Clinical and Experimental Vision and Eye Research (2021), 4, 52–52

CLINICAL IMAGE

Ocular findings in DiGeorge syndrome

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Received: 22-12-2021
Accepted: 07-01-2022
doi: 10.15713/ins.clever.79

A 17-year-old male with DiGeorge syndrome (chromosome 22q11.2 deletion) was referred to the ophthalmology clinic for an ocular examination during his regular checkup. The patient had no developmental delay or ocular complaints. His visual acuity was 20/30 in both eyes. The examination revealed symmetric findings of posterior embryotoxon [Figure 1] and star-shaped cataract [Figure 2]. History was negative for previous ocular trauma, radiation, electric shock, or treatment with chlorpromazine, possible etiologies in cataracts of this morphology. Cataract is a rare ocular finding amongst DiGeorge patients, and no particular shape was previously reported. Posterior embryotoxon is a common finding in these patients. It represents thickened and centrally displaced Schwalbe's ring (the periphery of the innermost surface of the cornea), which is seen as a sharply defined, concentric white line. This congenital defect may appear in different systemic conditions such as Alagille syndrome, Axenfeld-Rieger syndrome, or X-linked ichthyosis and may also occur in the general population.

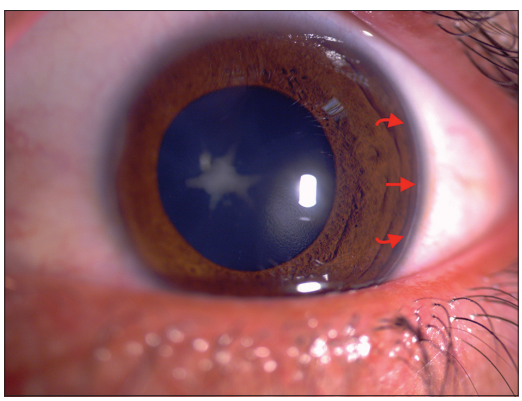

Figure 1: Posterior embryotoxon (red arrows)

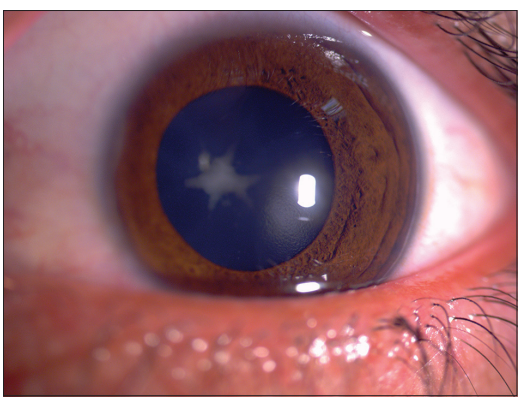

Figure 2: Star-shaped cataract

How to cite this article: Chorny A, Chiya B. Ocular findings in DiGeorge syndrome. Cli Exp Vis Eye Res J 2021;4(2):52.