Macular Microvascular Findings in Familial Exudative Vitreoretinopathy on Optical Coherence Tomography Angiography

S. Tammy Hsu1, Avni P. Finn1, Xi Chen1, Robert J. House1, Michael P. Kelly1, Cynthia A. Toth1,2, Lejla Vajzovic1

1 Department of Ophthalmology, Duke University School of Medicine, Durham, North Carolina, USA; 2 Biomedical Engineering Department, International University, Department of Biomedical Engineering, Duke University, Durham, North Carolina, USA

INTRODUCTION

- Familial exudative vitreoretinopathy (FEVR): Rare hereditary disorder1 Associated with genetic mutations in the Wnt signaling pathway necessary for retinal angiogenesis2,3 Characterized by incomplete and anomalous retinal vascular development Abnormalities have only been reported to be in the periphery4 Optical coherence tomography angiography (OCTA) can be used to noninvasively image retinal microvasculature without use of contrast dye OCTA provides depth-resolved images of the superficial and deep vascular complexes (SVC and DVC, respectively) Purpose: To determine if patients with familial exudative vitreoretinopathy (FEVR) have abnormalities in the macular microvasculature

METHODS

- Study approved by IRB, adhered to HIPAA & Declaration of Helsinki; obtained parental consent for each subject
- Prospective, observational case series
- Imaged 22 eyes of 6 FEVR and 6 control patients with OCTA
- FEVR: mean age 17.5 ± 7.5 yrs., median 20 yrs., range 2-25 yrs.
- Control: mean age 18.3 ± 15.8 yrs., median 15 yrs., range 12.5-64 yrs.
- OCTA images were obtained using the investigational Spectralis table top and portable Flex units (Figure 1) in the clinic and operating room, respectively
- Masked graders analyzed the OCTA images (SVC and DVC) for abnormal vascular features (Table 1)
- Compared OCTA to fluorescein angiography (FA) findings

RESULTS

- OCTA of 7 of 11 eyes with FEVR displayed abnormal macular findings in the microvasculature (Table 2, Figure 2):
  - SVC: dilation, disorganization, straightening, heterogeneous vessel density, and curls
  - DVC: areas of decreased density, disorganization, curls, and end-bulbs

- Vascular curls and loops
- Decreased density, disorganization, end-bulbs
- Abnormally straightened vessels

DISCUSSION

- End-bulbs could correlate with vessel termination, with suspended red blood cells in motion captured on OCT-A within the stubs5
- Association with animal models with defective Wnt signaling
- DVC vascular end-bulbs visible on OCT-A in humans (including one with documented LRPS mutation) parallel mouse models of FEVR with mutations in the Wnt signaling pathway6
- Mice with defective Nomin/Fzd4 signaling or LRPS signaling demonstrate retinal hypovascularization with delayed radial migration of endothelial cells and defective arborization of deeper capillaries following the vertical endothelial innervation from the vitreal surface7
- Limitations: Small sample size Cross-sectional study cannot assess longitudinal development Heterogeneity of FEVR disease stages, treatment history, and clinical findings
- Next steps: Increase sample size of patients with FEVR Correlate with histology of human retinas with FEVR

CONCLUSION

- OCTA revealed distinctive depth-resolved macular vasculature abnormalities in patients with FEVR.
- FEVR may not be a disease of peripheral retinal but also of the macula and deep retinal vasculature.
- Unique vascular patterns may assist in diagnosis and prognosis

REFERENCES


ACKNOWLEDGMENTS

Correspondence: s.tammyhsu@duke.edu