# Clinical Phenotype and Optical Coherence Tomography in Patients with Achiasma

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## Introduction

- OCT is one of the most common techniques in assessing patients with retinal and optic nerve (ON) pathology.
- Achiasma is a rare visual pathway maldevelopment with reduced decussation of the axons in the optic chiasm. In the literature mainly single patients with achiasma have been described, with a maximum of nine patients examined .
- Limited literature is available regarding OCT in patients with achiasma.

## Purpose:

To investigate optic nerve and macular morphology in patients with achiasma using spectral-domain optical coherence tomography (SD-OCT).
Design: Prospective, case - control observational study.
Participants: 12 participants with achiasma (8 males and 4 females; 29.6±18.4 years (mean±SD) including 2 children 9 and 2 years old) and 24 gender-, age-, ethnicity- and refraction-matched healthy controls using a high resolution HH SD-OCT device (Copernicus; Optopol Technology S.A., Zawiercie, Poland (ADULTS); and Bioptigen Inc., Research Triangle Park, NC) without anaesthesia or sedation (FOR CHILDREN; Figure 1).
All patients had full ophthalmology assessment, eye movement recording (EyeLink 2, SR Research Ltd, Osgoode, Canada for ADULTS and EyeLink 1000, SR Research Ltd, Osgoode, Canada for CHILDREN ) and MRI scanning at 1.5 Tesla (Siemens Symphony, Erlangen, Germany) including sagittal MPRAGE (TR/TE/TI 1850/3.9/1100, flip angle = 15°, base resolution = 256, FOV = 300, voxel size 1.2mm x 1.2mm x 1.2mm) and axial CISS (TR/TE 11.4/5.7, flip angle 70°, base resolution = 256, FOV = 200, voxel size 0.8 x 0.8 x 1mm). )



Figure 1: A hand-held spectral domain portable device (Bioptigen Inc., Research Triangle Park, NC) 840 nm wavelength, 2.6 mm theoretic axial resolution was used. The advantage of the device is a separate probe that can be widely used in paediatric practice as patients' fixation is not required.

## Results:

- Visual acuity in achiasma patients was 0.63±0.19 and 0.53±0.19 for the right and left eye, respectively; mean refractive error (spherical equivalent) was -0.13 Dioptres.
- All patients had see-saw nystagmus, no stereo vision, strabismus was reported in 58.33% of patients and left (25%)/alternating (8.33%) head turn, or tilt (25%) also noted.
- OCT showed normal optic nerve structure and significantly thinner paracentral IS (p=0.002) with visual flattening of the ellipsoid line.
- 75% of patients had some degree of decussation in chiasm and in 25% (3 patients) crossing was fully altered as measured by MRI.

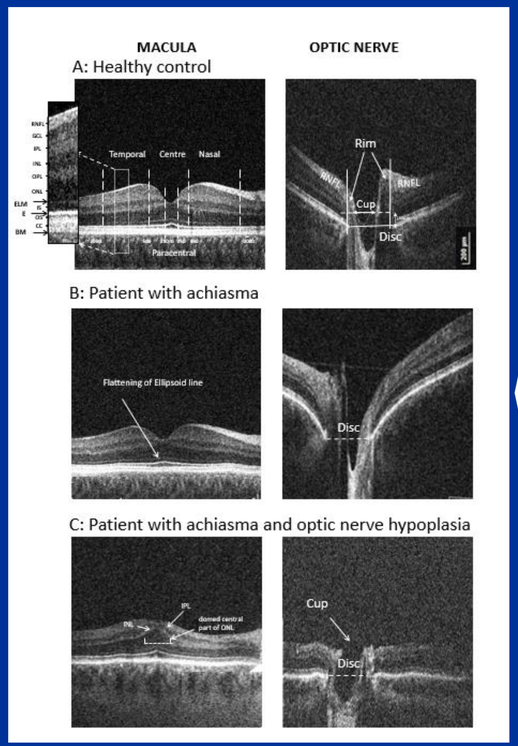



Figure 2: Horizontal OCT B-scan images of the macula area and the optic nerve head of a control subject (A), a patient with achiasma (B) and a patient with achiasma and co-existent optic nerve hypoplasia (ONH, C). Figure 1A (left) shows the position of the different retinal layers; RNFL = retinal nerve fibre layer; GCL = ganglion cell layer; IPL = inner plexiform layer; INL = inner nuclear layer; OPL = outer plexiform layer; ONL = outer nuclear layer; ELM = external limiting membrane; IS = inner segment; E = ellipsoid; OS = outer segment; CC = contact cylinder; BM = Bruch's membrane.
The horizontal macula scan of the patient with achiasma (1B) showed flattening of the ellipsoid line. The patient with co-existing ONH (1C) has also features typical for macula hypoplasia (thin retina, flat foveal pit, thin RNFL and GCL, continuation of the IPL and INL, domed central ONL and absence of the upward deflection of the ellipsoid line. Optic disc/cup size, peripapillary RNFL thickness of the patient with both achiasma and ONH (1C right) is considerably reduced.

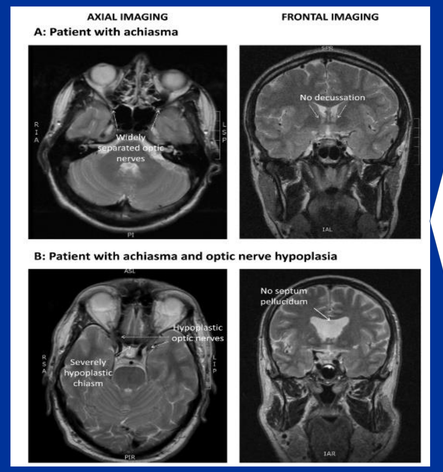



Figure 3:MRI of the patients with Achiasma and co-existing and septo-optal diplasia (B). A: Axial scan shows separated optic nerves without decussation. Coronal image shows separate non-crossed optic tracts in a patient with achiasma (A, right).
B: Axial scan indicates severe thinning of optic nerves in both sides with hypoplastic chiasma. On the coronal image of the same patient, the septum pellucidum is absent (B, right) .

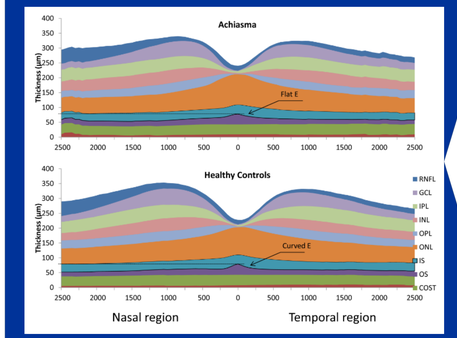



Figure 4: Cross sectional schematic diagram of individual retinal layers in patients with achiasma and controls. Horizontal dotted line shows the height of the ellipsoid line (E).

## Conclusions:

- This is the largest study of the achiasma patients and demonstrates for the first time changes in the fovea with IS thinning and ellipsoid line flattening.
- The optic nerve head was normal in 75% of patients despite severe changes in the chiasm.
- Co-existing foveal changes in 33.33% of patients (ONH, SOD) in our series and the literature suggest that these various abnormalities are different manifestation of events during foetal developmental occurring at different times and with different severity.