Case 15330

Eurorad

Congenital disorder of glycosylation, a neuroradiologic case report

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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Imaging sequences
Imaging Technique: CT
Imaging Technique: MR
Special Focus: Metabolic disorders Case Type: Clinical Cases
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Patient: 16 years, male

Clinical History:

A 16-year-old male patient with a known congenital disorder of glycosylation, presents to the pediatrics consultation with symptoms of a stroke-like event and transitory paresis of the left arm. After an initial CT, an MRI was scheduled.

Imaging Findings:

The CT showed no early or old signs of ischaemia, nor any intracranial bleeding.
The MRI confirmed no signs of recent ischaemia on the diffusion-weighted images. The CT and MRI both showed a marked hypoplasia and atrophy of the cerebellar folia, involving both hemispheres and the vermis, with enlargement of the cerebellar fissures. The cerebellar parenchyma showed marked T2- and FLAIR-hyperintensity in comparison to the cerebral parenchyma. Associated pontine atrophy was present. The cerebellar and pontine atrophy had clearly progressed in comparison to an earlier MRI of 16 years ago, at the age of 6 months. Upon looking at the first MRI, we learned that there was already clear cerebellar hypoplasia.
Secondary to the cerebellar atrophy, there was a large T2-hyperintense CSF accumulation inferior of the cerebellar vermis. There was no displacement of the cerebellar tentorium. There was no hydrocephalus or ventricle dilatation present.

Discussion:

Congenital disorders of glycosylation (CDG) is an autosomal recessive disorder which is caused by abnormal glycosylation of oligosaccharides, leading to a variety of symptoms and affecting multiple systems. The most common form is CDG type-Ia and is caused by a mutation of the PPM-2 gene, which encodes a cytosolic enzyme phosphomannomutase. This specific mutation was diagnosed in our case at the age of 10 months. [1]
Typically, CDG-Ia patients will demonstrate neurological involvement, with muscular hypotonia, failure to thrive and

psychomotor retardation, often in the first year of life. [1]

MR-imaging typically shows a small hypoplastic cerebellum, including the vermis and both hemispheres in the neonatal period, with progressive volume loss (atrophy) upon follow-up imaging.
The distinction between cerebellar hypoplasia and cerebellar atrophy is not always clear in practice. Cerebellar hypoplasia (CH) is a congenital condition, which presents as an underdevelopment or incomplete development of the cerebellum. In CH, the cerebellar structures are not filling a normal configured posterior fossa. Cerebellar atrophy (CA) however, implies progressive volume-loss of an initial normal cerebellum. In CA, the cerebellar structures are normally formed, yet loss of folial tissue has occurred and secondary, the fissures or interfolial spaces have progressively enlarged. [2]

This case shows a clear hypoplastic cerebellum shortly after birth, on the T1-weighted images, performed at theage of 6 months. Sixteen years later, we notice a progressive atrophy of the cerebellar folia and the pons. In accordance with other studies, we can conclude that that cerebellar atrophy can be superimposed on cerebellar hypoplasia, in CDG type-1a patients. [1, 2, 3]
A case-series by Feraco et. al, found that the presence of high T2/FLAIR signal intensity in the cerebellum was a universal finding in all of their five cases and could help in the differential diagnosis of CA and CH. In our case, this was also a marked finding. [1]
In dandy-walker malformation there is an enlarged posterior fossa, with partial to complete vermian aplasia, and secondary elevated cerebellar tentorium. Malformations like this, are therefore easily excluded from the differential diagnosis.

The stroke-like event our patient presented with, is reportedly a frequent finding in CDH-1a patients, and is most likely caused by an active epileptic inhibitory process rather than real ischaemia. [4]

To summarize, we present a typical case of CDG-1a related ponto-cerebellar degeneration. A small T2-FLAIR hyperintense and hypoplastic cerebellum, often associated with superimposed pontocerebellar atrophy can be suggestive for the diagnosis of CDG.

Differential Diagnosis List: Congenital disorder of glycosylation type-1a, Dandy-walker malformation, Pontocerebellar atrophy of other origin.

