Gift of Sight Clinical Research Award
2019 Winner: Daniel Osborne

Title

Spectral-domain optical coherence tomography of children with Down syndrome

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Purpose

Down syndrome is known to be associated with numerous ophthalmic manifestations including corneal ectasia, iris stromal hyperplasia, cataracts and retinovascular anomalies. Foveal hypoplasia has been reported in 1-10% of children with Down Syndrome (Stephen *et al.* 2007, Kranj 2012).

Spectral-domain optical coherence tomography (SD-OCT) is the most accurate non-invasive in-vivo modality for assessing the structure of the retina, and to the best of our knowledge no study has previously reported SD-OCT findings in children with Down syndrome.

The primary outcome for this study was to assess the feasibility of obtaining optic nerve and macular SD-OCT data from this population. Our secondary outcome was to evaluate the retinal structure of patients with Down syndrome for any differences in comparison to previously established age-specific normative data.

Methods

We recruited 11 patients with an established diagnosis of Down syndrome. 9 patients underwent a full orthoptic and ophthalmological examination and handheld SD-OCT imaging of the optic nerve and macula. In order to quantify optic disc and macular morphology, SD-OCTs were segmented using a customised macro in ImageJ. We measured the thickness of the 10 retinal layers, 1700 microns from the disc in two meridia, at the fovea, 1000 microns from the fovea in two meridian, and 2000 microns from the fovea in two meridia. These measurements were compared with parameters previously reported in the literature. (Lee *et al.* 2015, Patel *et al.* 2016)

Results

The success rate for obtaining usable OCT images from this population was 82% (9 out of 11 subjects).

The prevalence of foveal hypoplasia was found to be 67%, which is significantly higher than the estimated prevalence in Down syndrome reported in the literature.

Adjacent to the optic disc, the retinal thickness was found to be higher than previously reported in children without Down syndrome, and this was attributable to an increased retinal nerve fibre layer thickness.

Conclusions

OCT examination of children with Down syndrome using a handheld OCT device is feasible, although there are challenges to examination in this population. Children with Down syndrome may have atypical retinal morphology, and in particular here may be a higher prevalence of foveal hypoplasia in this population than previously described.

References

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