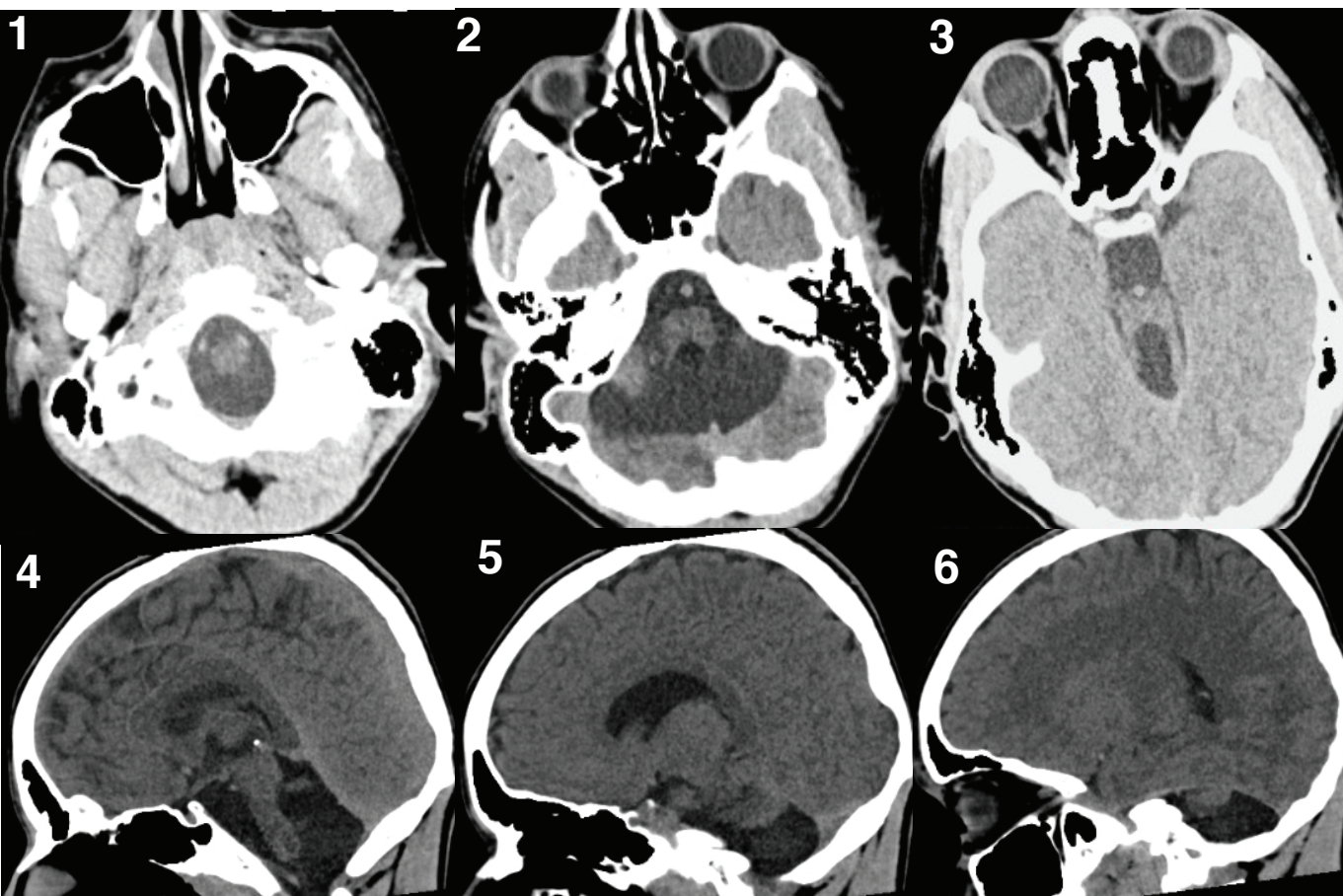
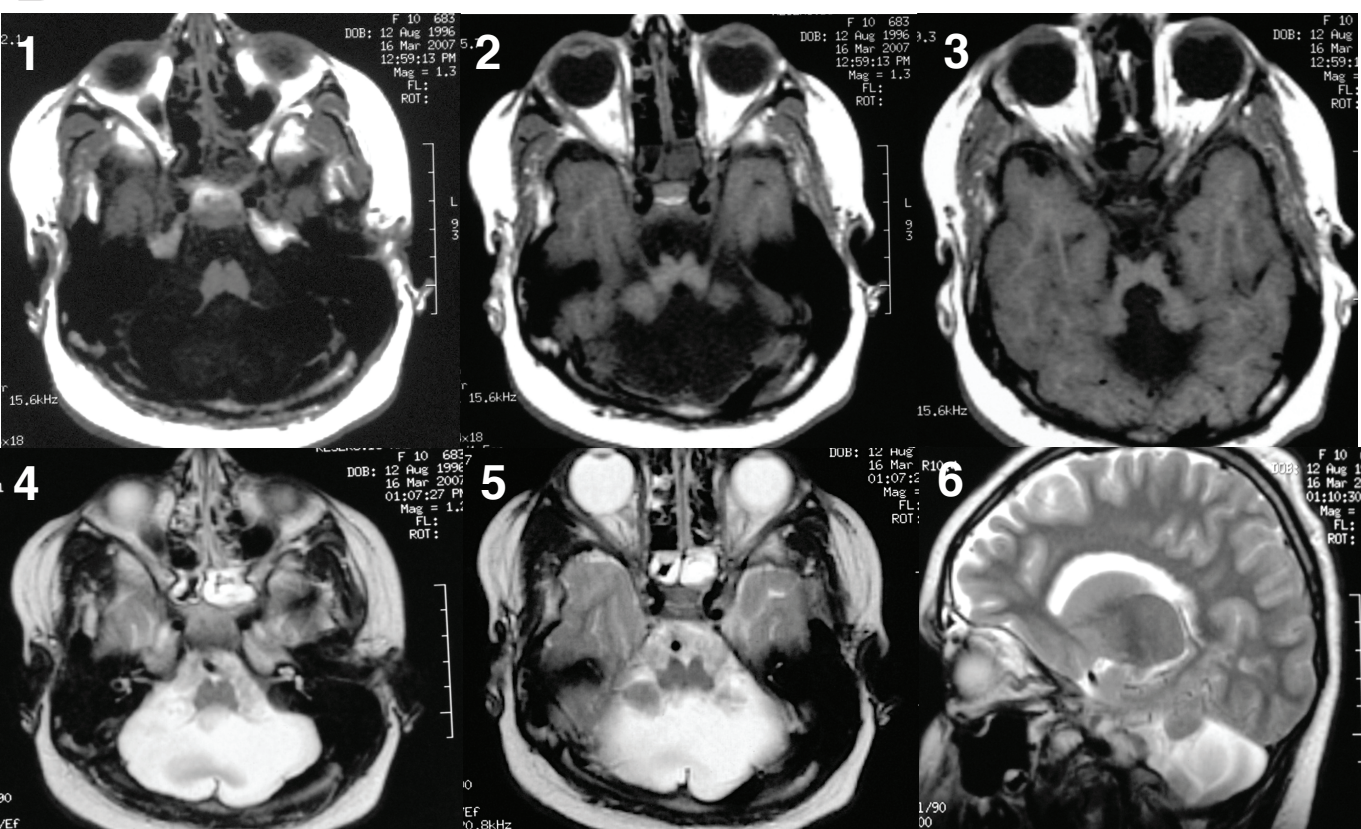
