

## Supplemental Online Content

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**eTable.** Gene variants detected in blood samples

**eFigure 1.** In vivo gross pathology highlights bony spicules characteristic of RP

**eFigure 2.** Further in vivo imaging of the patient supports asymmetric findings

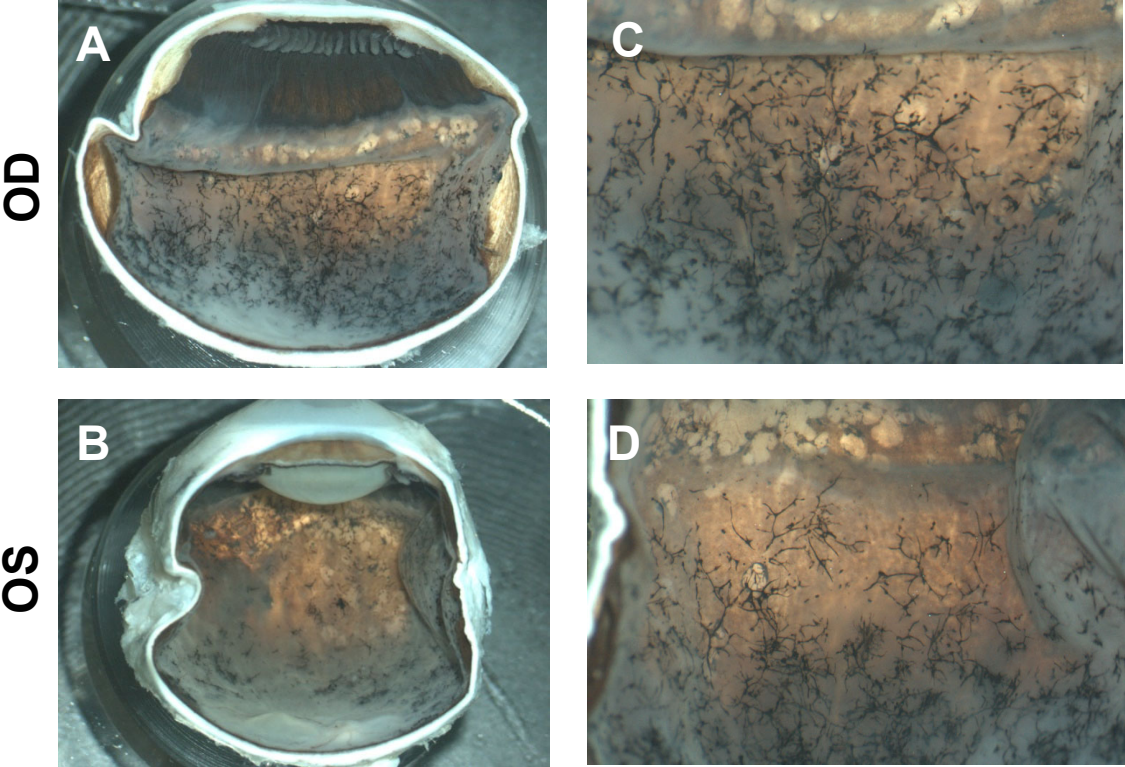
This supplementary material has been provided by the authors to give readers additional information about their work.

**eTable.** Gene variants detected in blood samples

Variant ID (GRCh37)	Gene	Transcript	Mode of Inheritance	Variant (cDNA, protein)
4-15539735-G-C	<i>CC2D2A</i>	NM_001080522.2	AR	c.1978G>C, p.Val660Leu
10-73574808-G-A	<i>CDH23</i>	NM_022124.5	AD, AR	c.9838G>A, p.Asp3280Asn
17-42429396-C-T	<i>GRN</i>	NM_002087.3	AD, AR	c.1193C>T, p.Ser398Leu
20-21112834-A-G	<i>KIZ</i>	NM_018474.6	AR	c.152+34A>G
18-7037641-G-A	<i>LAMA1</i>	NM_005559.3	AR	c.1673C>T, p.Ala558Val
4-110791654-CCTT-C	<i>LRIT3</i>	NM_198506.4	AR	c.1752_1754del, p.Leu585del
2-170094756-C-A	<i>LRP2</i>	NM_004545.2	AR	c.4351G>T, p.Val1451Phe
11-68191020-A-G	<i>LRP5</i>	NM_002335.3	AD, AR	c.2091A>G, p.Ile1031Val
2-234235804-C-A	<i>SAG</i>	NM_000541.4	AR	c.473C>A, p.Thr158Lys