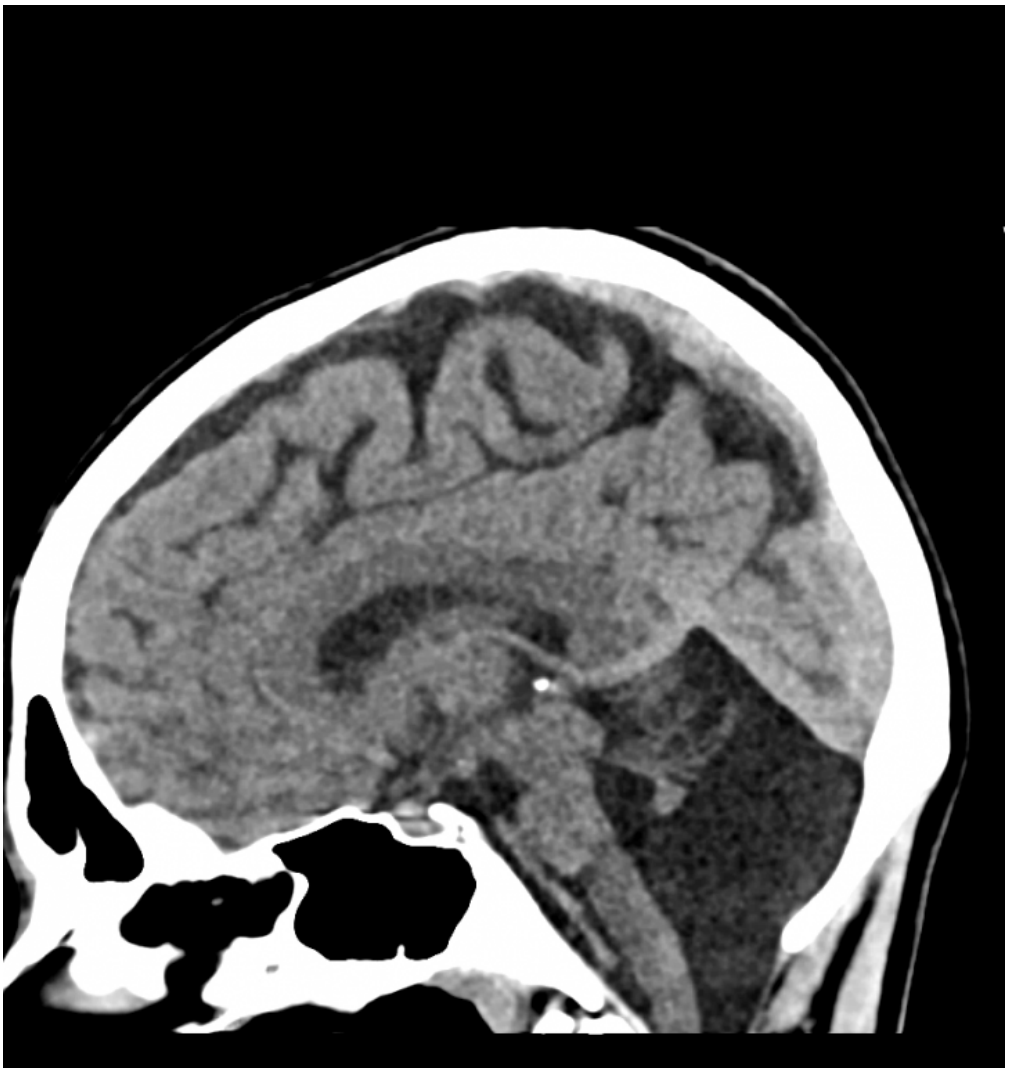
Final Diagnosis: Congenital disorder of glycosylation type-1a

References:

Feraco P, Mirabelli-Badenier M, Severino M, Alpigiani MG, Di Rocco M, Biancheri R, et al. (2012) The shrunken, bright cerebellum: a characteristic MRI finding in congenital disorders of glycosylation type 1a. AJNR Am J Neuroradiol 2012;33(11):2062–7 (PMID: 22723063)
Boltshauser E. et al. (2004) Cerebellum—small brain but large confusion: a review of selected cerebellar malformations and disruptions. Am J Med Genet A 2004; 126A: 376– 85 (PMID:15098235)
Poretti A, Wolf NI, Boltshauser E. (2008) Differential diagnosis of cerebellar atrophy in childhood. Eur J Paediatr Neurol 2008; 12: 155– 67 (PMID: 17869142)
Argirios Dinopoulos et al. (2007) Radiologic and Neurophysiologic Aspects of Stroke-like Episodes in Children With Congenital Disorder of Glycosylation Type Ia. Pediatrics Vol. 119 No. 3 March 01, 2007 (PMID:17308246)

