

Retinal and optic nerve changes in microcephaly

An optical coherence tomography study

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Study question

Is microcephaly associated with reductions in retinal thicknesses and optic disc diameters?

Summary answer

Microcephaly is associated with abnormally thin retinae. Although several patients had small optic discs, no statistical significance was reached for optic disc dimensions.

What is known and what this paper adds

Various ocular abnormalities have been reported in patients with microcephaly, and case series studies have reported retinal and choroid thinning. This study confirms, by means of quantitative handheld optical coherence tomography, that retinal and optic nerve abnormalities are common in children with microcephaly.

Participants and setting

This study recruited 27 children with primary microcephaly (15 girls; mean age, 9.4 ± 7.8 years) through the University Hospitals of Leicester. For each child with microcephaly, this study also recruited 1 age-, sex-, and ethnicity-matched healthy control (mean age, 9.0 ± 7.2 years) through National Health Service clinics in Leicestershire.

Design, size, and duration

The participants underwent ophthalmologic examinations that included best-corrected visual acuity measurements (where possible), cycloplegic refraction, orthoptic examinations, slit-lamp examinations, and funduscopy. The participants' retinae and optic discs were scanned with a handheld optical coherence tomography device, and the images were analyzed in ImageJ. General linear models were used to compare findings in the patients to those in the healthy controls.

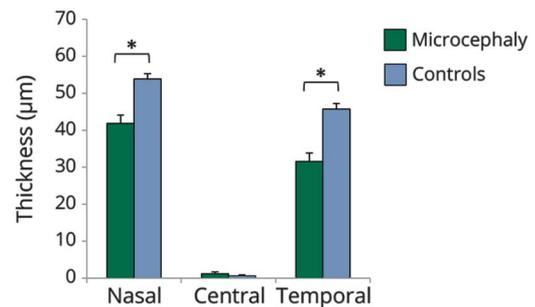
Primary outcomes measures

The primary outcomes were retinal thicknesses and optic disc dimensions.

Main results and the role of chance

Compared to the healthy controls, the patients with microcephaly had reduced parafoveal retinal thicknesses in the

Figure Mean thicknesses in an example retinal layer (ganglion cell layer) in the patient and healthy control groups



* $p < 0.05$.

nasal ($307 \pm 8.5 \mu\text{m}$ vs $342 \pm 3.7 \mu\text{m}$; $p < 0.001$) and temporal ($279 \pm 11.5 \mu\text{m}$ vs $325 \pm 3.1 \mu\text{m}$; $p < 0.001$) quadrants. The patients also had reduced total peripapillary retinal thicknesses in the temporal ($275 \mu\text{m}$ vs $318 \mu\text{m}$; $p < 0.001$) and nasal ($239 \mu\text{m}$ vs $268 \mu\text{m}$; $p = 0.013$) quadrants. Small optic discs occurred in several patients; however, no statistical significance was reached for the optic disc dimensions, possibly due to large variation.

Bias, confounding, and other reasons for caution

The underlying microcephaly etiologies were heterogeneous and sometimes unknown. Some patients were taking vigabatrin, which can cause retinal toxicity.

Generalizability to other populations

The heterogeneity of the patient sample favors the generalizability of this study's results.

Study funding/potential competing interests

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A draft of the short-form article was written by M. Dalefield, a writer with Editage, a division of Cactus Communications. The authors of the full-length article and the journal editors edited and approved the final version.

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