Optical Coherence Tomography Angiography (OCTA) Analysis of Retinal Microvascular Abnormalities in Genetically-confirmed Familial Exudative Vitreoretinopathy (FEVR)

Bascom Palmer Eye Institute
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INTRODUCTION

- FEVR is a genetic condition that affects retinal angiogenesis.
- The FEVR phenotype can vary greatly between individuals.
- While multimodal imaging (with FA and OCT) has advanced the understanding of FEVR, these modalities do not allow for depth-resolved assessment of the microvasculature, which could be an important indicator of disease severity and progression.

OBJECTIVE

To describe retinal microvasculature abnormalities in patients with genetically-confirmed FEVR using OCTA and assess for clinical and phenotypic differences among different genotypes.

METHODS

This study was a retrospective analysis of 24 eyes with genetically-confirmed FEVR who underwent prospective imaging using supine OCTA and FA during examination under anesthesia at Bascom Palmer Eye Institute from March 1, 2019 to May 31, 2021. The images were qualitatively analyzed to determine the presence or absence of an abnormal FAZ, decreased vessel density, vessel straightening, vessel anastomoses, presence of terminal bulbs, and presence of capillary loops.

RESULTS

DEMOGRAPHIC INFORMATION

24 eyes
12 patients

Mean Age
6.1±4.85 y

67% M
33% F

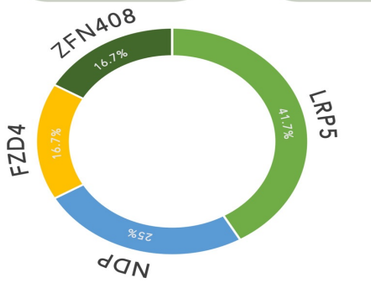


Fig 1. Percentage of patients with FEVR-associated genetic mutations.

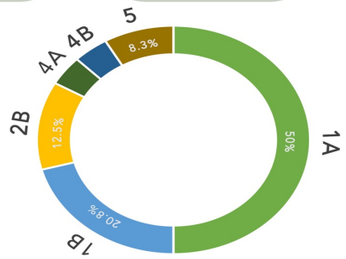
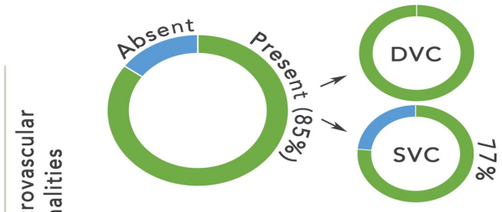


Fig 2. Distribution of FEVR stages.



Macular Microvascular Abnormalities

#1: decreased vessel density (60%)

#1: Blood vessel straightening (50%)

Patient with *NDP* Mutations
- ↑SVC abnormalities*
- ↑Absent FAZ*
- ↑Terminal bulbs (DVC)*
- ↑BV straightening*
- ↑Vessel dragging*

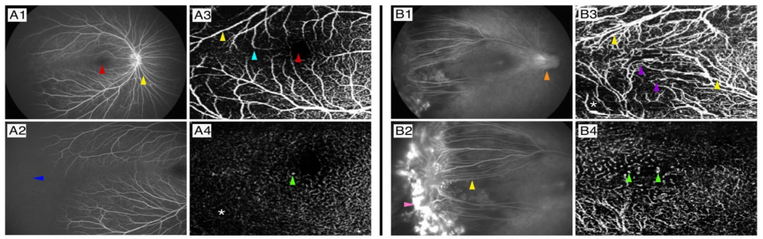


Fig 3. Representative OCTA and FA studies in two patients with FEVR and an NDP mutation.

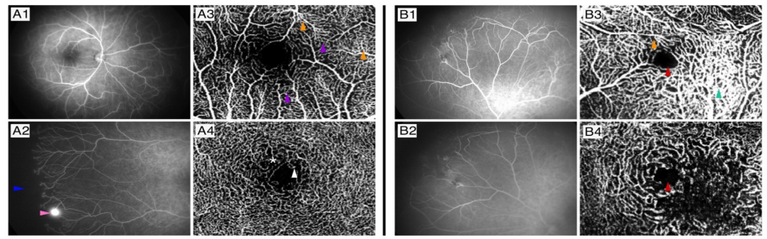


Fig 4. Representative OCTA and FA studies in two patients with FEVR and an LRP5 mutation.

CONCLUSIONS

FEVR is a retinal vascular condition with highly heterogeneous phenotypes and genotypes. OCTA allows for the identification of depth-resolved microvascular abnormalities beyond those seen with FA. The results presented in this study suggest that different genotypes may exhibit different patterns of microvascular abnormalities.

references