

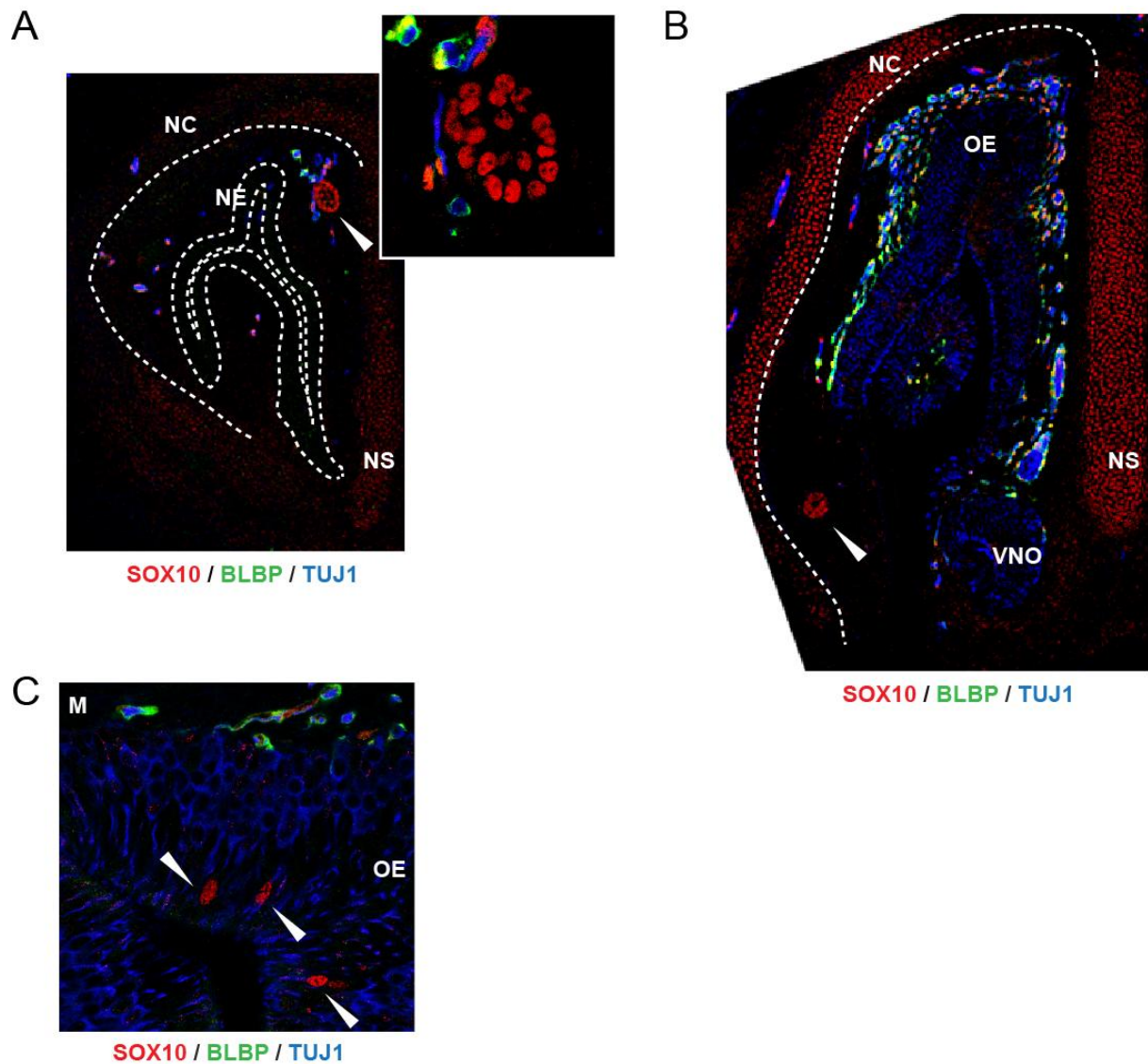
Supplemental Figure S1. Familial data and additional functional tests of the mutations

(A) Pedigrees of family A and family D along with the phenotyping and genotyping data available. The family tree legends are given in the box.

(B) Dominant-negative activity of the SOX10 mutant proteins in luciferase assay. The wild-type (wt) and increasing quantities of the mutant SOX10 expression vector were co-transfected with a reporter construct containing the *MITF* promoter. Reporter gene activation is presented as luciferase fold induction relative to the empty vector. Results are the mean \pm s.e.m of three different experiments, each performed in duplicate.

(C) Pedigree of family G along with the phenotyping and genotyping data available. The family tree legends are the same as in (A).

