

Supplementary Information

NGS-based Molecular diagnosis of 105 eyeGENE[®] probands with Retinitis

Pigmentosa

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Supplemental Table 1 Genes in our retinal capture panel

All genes in the panel	RP genes in the panel	Inheritance mode of RP genes
<i>ABCA4</i>	<i>ABCA4</i>	recessive
<i>ABCC6</i>	<i>BBS1</i>	recessive
<i>ABHD12</i>	<i>BEST1</i>	recessive/dominant
<i>ADAM9</i>	<i>C2orf71</i>	recessive
<i>TREX1</i>	<i>C8orf37</i>	recessive
<i>AHI1</i>	<i>CA4</i>	dominant
<i>AIPL1</i>	<i>CC2D2A</i>	recessive
<i>ALMS1</i>	<i>CERKL</i>	recessive
<i>ARL6</i>	<i>CLRN1</i>	recessive
<i>ARMS2</i>	<i>CNGA1</i>	recessive
<i>ATXN7</i>	<i>CNGB1</i>	recessive
<i>BBS1</i>	<i>CRB1</i>	recessive
<i>BBS10</i>	<i>CRX</i>	dominant
<i>BBS12</i>	<i>DHDDS</i>	recessive
<i>BBS2</i>	<i>EYS</i>	recessive
<i>BBS4</i>	<i>FAM161A</i>	recessive
<i>BBS5</i>	<i>FLVCR1</i>	recessive
<i>BBS7</i>	<i>FSCN2</i>	dominant
<i>BBS9</i>	<i>GNPTG</i>	recessive
<i>BEST1</i>	<i>GUCA1B</i>	dominant
<i>C1QTNF5</i>	<i>IDH3B</i>	recessive
<i>C2</i>	<i>IMPDH1</i>	dominant
<i>C2orf71</i>	<i>IMPG2</i>	recessive
<i>C3</i>	<i>KLHL7</i>	dominant
<i>C8orf37</i>	<i>LRAT</i>	recessive
<i>CA4</i>	<i>MAK</i>	recessive
<i>CABP4</i>	<i>MERTK</i>	recessive
<i>CACNA1F</i>	<i>NR2E3</i>	recessive/dominant
<i>CACNA2D4</i>	<i>NRL</i>	recessive/dominant
<i>CC2D2A</i>	<i>OFD1</i>	X-linked
<i>CDH23</i>	<i>PDE6A</i>	recessive
<i>CDH3</i>	<i>PDE6B</i>	recessive
<i>CDHR1</i>	<i>PDE6G</i>	recessive
<i>CEP164</i>	<i>PRCD</i>	recessive
<i>CEP290</i>	<i>PROM1</i>	recessive
<i>CERKL</i>	<i>PRPF3</i>	dominant

<i>CFB</i>	<i>PRPF31</i>	dominant
<i>CFH</i>	<i>PRPF6</i>	dominant
<i>CHM</i>	<i>PRPF8</i>	dominant
<i>CLN3</i>	<i>PRPH2</i>	dominant
<i>CLRN1</i>	<i>RBP3</i>	recessive
<i>CNGA1</i>	<i>RDH12</i>	dominant
<i>CNGA3</i>	<i>RGR</i>	recessive
<i>CNGB1</i>	<i>RHO</i>	recessive/dominant
<i>CNGB3</i>	<i>RLBP1</i>	recessive
<i>CNNM4</i>	<i>ROM1</i>	dominant
<i>COL11A1</i>	<i>RP1</i>	recessive/dominant
<i>COL2A1</i>	<i>RP2</i>	X-linked
<i>COL9A1</i>	<i>RP8</i>	dominant
<i>CRB1</i>	<i>RP9</i>	dominant
<i>CRX</i>	<i>RPE65</i>	recessive/dominant
<i>CYP4V2</i>	<i>RPGR</i>	X-linked
<i>DFNB31</i>	<i>SAG</i>	recessive
<i>DHDDS</i>	<i>SEMA4A</i>	dominant
<i>DMD</i>	<i>SNRNP200</i>	dominant
<i>EFEMP1</i>	<i>SPATA7</i>	recessive
<i>ELOVL4</i>	<i>TOPORS</i>	dominant
<i>ERCC6</i>	<i>TTC8</i>	recessive
<i>EYS</i>	<i>TTPA</i>	recessive
<i>FAM161A</i>	<i>TULP1</i>	recessive
<i>FBLN5</i>	<i>USH2A</i>	recessive
<i>FLVCR1</i>		
<i>FSCN2</i>		
<i>FZD4</i>		
<i>GNAT1</i>		
<i>GNAT2</i>		
<i>GNPTG</i>		
<i>GPR179</i>		
<i>GPR98</i>		
<i>GRK1</i>		
<i>GRM6</i>		
<i>GUCA1A</i>		
<i>GUCA1B</i>		
<i>GUCY2D</i>		
<i>HMCN1</i>		