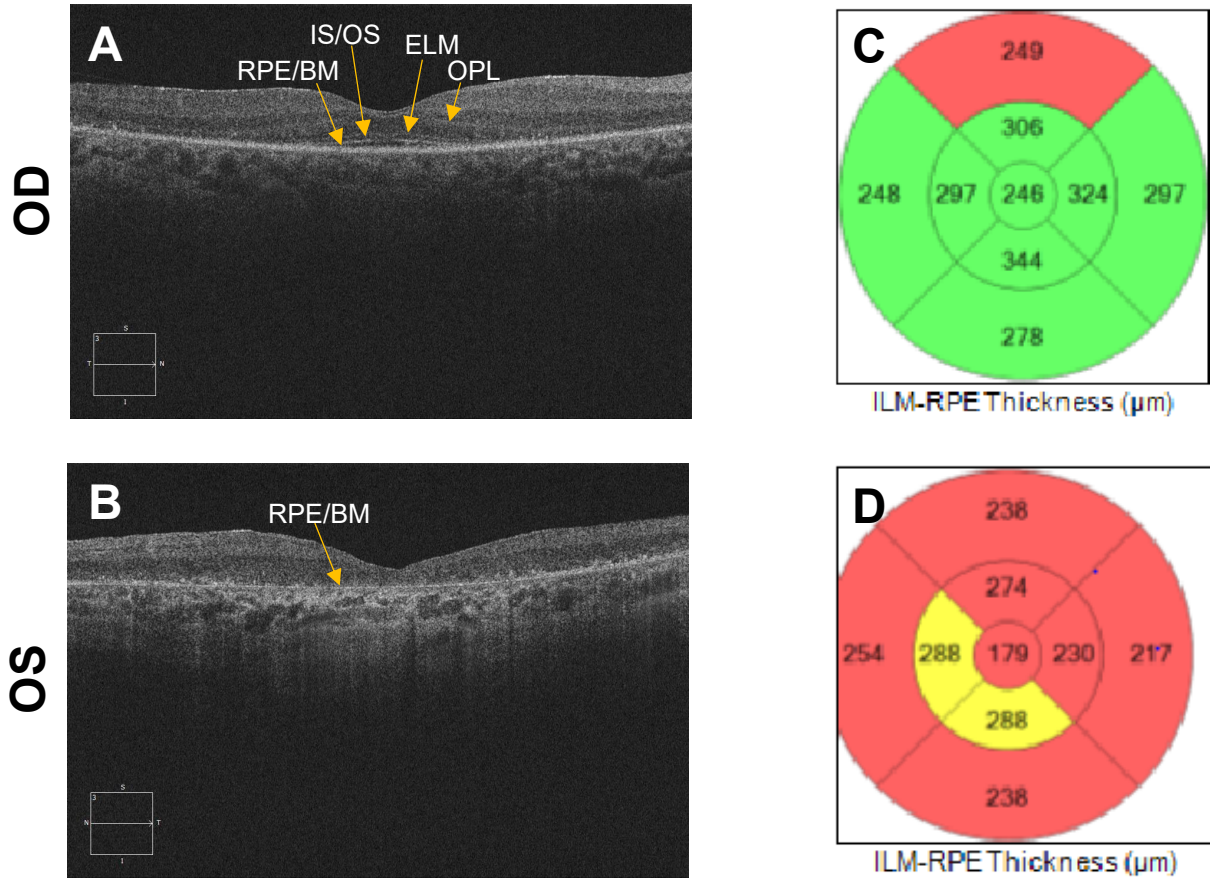
These are established heterozygous variants related to a variety of genetic conditions, including RP.

**eFigure 1.** In vivo gross pathology highlights bony spicules characteristic of RP



Gross pathology (D, H) illustrates the bony spicules present in OD (A, B) and OS (C, D). OS is more atrophic, so much so that there are fewer bony spicules remaining.

**eFigure 2.** Further in vivo imaging of the patient supports asymmetric findings



SD-OCT imaging of OD (A) and OS (B) demonstrates more widespread foveal atrophic changes (ONL and ELM) in the OS (B). Meanwhile, the IS/OS junction seems relatively intact within the fovea in OD compared to OS, in which the only remaining distinguishable feature is the RPE/BM complex. Retinal thickness maps of OD (C) and OS (D), measuring the thickness of the retina from the internal limiting membrane to the RPE in  $\mu\text{m}$ , further demonstrate thinning of the macula OS $\gg$ OD. ONL = outer nuclear layer. ELM = external limiting membrane. IS = inner segment. OS = outer segment. BM = Bruch's membrane.