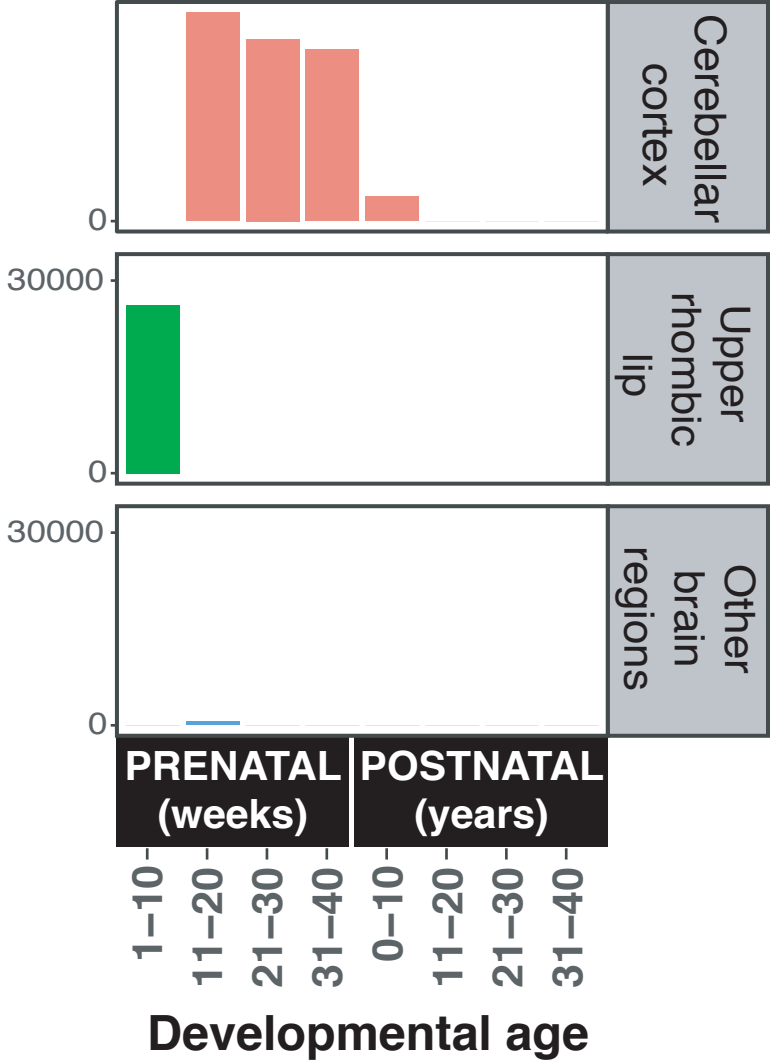