HTRA1		
IDH3B		
IFT140		
IMPDH1		
IMPG2		
INPP5E		
INVS		
IQCB1		
JAG1		
KCNJ13		
KCNV2		
KIF11		
KLHL7		
LCA5		
LRAT		
LRP5		
MAK		
MERTK		
MFN2		
MFRP		
MKKS		
MKS1		
MTTP		
MYO7A		
NDP		
NMNAT1		
NPHP1		
NPHP3		
NPHP4		
NR2E3		
NRL		
NYX		
OAT		
OFD1		
OPA1		
OPA3		
OPN1LW		
OPN1MW		
OPN1SW		

<i>OTX2</i>		
<i>PANK2</i>		
<i>PAX2</i>		
<i>PCDH15</i>		
<i>PDE6A</i>		
<i>PDE6B</i>		
<i>PDE6C</i>		
<i>PDE6G</i>		
<i>PDZD7</i>		
<i>PEX1</i>		
<i>PEX2</i>		
<i>PEX7</i>		
<i>PGK1</i>		
<i>PHYH</i>		
<i>PITPNM3</i>		
<i>PLA2G5</i>		
<i>PRCD</i>		
<i>PRD</i>		
<i>PROM1</i>		
<i>PRPF3</i>		
<i>PRPF31</i>		
<i>PRPF6</i>		
<i>PRPF8</i>		
<i>PRPH2</i>		
<i>PXMP3</i>		
<i>RAX2</i>		
<i>RB1</i>		
<i>RBP3</i>		
<i>RBP4</i>		
<i>RCD1</i>		
<i>RD3</i>		
<i>RDH12</i>		
<i>RDH5</i>		
<i>RGR</i>		
<i>RGS9</i>		
<i>RGS9BP</i>		
<i>RHO</i>		
<i>RIMS1</i>		
<i>RLBP1</i>		

ROM1		
RP1		
RP1L1		
RP2		
RP8		
RP9		
RPE65		
RPGR		
RPGRIP1		
RPGRIP1L		
RS1		
SAG		
SDCCAG8		
SEMA4A		
SLC24A1		
SNRNP200		
SPATA7		
TEAD1		
TIMM8A		
TIMP3		
TLR3		
TLR4		
TMEM126A		
TMEM237		
TOPORS		
TREX1		
TRIM32		
TRPM1		
TSPAN12		
TTC8		
TTPA		
TULP1		
UNC119		
USH1C		
USH1G		
USH2A		
VCAN		
WDPCP		
WDR19		

<i>WFS1</i>		
<i>WDPCP</i>		
<i>WDR19</i>		

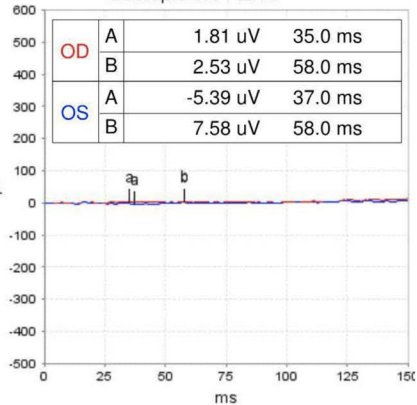
Supplemental Table 2 Genes screened for 16 probands before NGS-based sequencing

