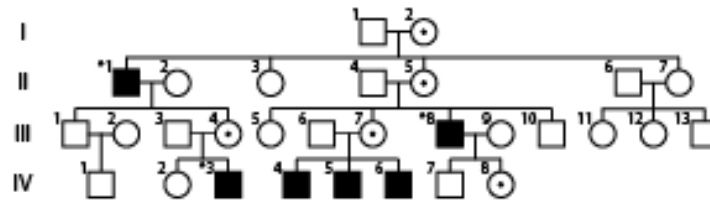


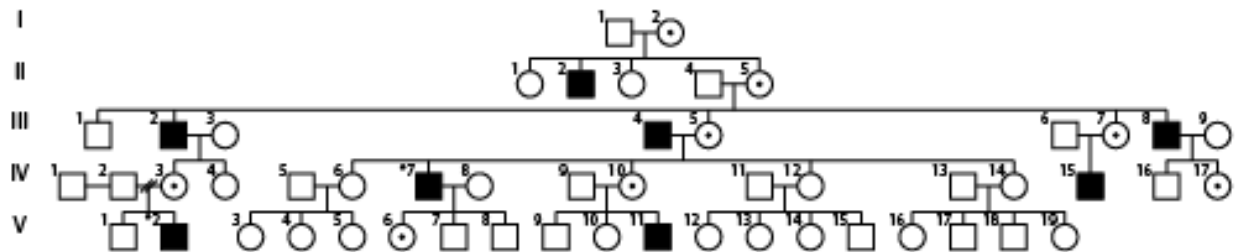
Supplemental Figure 1

Pedigrees for Families 6, 11, and 15, as indicated in Table 1. Asterisks denote patients included in this study.

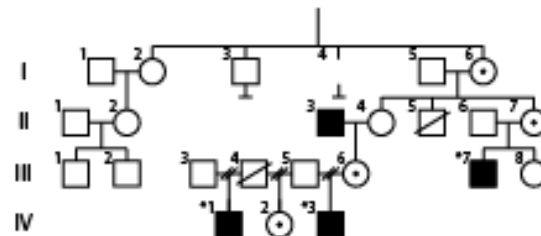
Family 6



Family 11

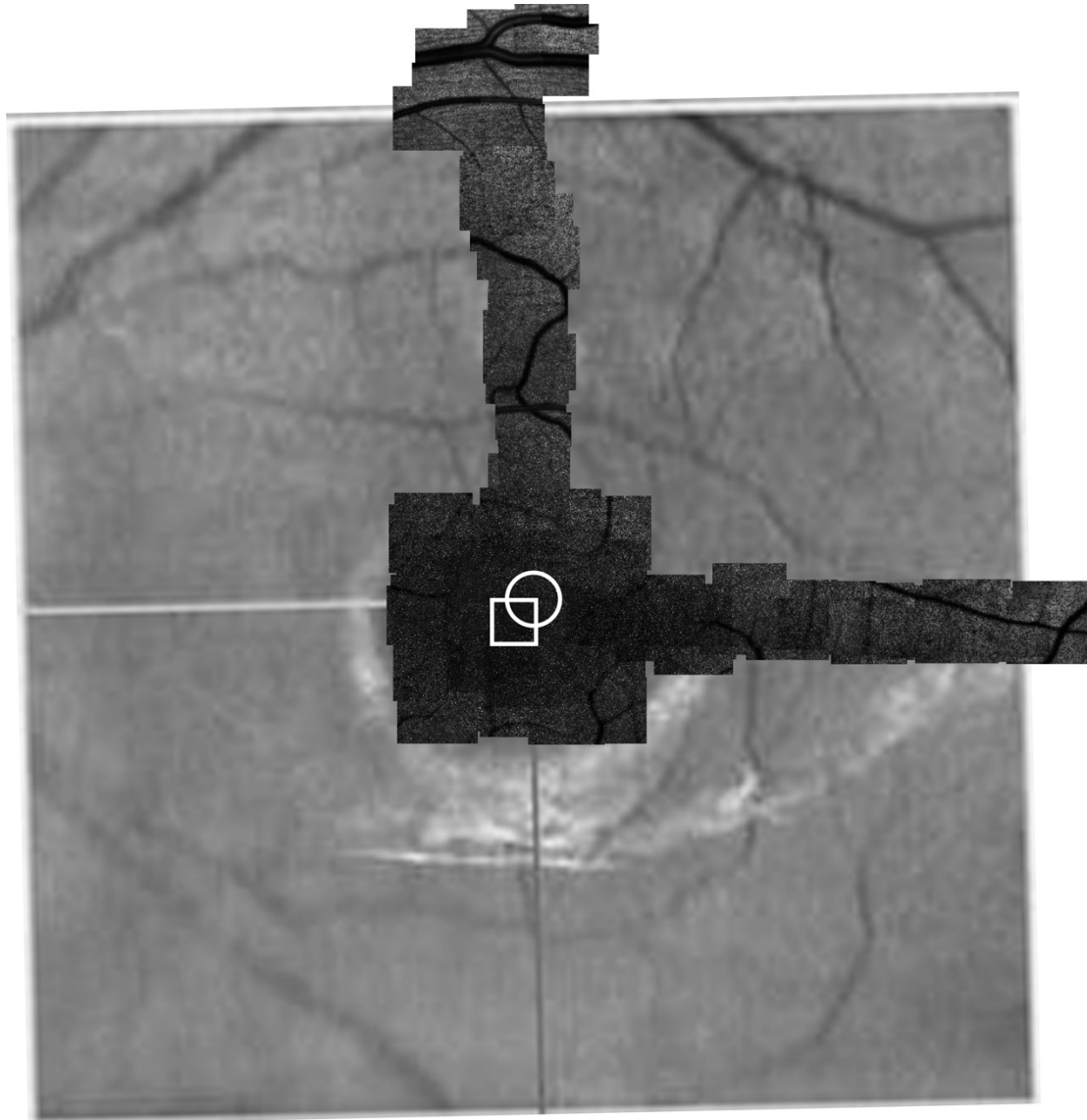


Family 15



Supplemental Figure 2

Alignment of the confocal adaptive optics scanning light ophthalmoscopy montage and the fundus image (Cirrus HD-OCT, Carl Zeiss, Meditec) for JC_10557; the patient with the largest displacement (186 μm) between the locations of peak cell density (white square) and anatomical fovea (white circle).



Supplemental Figure 3

Confocal (left) and corresponding split-detection images (right), centered on the location of peak cell density, of all ten patients with blue cone monochromacy for whom both modalities were analyzable. The anatomical fovea is marked with a star for all patients except JC_10558, for whom alignment between modalities was not possible.

