32-year-old Caucasian woman with OCA had a best spectacle corrected vision of 20/30 (6/9) bilaterally, -9.00 diopter myopia and absence of nystagmus. She had prominent iris transillumination defects, thickened foveae and albinotic fundi (figure 1) (Optos California, Optos, Marlborough, Massachusetts, USA). Optical coherence tomography (OCT) (Zeiss Cirrus 5000, Dublin, California, USA) and optical coherence tomography angiography (OCTA) revealed an immature macula and absence of both superficial and deep foveal avascular zone (FAZ) (Spectralis OCTA, Heidelberg Engineering, Heidelberg, Germany) (figure 1). Fundus autofluorescence showed the large choroidal vasculature (figure 2).

### Case 2

This 18-year-old Caucasian girl with OCA had a unocular spectacle corrected vision of 20/100 (6/30) in each eye and binocular vision of 20/80 (6/24) with 4 diopters of astigmatism. The macula was immature and thickened along with an albinotic fundus. The fast nystagmus was controlled by peribulbar anaesthesia allowing capturing a map of the retina and its vessels. OCTA (Revo NX by Optopol Technology SA, Zawiercie, Poland)



**Figure 1** Wide field fundus photography (A, B) shows an albinotic fundus bilaterally. SD-OCT (C, D) reveals a fovea plana in both eyes. 3×3 mm<sup>2</sup> macular scans fail to reveal a foveal avascular zone in superficial (E, G) and deep (F, H) capillary plexus of foveal region.

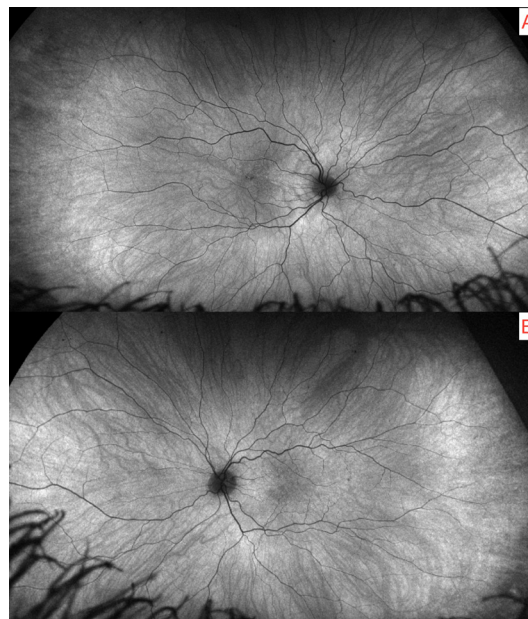
showed immature fovea with absence of both superficial and deep FAZ.

## INVESTIGATIONS

Patients declined genetic testing.

## OUTCOME AND FOLLOW-UP

Patients were lost to follow-up.



**Figure 2** Wide angle fundus autofluorescence of case 1 shows the large choroidal vessels in the right eye (A) and the left eye (B).



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## Case report

### DISCUSSION

Saunier *et al* found an absent superficial FAZ in a 12-year-old boy with OCA.<sup>3</sup> Monfermé *et al* had an unmeasurable superficial FAZ in a patient with OCA heterozygous with the R402Q variant of the *TYR* gene.<sup>4</sup> Pakzad-Vaezi *et al* showed no evidence of a FAZ in the superficial plexus with a small FAZ in the deep plexus in two OCA cases with bilateral visual acuities of 6/60 and 6/18, respectively, and with nystagmus.<sup>5</sup> Hamid *et al* detected absence of superficial FAZ with a small deep FAZ with normal vision and no nystagmus in a 30-year-old man with *OCA2* gene in the context of Prader-Willi syndrome.<sup>6</sup> Superficial and deep FAZ can be absent in nanophthalmos and in retinopathy of prematurity.<sup>7</sup> Also, Yokoyama *et al* found absent FAZ in 4 of 267 normal eyes of 255 patients with normal foveal depression, normal macular pigment and good vision.<sup>8</sup> OCTA acquisition times are longer than for standard OCT representing an additional challenge when working with eyes with fast nystagmus. In such eyes, visualisation of the capillary foveal plexus is possible by local ocular anaesthesia or by use of portable investigational OCTA devices.<sup>9</sup> More research is needed in analysing FAZ in OCA especially in eyes with severe nystagmus.

The macular pigment appears around 17 weeks gestational age and is likely one driving force to the formation of the FAZ.<sup>10</sup> Foveal pit formation does not take place until the FAZ starts to

form with morphology of the foveal pit correlating positively with the FAZ area.<sup>10</sup> It is hypothesised that lack of macular pigment halts FAZ formation which in turn delays also foveal pit formation.<sup>10</sup> The current study suggests that the persistence of both the superficial and the deep foveal capillaries may be involved in the aetiology of foveal hypoplasia in OCA.

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### Learning points

- ▶ A hallmark of oculocutaneous albinism (OCA) is foveal hypoplasia, with retention of the inner retinal layers in the fovea as shown on optical coherence tomography.
- ▶ Optical coherence tomography angiography showed persistence of both the superficial and the deep retinal capillary plexus in two cases with OCA adding another vascular feature to the foveal hypoplasia.
- ▶ Lack of macular pigment in OCA halts the formation of the foveal avascular zone which in turn delays the formation of a foveal pit.

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