Supplementary Online Content

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**eFigure 1.** Blue light reflectance images and macular optical coherence tomography of two MacTel patients.
**eFigure 2.** J28/F289 Family
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This supplementary material has been provided by the authors to give readers additional information about their work.
**eFigure 1.** Blue light reflectance images and macular optical coherence tomography of two MacTel patients.

Blue light reflectance images and macular optical coherence tomography of the 52-year-old father (a, b) and the 21-year-old MacTel proband (c, d) from the family shown in **Figure 2b.** Blue light reflectance shows an increased ring-like parafoveal reflectance both in the father (a) and his son (c). Macular OCT shows ellipsoid zone disruption in the proband (d) with more prominent juxtafoveal cavitations in the proband’s father (b).
**eFigure 2. J28/F289 Family**

Probands are marked with a black arrow. A red diamond denotes individuals enrolled in the study; filled symbols denote confirmed MacTel diagnoses.
**eFigure 3. UT001/F287 Family**

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