

Supplemental Figures

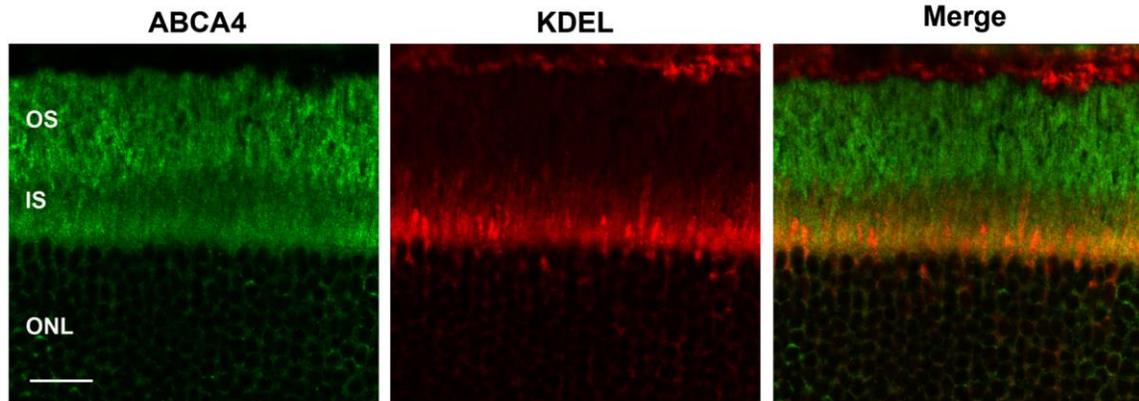


Figure S-1 Fluorescence microscopy images showing co-localization of p.Asn965Ser ABCA4 and KDEL in the ER of photoreceptors. Retinal cryosection of a p.Asn965Ser mouse was labeled for ABCA4 with the Rim 5B4 monoclonal antibody (green) and a polyclonal antibody (red) to the KDEL ER retention sequence. Merged image shows the co-localization of a fraction of the ABCA4 mutant with the ER marker in the inner segment (IS) just above the outer nuclear layer (ONL). ABCA4 is also present in the outer segment (OS) layer. Bar – 20 μm .

Targeting Vector Schematic

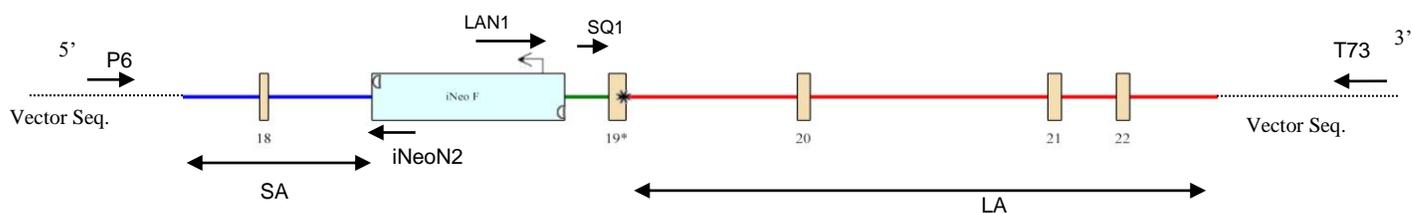


Figure S-2: Targeting Vector Schematic for generation of the p.Asn965Ser ABCA4 mutant mouse: The targeting vector was confirmed by restriction analysis after each modification step and by sequencing using primers designed to read from the selection cassette into the genomic sequence containing the point mutation. P6 and T73 primers anneal to the BAC subclone sequence and read into the 5' and 3' ends of the subcloned vector. The sequencing results with primers confirmed the introduction of the point mutation in exon 19 and that no error was introduced into the PCR modified region. SA – short arm; LA – long arm.

PCR primers used for sequencing:

Primer P6: 5' - GAG TGC ACC ATA TGG ACA TAT TGT C-3'
 Primer T73: 5' - TAA TGC AGG TTA ACC TGG CTT ATC G-3'
 Primer LAN1: 5' - CCA GAG GCC ACT TGT GTA GC-3'
 Primer iNeoN2: 5' - AGT ATG GCT TTC CTT CCC GAT GG-3'
 Primer SQ1: 5' - GCA GGT GAA GTT ACA GCC AGA G-3'