**Supplementary Materials**

# Target 5000: Target Capture Sequencing for Inherited Retinal Degenerations

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**Table S1.** Full list of genes captured in NGS panel.

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| **A** | *BBS9* | *CNGB3* | *GNAT1* | **K** | *NEK2* | *PEX1* | *RDH12* | **T** | *WFS1* |
| *ADIPOR1* | *BEST1* | *CNNM4* | *GNAT2* | *KCNJ13* | *NEUROD1* | *PEX2* | *RDH5* | *TEAD1* | *WFS1* |
| *ABCA4* | **C** | *COL11A1* | *GNB3* | *KCNV2* | *NMNAT1* | *PEX7* | *RGR* | *TIMM8A* | *WHRN* |
| *ABCC6* | *C12orf65* | *COL2A1* | *GNPTG* | *KIAA1549* | *NPHP1* | *PGK1* | *RGS9* | *TIMP3* | **Z** |
| *ABHD12* | *C1QTNF5* | *COL9A1* | *GPR179* | *KIF11* | *NPHP3* | *PHYH* | *RGS9BP* | *TMEM126A* | *ZNF408* |
| *ACBD5* | *C21orf2* | *CRB1* | *GRK1* | *KIZ* | *NPHP4* | *PITPNM3* | *RHO* | *TMEM216* | *ZNF423* |
| *ADAM9* | *C2orf71* | *CRX* | *GRM6* | *KLHL7* | *NR2E3* | *PLA2G5* | *RIMS1* | *TMEM237* | *ZNF513* |
| *ADAMTS18* | *C8orf37* | *CSPP1* | *GUCA1A* | **L** | *NR2F1* | *PLK4* | *RLBP1* | *TOPORS* | |
| *ADGRA3* | *CA4* | *CTBP2* | *GUCA1B* | *LAMA1* | *NRL* | *PNPLA6* | *ROM1* | *TREX1* | |
| *ADGRV1* | *CABP4* | *CTNNA1* | *GUCY2D* | *LCA5* | *NYX* | *POC1B* | *RP1* | *TRIM32* | |
| *ADIPOR1* | *CACNA1F* | *CYP4V2* | **H** | *LRAT* | **O** | *POMGNT1* | *RP1L1* | *TRNT1* | |
| *AGBL5* | *CACNA2D4* | **D** | *HARS* | *LRIT3* | *OAT* | *PRCD* | *RP2* | *TRPM1* | |
| *AHI1* | *CAPN5* | *DHDDS* | *HGSNAT* | *LRP5* | *OFD1* | *PRDM13* | *RP9* | *TSPAN12* | |
| *AIPL1* | *CC2D2A* | *DHX38* | *HK1* | *LZTFL1* | *OPA1* | *PROM1* | *RPE65* | *TTC8* | |
| *ALMS1* | *CDH23* | *DMD* | *HMCN1* | **M** | *OPA3* | *PRPF3* | *RPGR* | *TTLL5* | |
| *ARL2BP* | *CDH3* | *DRAM2* | *HMX1* | *MAK* | *OPN1LW* | *PRPF31* | *RPGR* | *TTPA* | |
| *ARL3* | *CDHR1* | *DTHD1* | **I** | *MAPKAPK3* | *OPN1MW* | *PRPF4* | *RPGRIP1* | *TUB* | |
| *ARL6* | *CEP164* | **E** | *IDH3B* | *MERTK* | *OPN1SW* | *PRPF6* | *RPGRIP1L* | *TUBGCP4* | |
| *ASRGL1* | *CEP250* | *EFEMP1* | *IFT140* | *MFN2* | *OTX2* | *PRPF8* | *RS1* | *TUBGCP6* | |
| *ATF6* | *CEP290* | *ELOVL4* | *IFT172* | *MFRP* | **P** | *PRPH2* | *RTN4IP1* | *TULP1* | |
| *ATXN7* | *CERKL* | *EMC1* | *IFT27* | *MFSD8* | *PANK2* | *PRPS1* | **S** | **U** | |
| **B** | *CFH* | *EXOSC2* | *IMPDH1* | *MIR204* | *PAX2* | **R** | *SAG* | *UNC119* | |
| *BBIP1* | *CHM* | *EYS* | *IMPG1* | *MKKS* | *PCDH15* | *RAB28* | *SDCCAG8* | *USH1C* | |
| *BBS1* | *CIB2* | **F** | *IMPG2* | *MKS1* | *PCYT1A* | *RAX2* | *SEMA4A* | *USH1G* | |
| *BBS10* | *CLN3* | *FAM161A* | *INPP5E* | *MTTP* | *PDE6A* | *RB1* | *SLC24A1* | *USH2A* | |
| *BBS12* | *CLRN1* | *FLVCR1* | *INVS* | *MVK* | *PDE6B* | *RBP3* | *SLC25A46* | **V** | |
| *BBS2* | *CLUAP1* | *FSCN2* | *IQCB1* | *MYO7A* | *PDE6C* | *RBP4* | *SLC7A14* | *VCAN* | |
| *BBS4* | *CNGA1* | *FZD4* | *ITM2B* | **N** | *PDE6G* | *RCBTB1* | *SNRNP200* | **W** | |
| *BBS5* | *CNGA3* | **G** | **J** | *NBAS* | *PDE6H* | *RD3* | *SPATA7* | *WDPCP* | |
| *BBS7* | *CNGB1* | *GDF6* | *JAG1* | *NDP* | *PDZD7* | *RDH11* | *SPP2* | *WDR19* | |

