Neuro-Ophthalmology

CONGENITAL ANOMALIES OF THE OPTIC DISC: AN OPTICAL COHERENCE TOMOGRAPHY PERSPECTIVE

Maria Filipa Cunha Madeira, Marta Correia, Margarida Baptista, Ana Cabugueira
Oftalmologia, Hospital De Egas Moniz (Centro Hospitalar De Lisboa Ocidental), Portugal

PURPOSE:
Congenital anomalies of the optic disc (OD) are diagnosed by fundoscopy with clinical overlap. Optical coherence tomography (OCT) allows non-invasive, high-resolution imaging of their distinguishing features.

METHODS:
Papillary areas of OD conformation and pseudopapilledema anomalies were examined in twenty patients with spectral-domain OCT.

RESULTS:
Horizontal OCT scans across the optic pit revealed a triangular cavity extending posteriorly from the lamina cribrosa at the temporal border. Adjacent scans detailed further interconnected cavities within the herniated dysplasic retina, floating vitreous fibers and a macular neurosensory retinal detachment.

Reconstruction of OD coloboma conveyed an eccentric shallow retinochoroidal-scleral excavation. A concave membrane spanning the OD and a reticulated, tractioned retina surface were seen in horizontal scans.

The posterior staphyloma had a steeper excavation (incompletely captured by OCT) encompassing the surrounding peripapillary area.

Morning glory disc is identified on OCT by its central glial tuft. The traction exerted by the preretinal membrane and vitreous pulls the retina away from the scleral excavation.

Horizontal OCT scan across OD drusen detail signal poor structures, bordered by hyperreflective margins above the lamina cribrosa, with overlapping hyperfluorescence.

Peripapillary hyperreflective ovoid mass-like structure (PHOMS) have a greater reflectivity and are externally located, as they represent bulging retinal nerve fibre layer (RNFL).

Myelinated fibers have a thickened hyperreflective appearance on OCT scan continuous with RNFL, shadowing inferior layers, but totally sparing the OD.

CONCLUSIONS:
Delving into the anatomy of OD congenital anomalies with OCT enhances their differential diagnosis, guiding further ocular and systemic investigations.

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