Figure 1

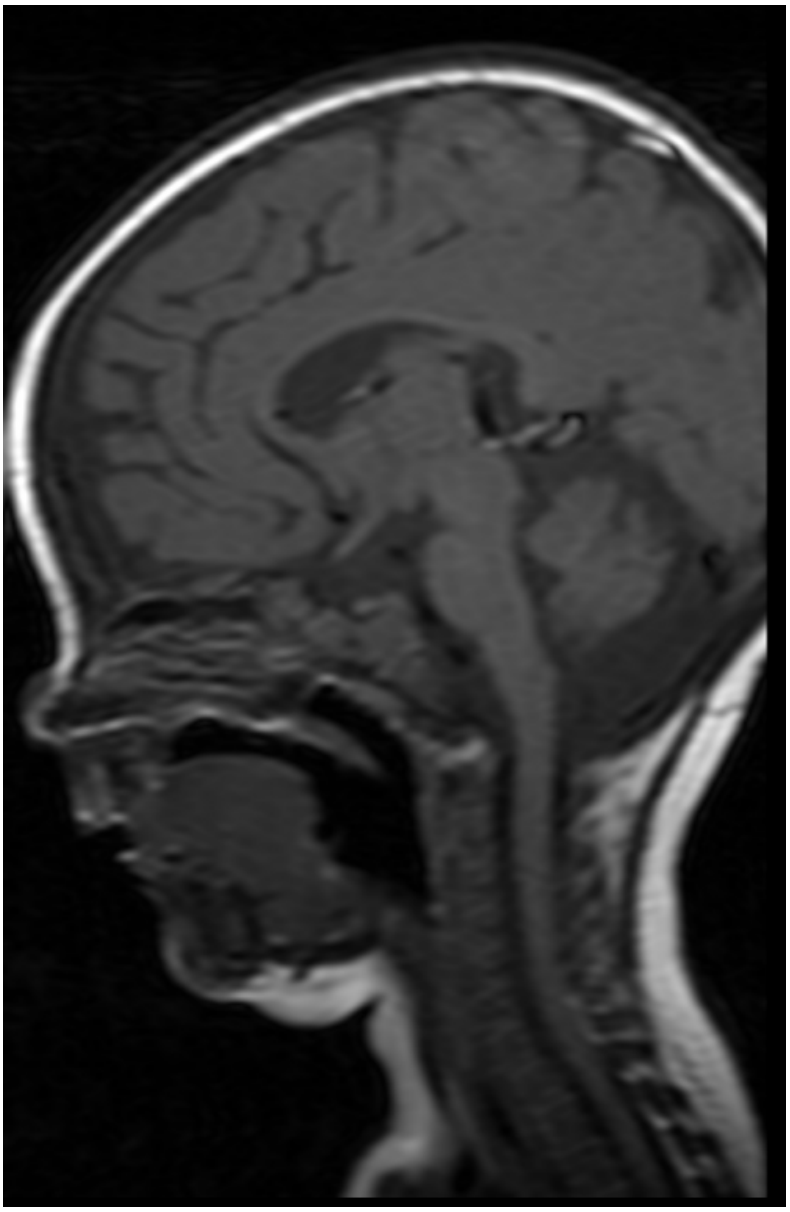
a



Description: Sagittal reconstruction of a CT shows an increased volume-loss of the cerebellum when compared with previous imaging (Figure 3).
Note the progressive widening of the cerebellar folia and progressive pontine atrophy. **Origin:** Radiology department of the university hospital of Leuven.