(D) Luciferase reporter gene analysis of the p.Met108Thr and p.Arg151Cys mutants. The wild-type (wt) or mutant SOX10 expression vector was co-transfected with a reporter construct containing the *MITF* promoter and the PAX3 cofactor expression vector. Reporter gene activation is presented as luciferase fold induction relative to the empty vector. Results are the mean \pm s.e.m of three different experiments, each performed in duplicate.

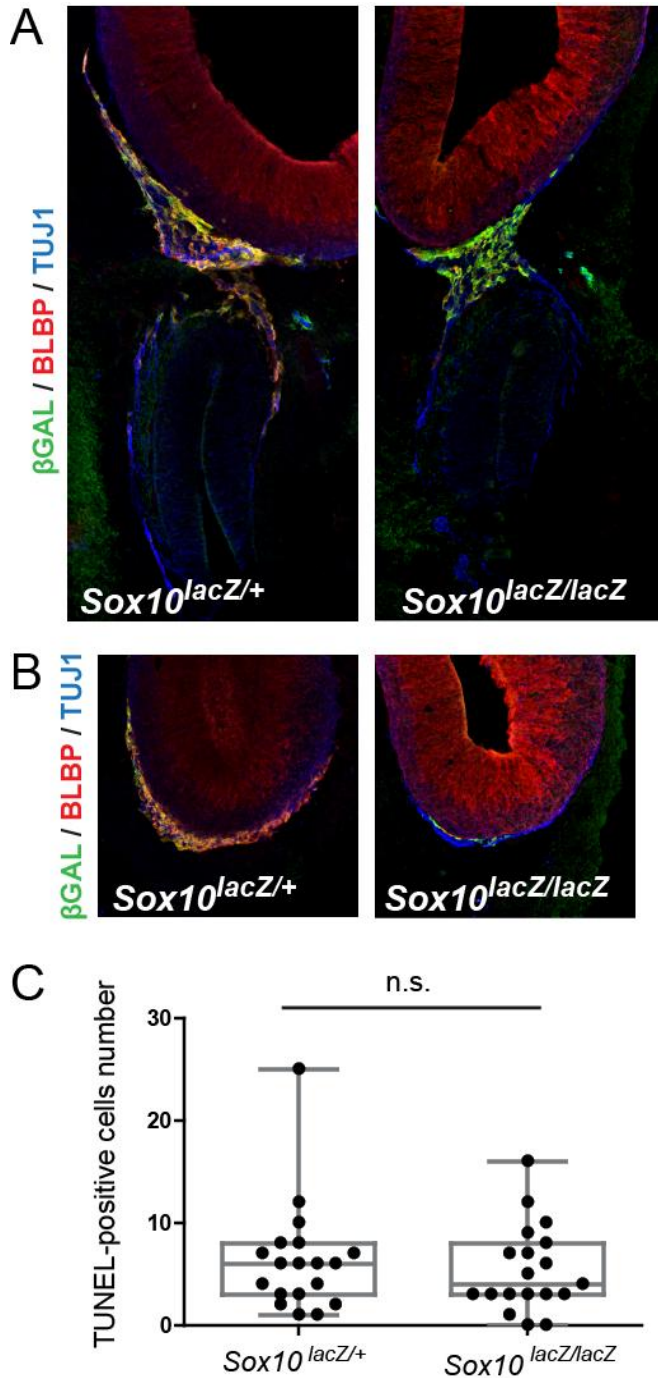


Supplemental Figure S2. SOX10 expression outside of OECs in the olfactory system of E14.5 mice

(A and B) Triple labeling of E14.5 mouse embryo nasal (A) and olfactory (B) epitheliums (coronal sections) for SOX10 (red), the neuronal marker TUJ1 (blue), and the OEC marker BLBP (green). The arrowhead indicates glandular structures in the nasal mesenchyme. The inset in (A) shows a higher magnification.

(C) Triple labeling of E14.5 mouse olfactory epithelium (coronal sections, higher magnification) for SOX10 (red), the neuronal marker TUJ1 (blue), and the OEC marker BLBP (green). Arrowheads indicate SOX10-positive nuclei within the olfactory epithelium.

NC: nasal capsule; NE: nasal epithelium; NS: nasal septum; OE: olfactory epithelium; VNO: vomeronasal organ.