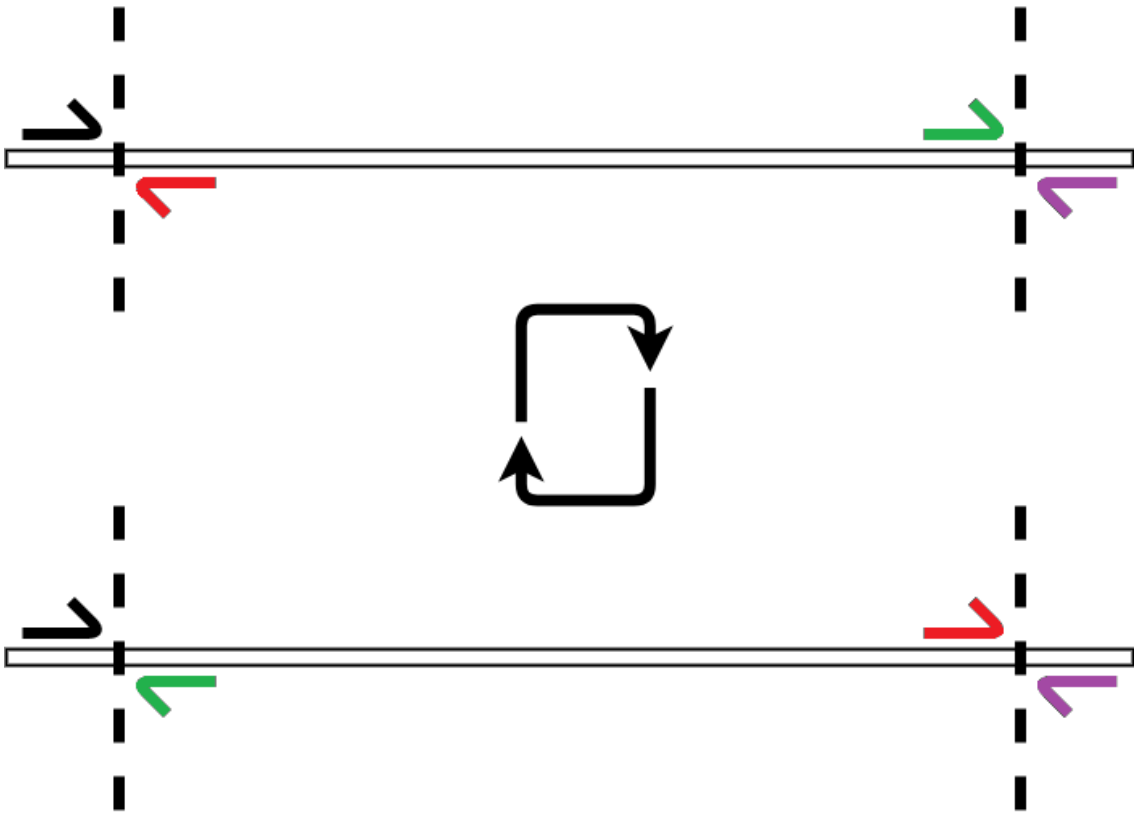

**Figure S1.** An illustration of the PCR strategy designed to detect a large homozygous inversion. Primers are indicated by colour; OAT-1 (**black**), OAT-2 (**red**), OAT-3 (**green**) and OAT-4 (**purple**). Broken lines indicate genomic breakpoints. Circular arrows indicate an inversion event. The top illustration depicts how primers would anneal around the breakpoints in a control sample. The bottom illustration shows how the internal primers (OAT-2 and OAT-3) are shuffled in an inversion event to form new primer pairings, detectable by PCR analysis.

