



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



Cerebellar agenesis

Luigi Titomanlio, MD; Alfonso Romano, MD; and
Ennio Del Giudice, MD, Naples, Italy

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.

Address correspondence and reprints requests to Dr. Ennio Del Giudice, Child Neurology Unit, Department of Pediatrics, Via S. Pansini, 5, 80131 Naples, Italy.

1. Glickstein M. Cerebellar agenesis. *Brain* 1994;117:1209–1212.
2. Timmann D, Dimitrova A, Hein-Kropp C, Wilhelm H, Dorfler A. Cerebellar agenesis: clinical, neuropsychological and MR findings. *Neurocase* 2003;5:402–413.

Neurology[®]

Cerebellar agenesis

Luigi Titomanlio, Alfonso Romano and Ennio Del Giudice

Neurology 2005;64;E21

DOI 10.1212/WNL.64.6.E21

This information is current as of March 21, 2005

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/64/6/E21.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2005/03/20/64.6.E21.DC1 http://n.neurology.org/content/suppl/2007/04/02/64.6.E21.DC2
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/64/6/E21.full#ref-list-1
Citations	This article has been cited by 2 HighWire-hosted articles: http://n.neurology.org/content/64/6/E21.full##otherarticles
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright . All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

