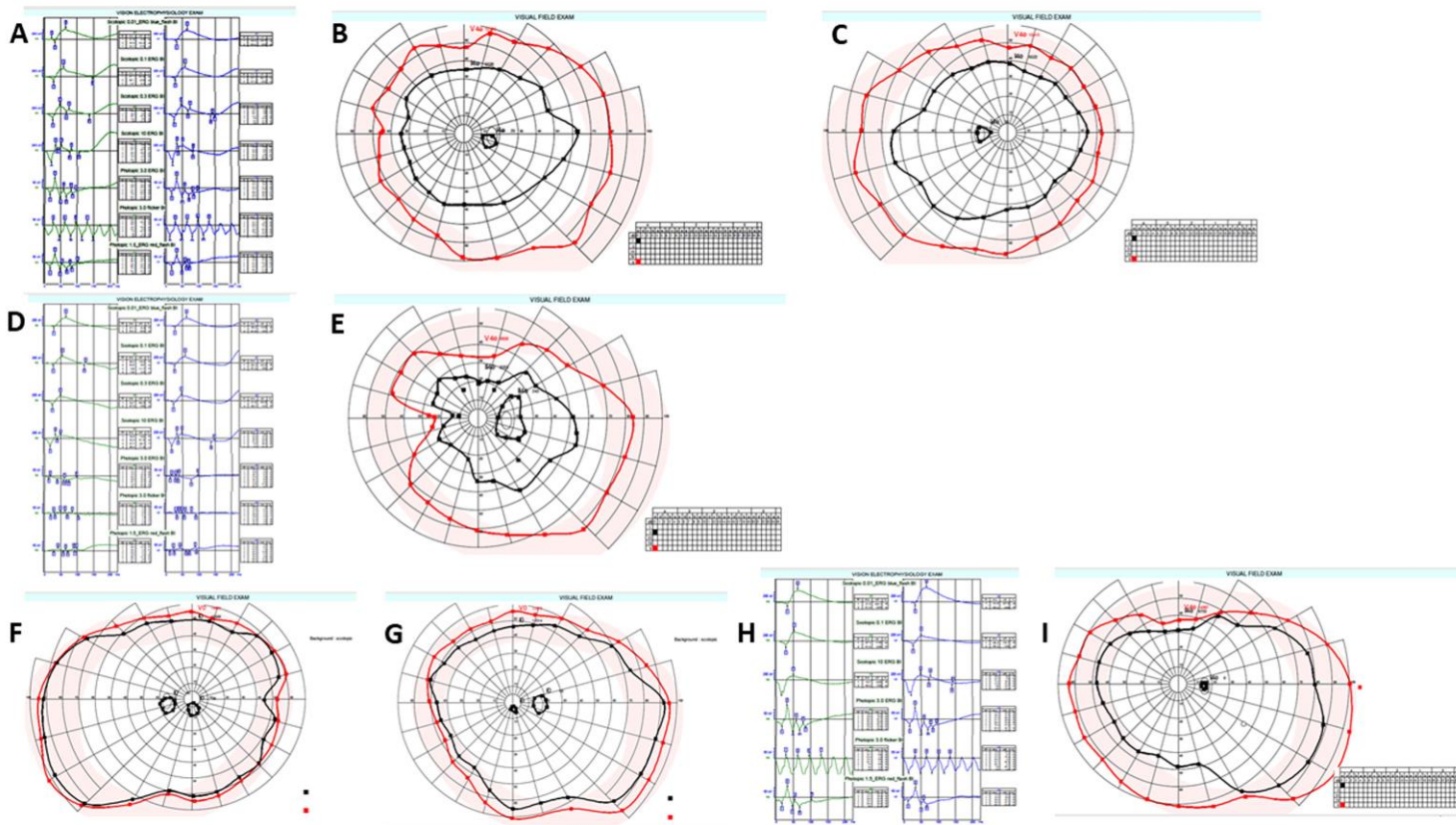


Supplementary Fig. 1

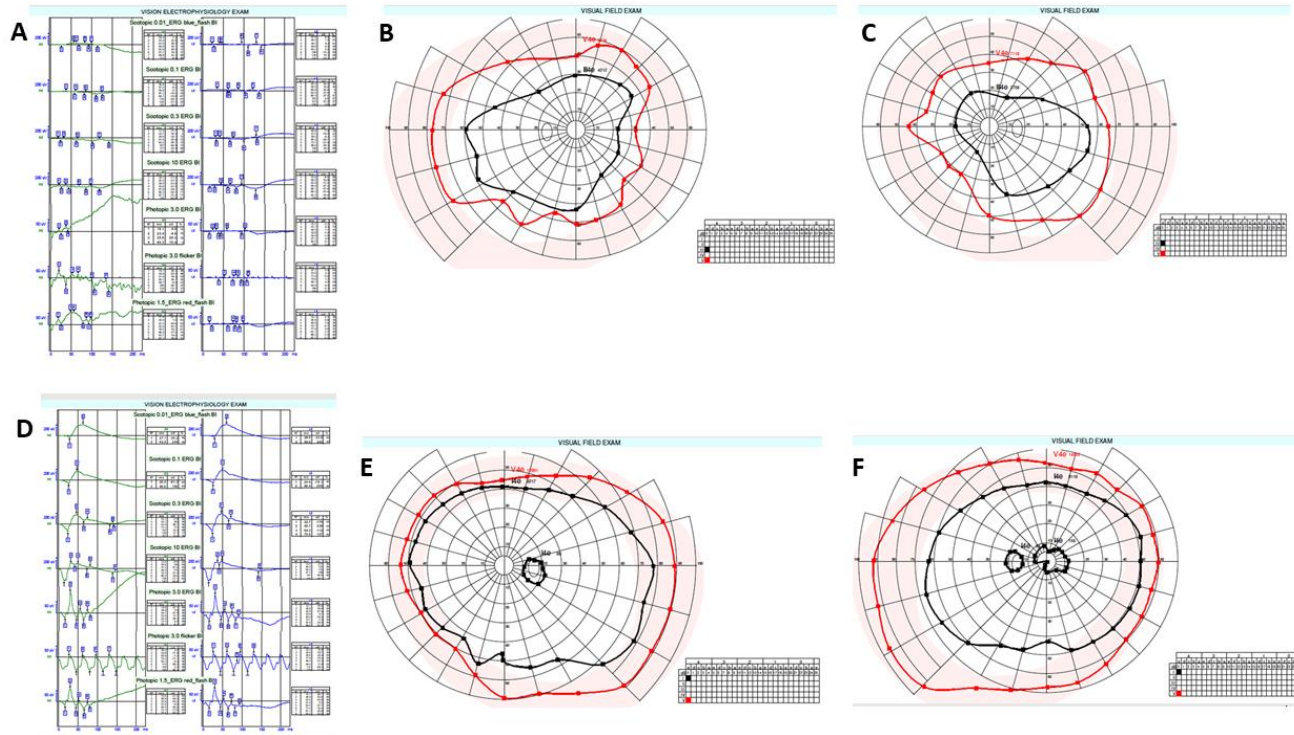
Representative ERG traces and visual field tests for patients #1(A-C), #2 (D,E), #3 (F,G), and #5 (H,I) (PNG 3457 kb)



Supplementary Fig. 2

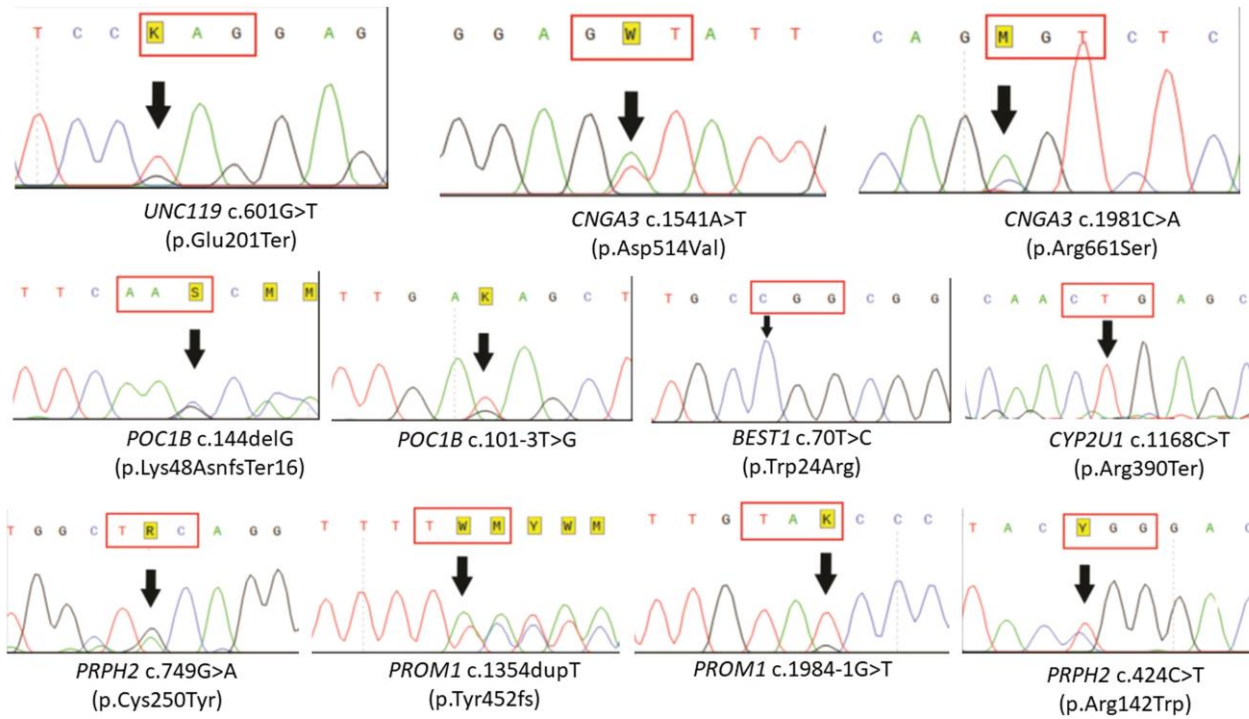
Representative ERG traces and visual field tests for patients

#7 (A-C) and #8 (D-F) (PNG 3150 kb)



Supplementary Fig. 3

Sanger sequencing confirmation of pathogenic and likely pathogenic variants.
(PNG 1689 kb)



Supplementary file.- References for published *POC1B* disease-causing variants.

