Figure. T1-weighted sagittal (A) and coronal (B) brain MRIs showing total cerebellar agenesis.

Cerebellar agenesis

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A 17-year-old boy with a history of neonatal hypotonia was first observed by us at age 4 years because of persistent ataxia. Brain MRI revealed isolate cerebellar agenesis (CA) (figure), the empty cerebellar space having its signal similar to CSF in all performed sequences. At age 17 years he showed moderate ataxia

(video 1), mild dysmetria (video 2), and no nystagmus. Neuropsychological evaluation evidenced mild mental retardation. Further examination was normal. He attends a normal school, to which he gets by bicycle.

Total or subtotal CA is an extremely rare congenital defect and is thought to be associated with profound deficits in movement.¹ Clinical presentation ranges from early death to variable degrees of cerebellar dysfunction.²

Reports of living patients address the question of whether CA is compatible with functional motor development. Cerebellar development occurs early during embryogenesis, so that plasticity of the remaining brain could explain functional compensation.

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