Abnormal cerebellar foliation in *EBF3* mutation

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Figure Dandelion cerebellar sign

MRI T2-weighted imaging in patient, top, and healthy participant, bottom. Radial shape (dandelion sign, A and B) and blurred wavy boundaries (C) of cerebellar folia are seen. Vermis hypoplasia lacking the normal lobules subdivision (A and B vs E and F), facing cerebellar hemispheres, and flattened course of the folia white matter stem (D) in respect to normal rounded appearance (G, H) are noted.

A 5-month-old patient was first evaluated for a hypotonic hyporeflexic syndrome; over years, the phenotype evolved to involve ataxia, developmental delay, and coarse facies. Brain MRI showed abnormal configuration of cerebellar folia, rearranged in radial shape (figure). Whole exome sequencing analysis revealed the c.512 G>A (p.G171D) de novo mutation in *EBF3*. *EBF3* gene function is crucial for neuronal migration during corticogenesis.¹ Cerebellar foliation is complex: folia sprout from anchoring centers,² whose distribution and number determine the shape of lobules and fissures; *EBF3* mutations may disrupt this process, thus causing the peculiar cerebellar lobule appearance seen here.

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Disclosure

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Telethon Undiagnosed Diseases Programme coinvestigators are listed in the appendix 2 at the end of the article.

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