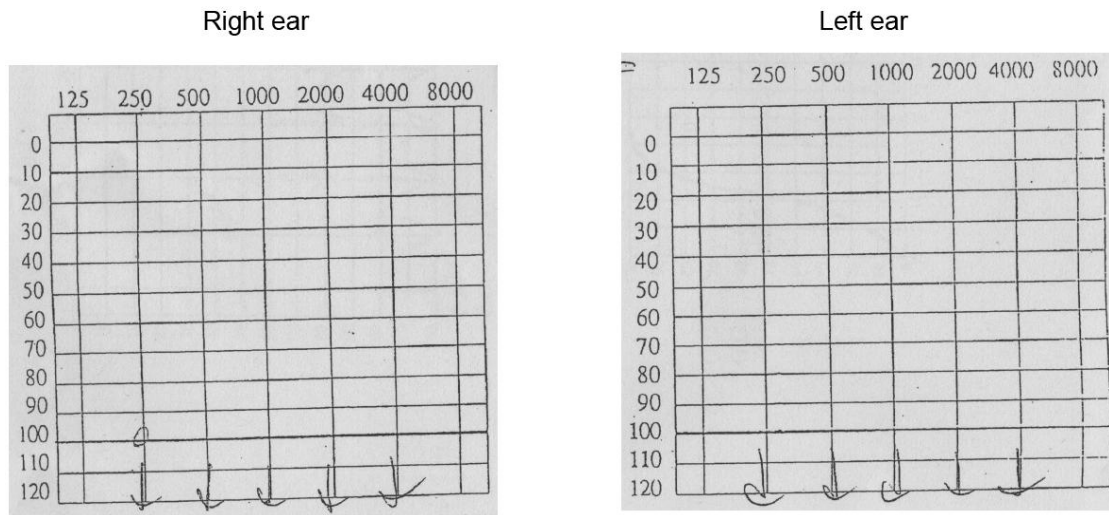
Supplemental Figure S3. Early defect of OECs in the *Sox10* mutant mice

(A) General overview of the olfactory system at E12.5 with triple immunolabeling for β -galactosidase (green), the neuronal marker TUJ1 (blue), and the OEC marker BLBP (red) in heterozygous (*Sox10*^{lacZ/+}, left panel) and homozygous (*Sox10*^{lacZ/lacZ}, right panel) mutant embryos.

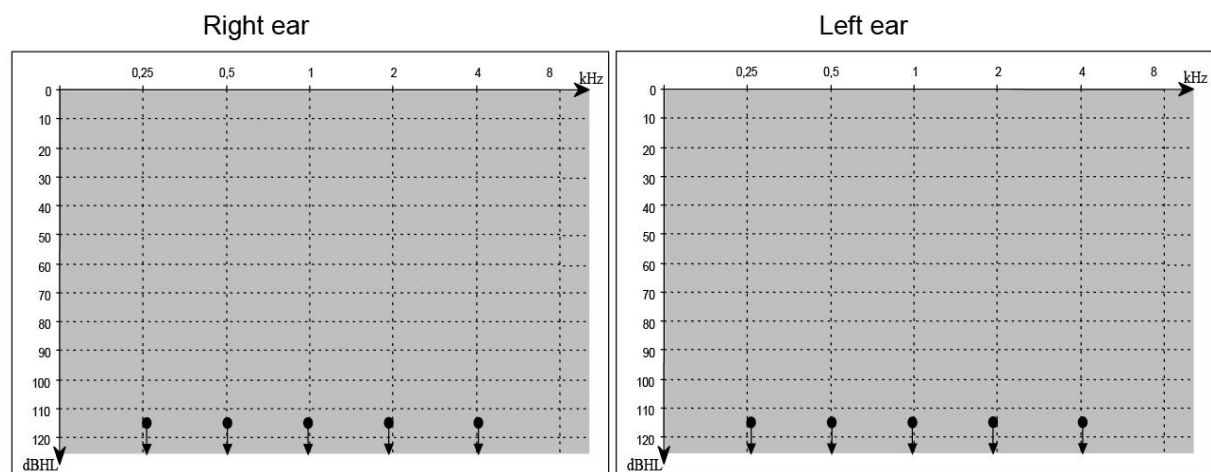
(B) A more rostral section of the olfactory bulb in heterozygotes (left panel) and homozygotes (right panel).

(C) Box plots showing quantification of apoptotic cells. Each dot represents counting of TUNEL-positive cells in the nasal mesenchyme at E12.5, on one side of a section. The top and bottom of each box represent the 25th and 75th percentiles, respectively. The middle line is the median. Statistical significance was tested with Student's ttest. n.s., not significant.

A Patient D



B Patient E



Supplemental Figure S4. Audiograms

(A and B) Audiograms of patients D and E, respectively.