Supplementary Tables

Table S1. Clinical data on 59 individuals with a *CACNA1F* mutation.

Individual #59 was excluded as this male had a *CABP4* mutation. BCVA = best corrected visual acuity, LogMar = Logarithm of Minimal angle of resolution, eq. sph.= equivalent sphere, D = diopters, P = light perception, -*High myopia prior to cataract operation, †Atypical course (severe retinal dystrophy), ‡ Progressive cone dystrophy, +/-= present/absent, nd = no data.

Table S2. Foveal Morphology in AED.

Cirrus and Spectralis grading of hypoplasia was masked. Agreement between modes for this subjective assessment was 100%. Also, in all cases where both eyes could be graded, there was symmetry. Y=yes, N=no, ND=no data, NA=image not analyzable

Table S3. Foveal Outer Segment Length in AED.

Mean ± standard deviation outer segment length for CSNB2A was 41.65 ± 4.99µm, OD (n=48) and 42.07 ± 5.65µm, OS (n=48). For comparison, normal outer segment length is 46.04 ± 4.34µm. Normal data derived from Wilk et al.[6] n=23 subjects; age (mean ± standard deviation) = 30 ±16 years; range 8-67 years. ND=no data, NA=image not analyzable. Subject #59 was not included in the calculation of the mean values.

Table S4. Subfoveal choroidal thickness in AED

Mean ± standard deviation choroidal thickness for AED was 195.74 ± 77.36µm, OD (n=40) and 187.85 ± 87.27µm, OS (n=37). The values appear significantly below previously published normative data. ND=no data, NA=image not analyzable. Subject #59 was not included in the calculation of the mean values.

Table S5. *In silico* prediction of the pathogenicity of identified missense variants

[a] SIFT (sift.jcvi.org/), the numbers in brackets are (score; median). [b] Polyphen2 (http://genetics.bwh.harvard.edu/pph2/), the number in brackets is the score using the HumVar model. AlignGVGD (http://agvgd.iarc.fr/), MutationTaster (http://www.mutationtaster.org/). [c] LuCAMP data are from exome sequencing of 2000 persons residing in Denmark (ref). [d] dbSNP is from version 142. [e] ESP is the Exome variant server (http://evs.gs.washington.edu/EVS/). [f] The class is based on an in house classification system based on mutation type, segregation data, population frequencies and functional studies.