Supplementary material of:

A Slc38a8 mouse model of FHONDA syndrome faithfully recapitulates the visual deficits of albinism without pigmentation defects

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Supplementary Figure 1

BLASTP (NIH/NCBI) alignment of SLC38A8 human (A6NNN8 UniProtKB, 434 amino acides) and Slc38a8 (Q5HZH7 UniProtKB, 431 amino acids) mouse proteins. The p.200Q (human) and p.197Q (mouse) amino acid residues are highlighted in bold and indicated with a vertical arrow.

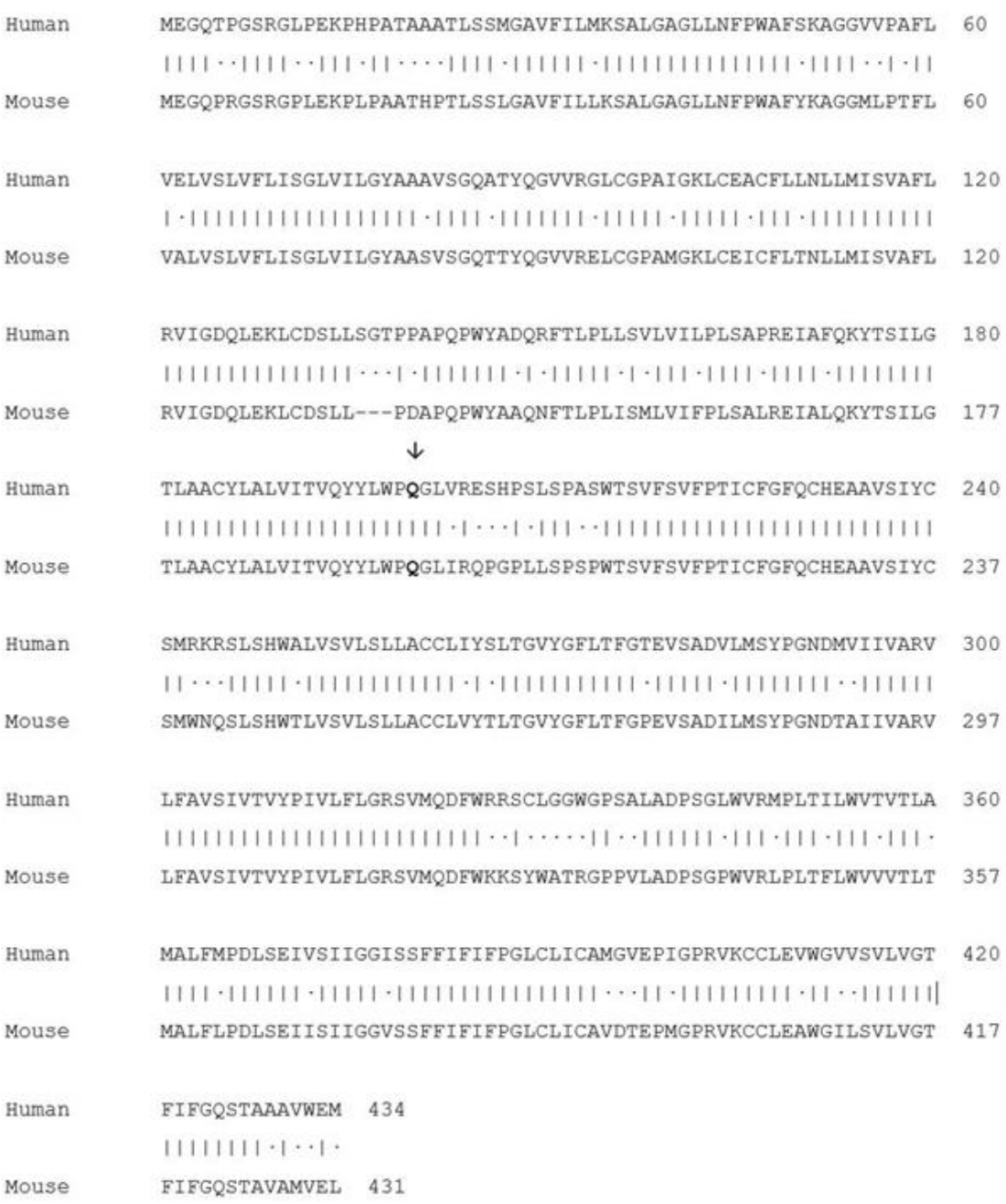
Supplementary Figure 2

Identification of FHONDA mutant founder (F0) mice after genome editing with CRISPR-Cas9 tools. **A**. Detection of FHONDA mutant founder (F0) mice by PCR. Three positive founder mice are shown, including #A8578, the mouse that carries the p.199Pro* mutant allele eventually selected for this study. Larger bands correspond to insertions and