Sample	Genes Screened
RC+V.27	<i>RP2, RPGR</i>
3H5+K.42	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, RP2</i>
3HV+M.66	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, RP2</i>
3WP+3.68	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, TOPORS</i>
3JY+V.17	<i>RP2, RPGR</i>
3JE+8.28	<i>RP2, RPGR</i>
5H3+9.73	<i>RP2, RPGR</i>
UY2+R.10	<i>CDH23, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, USH1C, USH1G, USH2A</i>
5EF+L.58	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, TOPORS</i>
5WG+V.72	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, RP2</i>
LA+X.61	<i>RDS</i>
3W5+K.90	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, RP2</i>
375+8.59	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, RP2</i>
3VU+9.42	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, TOPORS</i>
3VF+9.12	<i>IMPDH1, KLHL7, NR2E3, PRPF3, PRPF31, PRPF8, RDS, RHO, RP1, TOPORS</i>
UPF+S.24	<i>ABCA4</i>

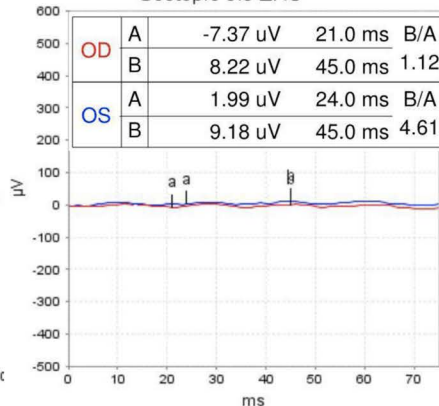
Figure Legends

Supplemental Figure S1. Electroretinogram results for patient UFC+7.74. Records shown here include Scotopic 0.01 (A), Scotopic 3.0 (B), Scotopic 3.0 Oscillatory Potentials (C), Photopic 3.0 (D), and Photopic Flicker [31 Hz] (E). Red lines represent right eye (OD) and blue lines represent left eye (OS). In all conditions, ERG responses were severely reduced or non-recordable.

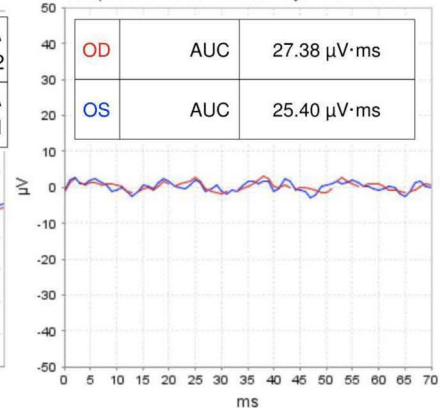
Scotopic 0.01 ERG



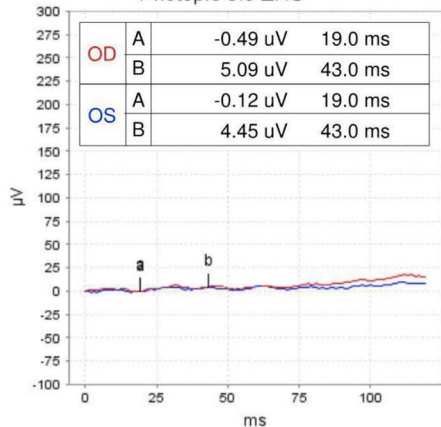
Scotopic 3.0 ERG



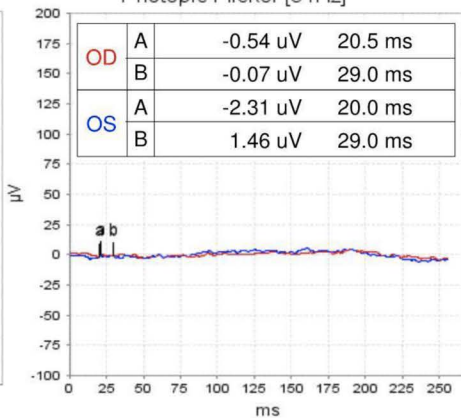
Scotopic 3.0 ERG Oscillatory Potentials



Photopic 3.0 ERG



Photopic Flicker [31Hz]



— OS
— OD