

wildtype

mutation

Supplementary Figure S4. The hs. *SLC4A2* mutation c.503T>C, p.lle168Thr may affect a hydrophobic interaction in SLC4A2.

The hs. *SLC4A2* mutation was modeled on the crystal structure of its human SLC4A1 paralog (Arakawa T Science 350:680-4, 2015).²² Ala400 in SLC4A1 corresponds to Ala703 in SLC4A2, and faces the hydrophopic amino acid Ile761. The mutation p.Ala703Thr introduces a polar, hydrophilic amino acid with a longer side change and may interfere with the hydrophobic interaction.