A**B**

ATOH1 expression levels

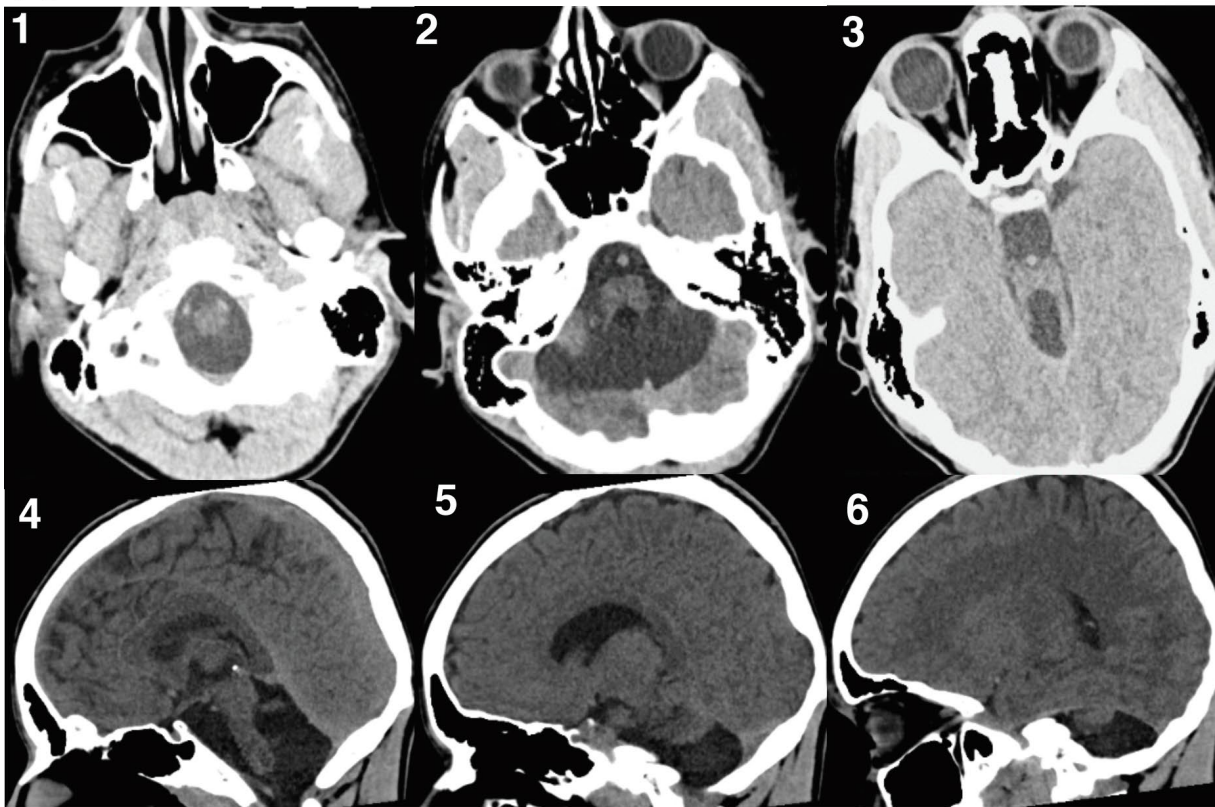
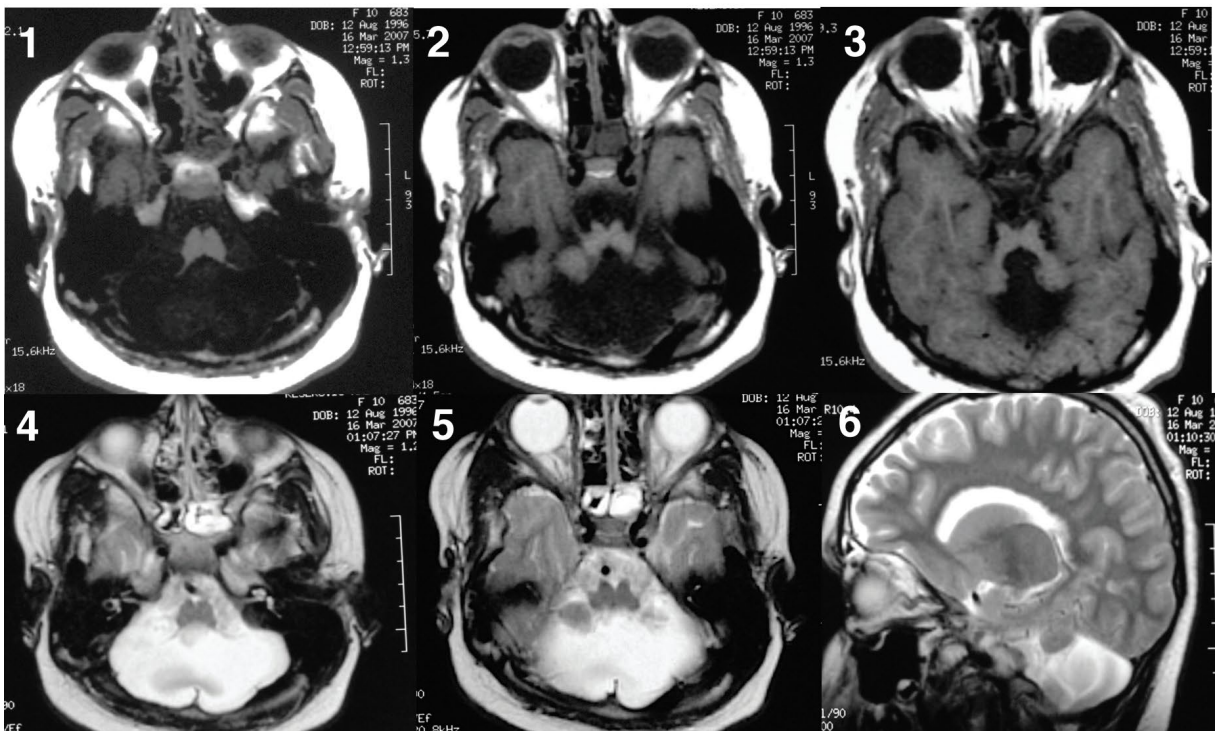


Supplemental online data

eFigure 1 CT and MR images of a proband II-1 and his affected sister II-3.