- Birtel J, Eisenberger T, Gliem M, Müller PL, Herrmann P, Betz C, Zahnleiter D, Neuhaus C, Lenzner S, Holz FG, Mangold E, Bolz HJ, Charbel Issa P. Clinical and genetic characteristics of 251 consecutive patients with macular and cone/cone-rod dystrophy. *Sci Rep*. 2018; 8:4824.
- Durlu YK, Köroğlu Ç, Tolun A. Novel recessive cone-rod dystrophy caused by *POC1B* mutation. *JAMA Ophthalmol*. 2014; 132:1185-91.
- Jin X, Chen L, Wang D, Zhang Y, Chen Z, Huang H. Novel compound heterozygous mutation in the *POC1B* gene underlie peripheral cone dystrophy in a Chinese family. *Ophthalmic Genet*. 2018; 39:300-6.
- Kameya S, Fujinami K, Ueno S, Hayashi T, Kuniyoshi K, Ideta R, Kikuchi S, Kubota D, Yoshitake K, Katagiri S, Sakuramoto H, Kominami T, Terasaki H, Yang L, Fujinami-Yokokawa Y, Liu X, Arno G, Pontikos N, Miyake Y, Iwata T, Tsunoda K; Japan Eye Genetics Consortium. Phenotypical characteristics of *POC1B*-associated retinopathy in Japanese cohort: Cone dystrophy with normal fundusoscopic appearance. *Invest Ophthalmol Vis Sci*. 2019; 60:3432-46.
- Kominami A, Ueno S, Kominami T, Nakanishi A, Ito Y, Fujinami K, Tsunoda K, Hayashi T, Kikuchi S, Kameya S, Iwata T, Terasaki H. Case of cone dystrophy with normal fundus appearance associated with biallelic *POC1B* variants. *Ophthalmic Genet*. 2018; 39:255-62.
- Peturson AC, Noel NCL, MacDonald IM. A homozygous *POC1B* variant causes recessive cone-rod dystrophy. *Ophthalmic Genet*. 2021; 42:349-53.
- Roosing S, Lamers IJ, de Vrieze E, van den Born LI, Lambertus S, Arts HH; *POC1B* Study Group, Peters TA, Hoyng CB, Kremer H, Heterschijt L, Letteboer SJ, van Wijk E, Roepman R, den Hollander AI, Cremers FP. Disruption of the basal body protein *POC1B* results in autosomal-recessive cone-rod dystrophy. *Am J Hum Genet*. 2014; 95:131-42.
- Toulis V, Cortés-González V, Castro-Miró M, Sallum JF, Català-Mora J, Villanueva-Mendoza C, Ciccioli M, González-Duarte R, Valero R, Marfany G. Increasing the Genetic Diagnosis Yield in Inherited Retinal Dystrophies: Assigning Pathogenicity to Novel Non-canonical Splice Site Variants. *Genes (Basel)*. 2020; 11:378.

Weisschuh N, Mazzola P, Bertrand M, Haack TB, Wissinger B, Kohl S, Stingl K. Clinical Characteristics of *POC1B*-Associated Retinopathy and Assignment of Pathogenicity to Novel Deep Intronic and Non-Canonical Splice Site Variants. *Int J Mol Sci.* 2021; 22:5396.

LOVD and ClinVar variant identifiers.

Gene	Variant	LOVD Variant ID	ClinVar Submission
<i>UNC119</i>	c.601G>T, p.Glu201Ter	0000846838	SUB11357785
<i>CNGA3</i>	c.1541A>T, p.Asp514Val c.1981C>A, p.Arg661Ser	0000846839 0000846841	SUB11360215
<i>POC1B</i>	c.144delG, p.Lys48AsnfsTer16 c.101-3T>G	0000846843 0000846844	SUB11360319
<i>BEST1</i>	c.70T>C, p.Trp24Arg	0000846845	SUB11360600
<i>CYP2U1</i>	c.1168C>T, p.Arg390Ter	0000846846	SUB11360770
<i>PRPH2</i>	c.749G>A, p.Cys250Tyr	0000846847	SUB11361105
<i>PROM1</i>	c.1354dupT, p.Tyr452fs c.1984-1G>T	0000846852 0000846853	SUB11361273
<i>PRPH2</i>	c.424C>T, p.Arg142Trp	0000846854	SUB11361311