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PHENOTYPIC SIMILARITY OF PFV AND CHRRPE: DISTINCT ENTITIES OR A POTENTIAL CONNECTION?

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Introduction:

Persistent fetal vasculature (PFV) is a congenital anomaly caused by failure of regression of the primary vitreous and hyaloid vasculature. PFV is associated with a surprisingly wide spectrum of presentations. While one of the most common presentations is tent-shaped tractional retinal detachment (TRD) due to a hyaloid artery remnant emanating from the optic nerve, there are rare instances of a hyperpigmented elevated mass-like lesion observed in these eyes. This previously unreported finding in PFV eyes resembles the appearance of a combined hamartoma of the retina and retinal pigment epithelium (CHRRPE). CHRRPE is a variably pigmented benign tumor of the retina and retinal pigment epithelium, characterized by overlying glial cell proliferation, which frequently results in epiretinal membrane formation and distortion of the retinal architecture and vessels. Interestingly, in some cases of CHRRPE, preretinal gliosis overlying the peripapillary tumor is more prominent and exhibits a hyaloidal stalk-like appearance that extends into the vitreous cavity. In this study, we aim to investigate cases exhibiting overlapping features of persistent fetal vasculature(PFV) and combined hamartoma of the retina and retinal pigment epithelium(CHRRPE) and to explore potential associations between these developmental ocular anomalies.

Materials and methods:

Descriptive case-series including nine eyes of eight patients aged 0-7 years with shared clinical features of PFV and CHRRPE. Diagnoses were established through clinical examination and intraoperative findings.

Results:

All eyes exhibited common features, including elevated pigmented retinal thickening, increased vascular tortuosity, and preretinal fibrotic/gliotic changes or epiretinal membranes. Macular involvement was observed in 56% of cases, and peripapillary involvement in 44%. Four eyes showed hyaloid stalk-like fibrotic remnants extending from the lesion to the posterior lens surface, suggestive of PFV component, while the other five harbored isolated CHRRPE. Notably, one patient presented with PFV in one eye, and CHRPPE in the other.

Conclusions:

There is a clinical overlap between PFV and CHRRPE, with some cases displaying features typically associated with the other condition. The presence of both diagnoses in the same patient further suggests a potential association between these entities, possibly representing two ends of the same spectrum rather than distinct entities. Further research, including molecular studies, is needed to explore this potential connection and deepen our understanding of ocular development.

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