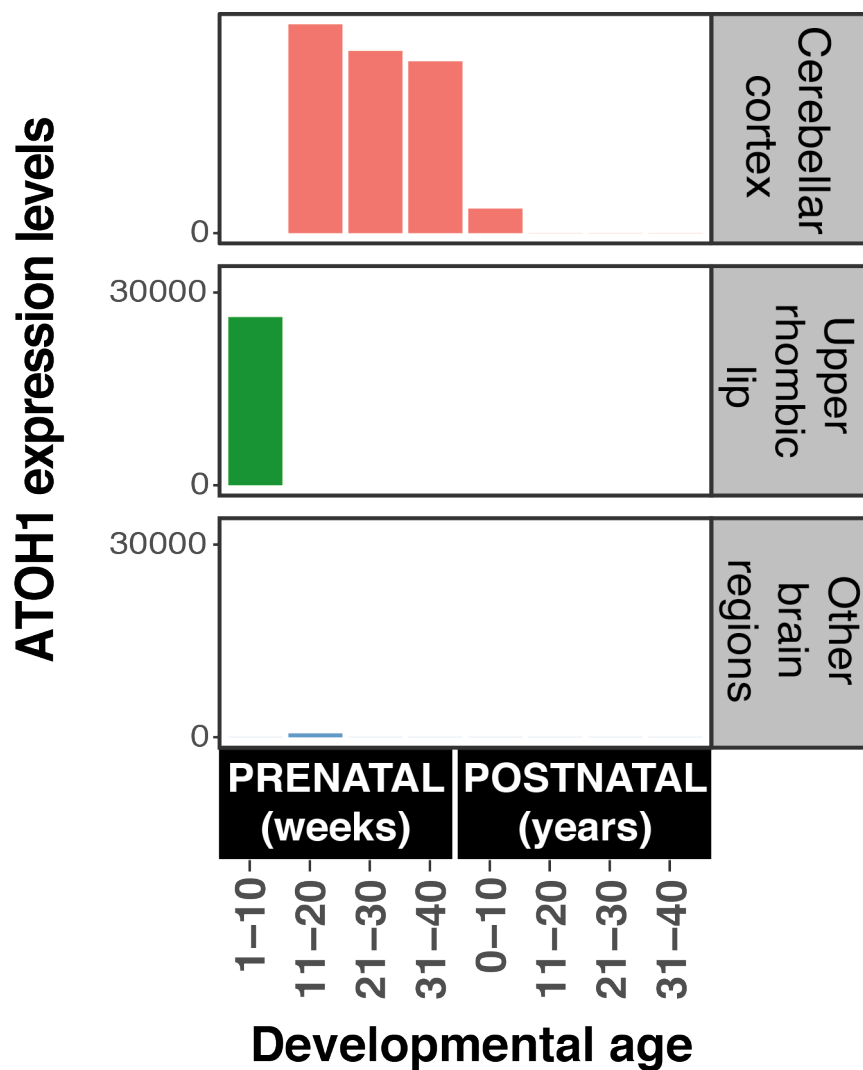
(A) CT images of a proband II-1. Please notice: 1 - normal medulla oblongata; 2 – hypoplastic inferior cerebellar peduncle; 3 – exposed superior cerebellar peduncles of the absent vermis – molar tooth sign; 4 – low cerebellar tentorial position, flattened splenium of corpus callosum; 5 and 6 – severely hypoplastic cerebellar hemispheres.

(B) MR images of an affected sister II-3. 1 – cystic dilatation of the 4th ventricle communicating with cisterna magna; 2 - cystic dilatation of the 4th ventricle communicating with cisterna magna, severely hypoplastic pons with anterior incisure and absent transverse pontine fibres, prominent trigeminal nerves; 3 – hypoplastic mesencephalon; 4 – hypoplastic or absent statoacoustic nerve; 5 – severely hypoplastic pons, middle cerebellar peduncles and cerebellar hemispheres. Please, note enlarged ocular bulbs; 6 – very hypoplastic cerebellar hemispheres, widened frontal subarachnoid spaces indicating frontal atrophy.

A**B**

eFigure 2

The expression pattern of the *ATOH1* gene in various developmental stages of human brain development based on the RNAseq data in the Allen human brain development atlas. The *ATOH1* gene expression profile highlights that its expression is confined to the developing cerebellum, including the upper rhombic lip, which is the origin of cerebellar territory. Expression values are given in RPKM.



eTable 1 Special clinical features noticed on CT and MR images.

Clinical Features seen on CT and MR images	Proband II-1	Affected sister II-3
Inferior cerebellar peduncles	hypoplasia	hypoplasia
Middle cerebellar peduncles	hypoplasia	hypoplasia
Pons	hypoplasia	hypoplasia
Cerebellar hemispheres	hypoplasia	hypoplasia
Cerebellar vermis	absent	absent
Posterior fossa	small	
Corpus callosum	flattened splenium of corpus callosum	flattened posterior part of corpus callosum
Frontal lobes		atrophy
4 th ventricle	widened	enlarged with cystic dilatation
Lateral ventricles	mild widened frontal horns of lateral ventricles	
Optic tracts	hypoplasia	hypoplasia
Ocular bulb	enlarged ocular bulb with small coloboma	enlarged
Optic nerves	twisted	
Statoacoustic nerve		hypoplasia
Optic chiasm	dysplastic	hypoplasia