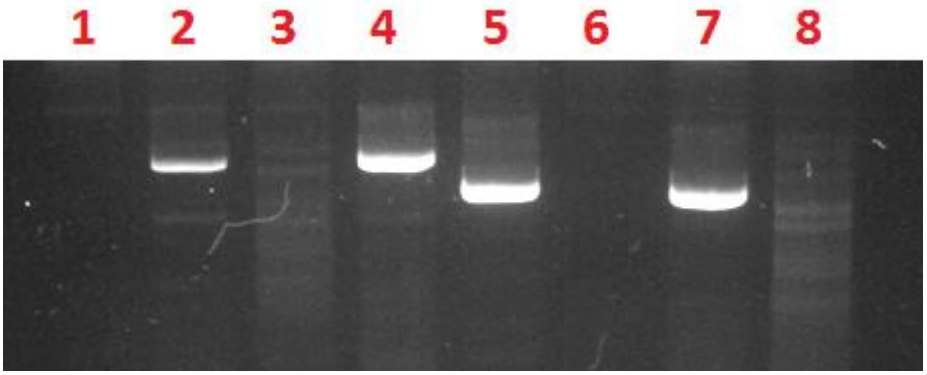


**Figure S2.** Confirmation of an *OAT* inversion using strategic PCR design. If the wildtype *OAT* sequence is present, the primer sets 1 + 2 and 3 + 4 can be used to generate a PCR product. If the inversion has occurred primer sets 1 + 3 and 2 + 4 can be used to generate of a PCR product. Unaffected patients is positive for products of a wildtype sequence. Patient H58 is positive only for inversion-specific products. Lane 1: Patient H58 Primers 1 and 2; Lane 2: Patient H58 Primers 3 and 4; Lane 3: Patient H58 Primers 1 and 3; Lane 4: Patient H58 Primers 2 and 4; Lane 5: Unaffected Patient Primers 1 and 2; Lane 6: Unaffected Patient Primers 3 and 4; Lane 7: Unaffected Patient Primers 1 and 3; Lane 8: Unaffected Patient Primers 2 and 4.

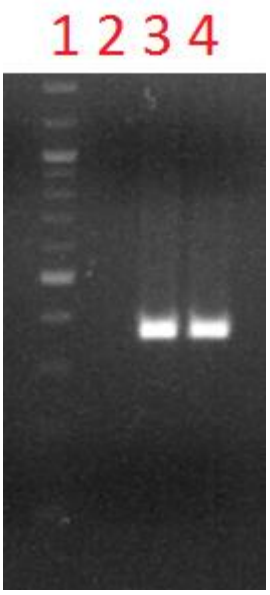


**Figure S3.** Gel confirmation of *USH2A* deletion. Primers were designed to target the region of deletion in affected patients C41 and E15. A product of the desired size (~300bp) would only be formed if the deletion was present. The sample from the unaffected sibling in the same pedigree, P36, did not form a product. A product was formed from the samples of C41 and E15. Lane 1: 100 bp Ladder; Lane 2: Unaffected Patient P36; Lane 3: Affected Patient C41; Lane 4: Affected Patient E15.

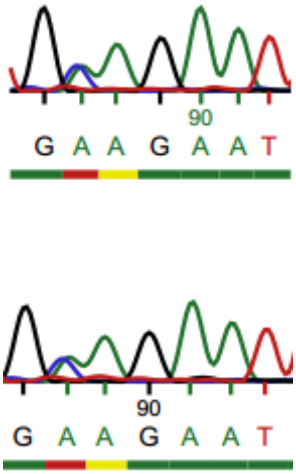


**Figure S4.** Sanger sequencing trace of *USH2A* mutation, p.Cys759Phe. The top trace is unaffected patient P36 and the bottom trace is affected patient E15. It is clear that both patients share the same heterozygous point mutation.

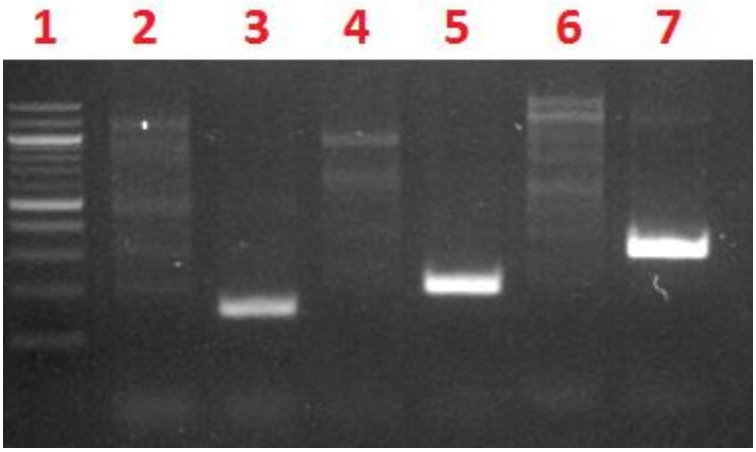


**Figure S5.** Analysis of *USH1C* deletion by use of PCR products. Primers were designed to amplify each exon believed to be deleted in Patient 1363. If the sequence for the exon was present a product would be formed. Exons 3 and 4 were designed as a single amplicon due to their proximity. The unaffected individual is positive for all of the *USH1C* exons. Patient 1363 is negative for all *USH1C* exons. Lane 1: 100 bp Ladder; Lane 2: Patient 1363 with primers for exon 1; Lane 3: Unaffected Patient with primers for exon 1; Lane 4: Patient 1363 with primers for exon 2; Lane 5: Unaffected Patient with primers for exon 2; Lane 6: Patient 1363 with primers for exons 3 + 4; Lane 7: Unaffected Patient with primers for exons 3 + 4.