**SLC13A5** is the second gene associated with Kohlschütter-Tönz syndrome

**Supplementary figure 1**

**Supplementary figure 1:** MRI of individual E:II:1 at age 11 months. A, B, D and E are axial T2-weighted images, E is a sagittal T1-weighted image. The left hemisphere is slightly smaller than the right (A, B). Otherwise there is no atrophy including a normal cerebellar volume (C, E). The signal of the pallidum is slightly hyperintense (B), fitting with non-completed myelination (adequate for age). The signal of the dorsal tegmental tracts is also slightly elevated (D), a nonspecific finding. (F) shows a proton spectroscopy from the left centrum semiovale; there are no visual abnormalities.