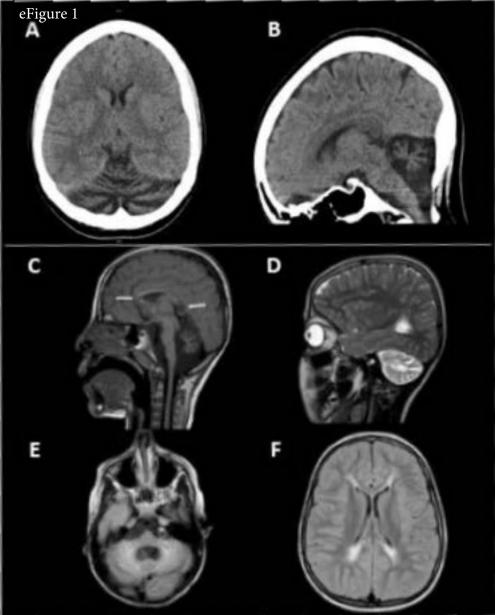
eFigure 1. Brain imaging. A and B Brain CT scan of Patient 1 at the age of 40 years. There is marked atrophy of the cerebellum and cerebellar peduncles and to a lesser extent atrophy of the brain stem. C – F Brain MRI scan of patient 3 at the age of 6 years. C: midsagittal T1 weighted image showing atrophy of the cerebellar vermis, thin corpus callosum and hypodense lesions in the rostrum and splenium of the corpus callosum (arrows). D: parasagittal T2 weighted image showing prominent atrophy of the cerebellar hemisphere. E: Axial FLAIR image showing atrophy and hyperintense signal of the cerebellar vermis and hemispheres. F: Axial FLAIR image showing normal cortical development, no cerebral atrophy, with bilateral hyperintense lesions in the periventricular and subcortical white matter of the frontal and occipital lobes.



eTable1: Collated table of individuals published with pathogenic variants in ANO10.

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