smaller bands correspond to deletions. **B**. Detection of FHONDA mutant founder (F0) mice by T7 Endonuclease I assay, which detects indels (insertions/deletions). Five positive founder mice are shown, including #A8578. **C**. Germline transmission test of FHONDA mouse mutant founder (F0) #A8578. The deleted product (smaller band) is found in numerous F1-derived animals.

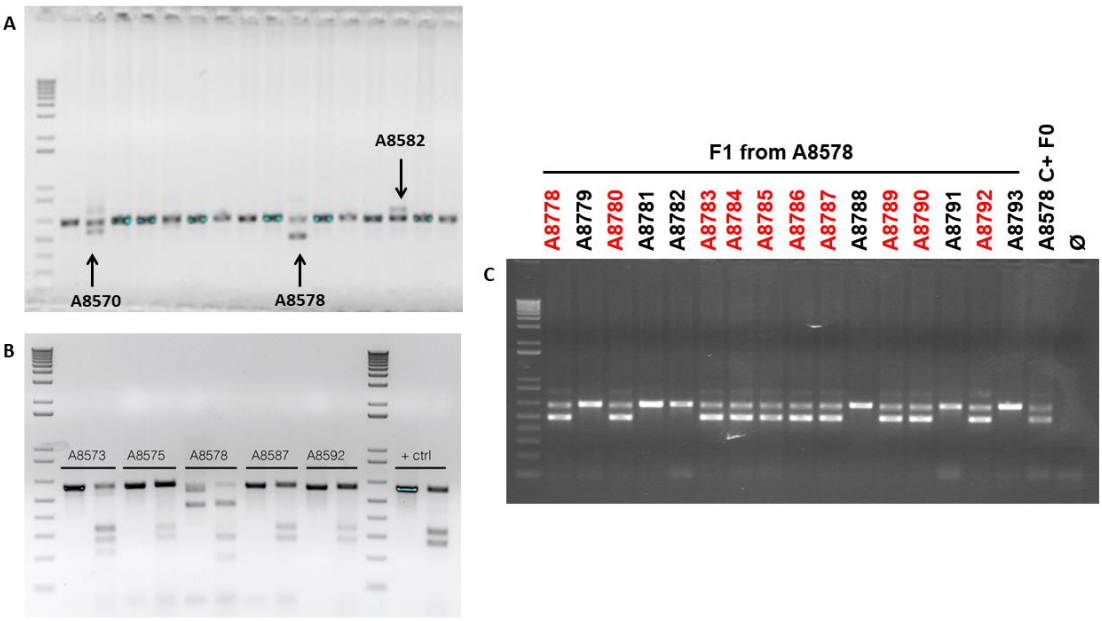
Supplementary Figure 3

Number of pigmented melanosomes per RPE cell, counted from electron microscope images, in adult (2-3 months old) wild-type pigmented C57BL/6J, FHONDA *Slc38a8* heterozygous “HET” (+/-) and FHONDA *Slc38a8* homozygous mutant “HOM” (-/-). Mean ± SEM, N=18-20 RPE cells, 1-factor ANOVA with Bonferroni correction for multiple comparisons. No statistically significant differences were detected.

Supplementary Figure 1



Supplementary Figure 2



Supplementary Figure 3