Figure 2

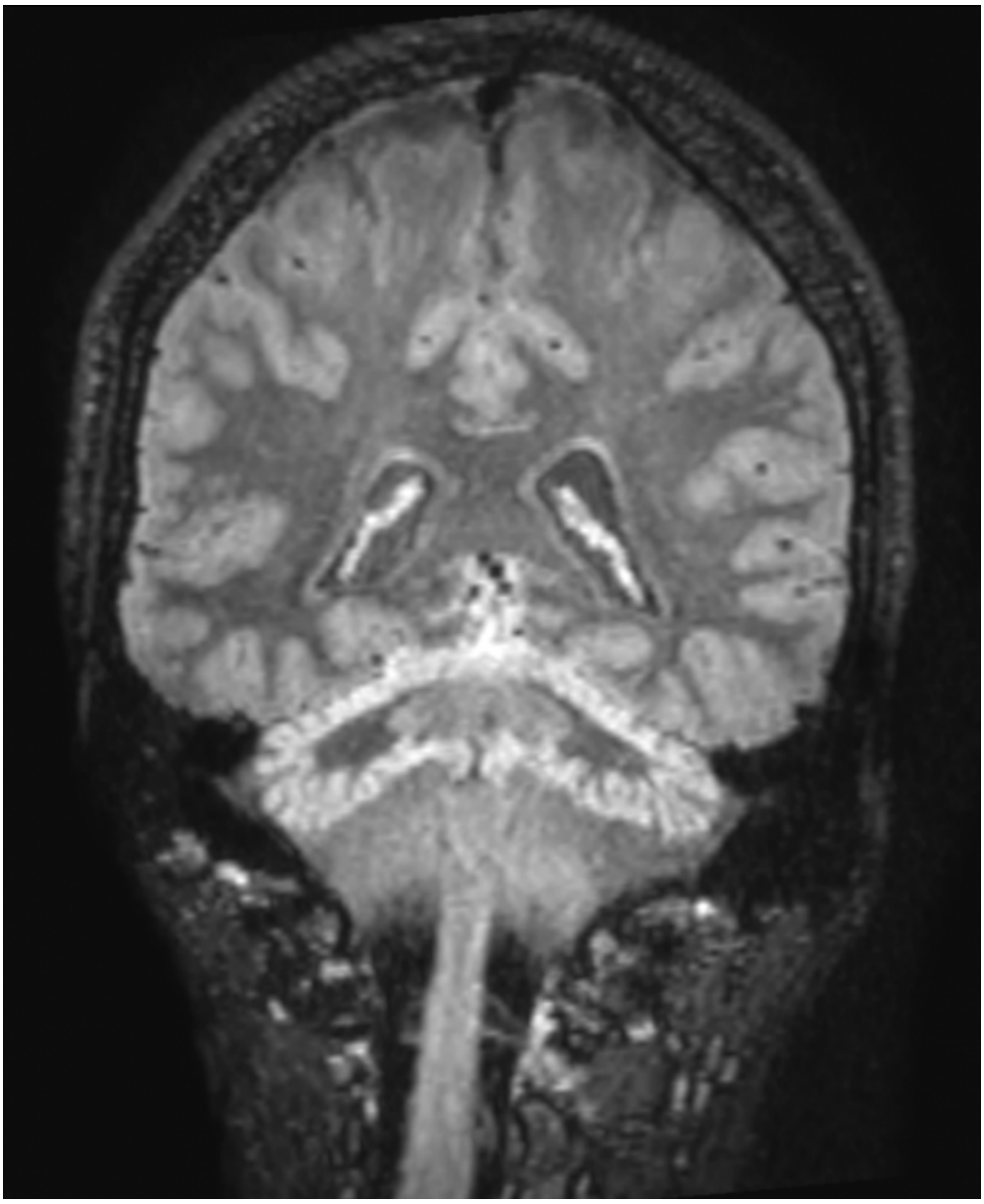
a



Description: Sagittal T1-weighted image, at 6 months shows a small cerebellum in an otherwise normal posterior fossa, suggestive of cerebellar hypoplasia. **Origin:** Radiologic deparment of the university hospital of Leuven, Belgium

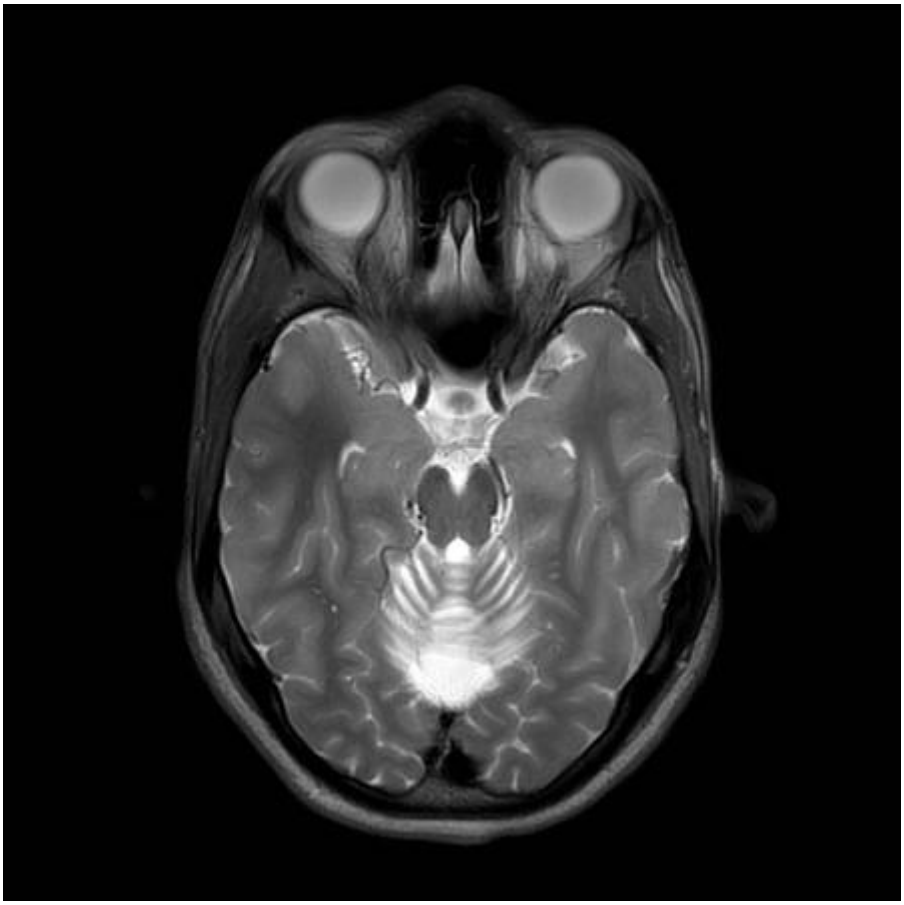
Figure 3

a



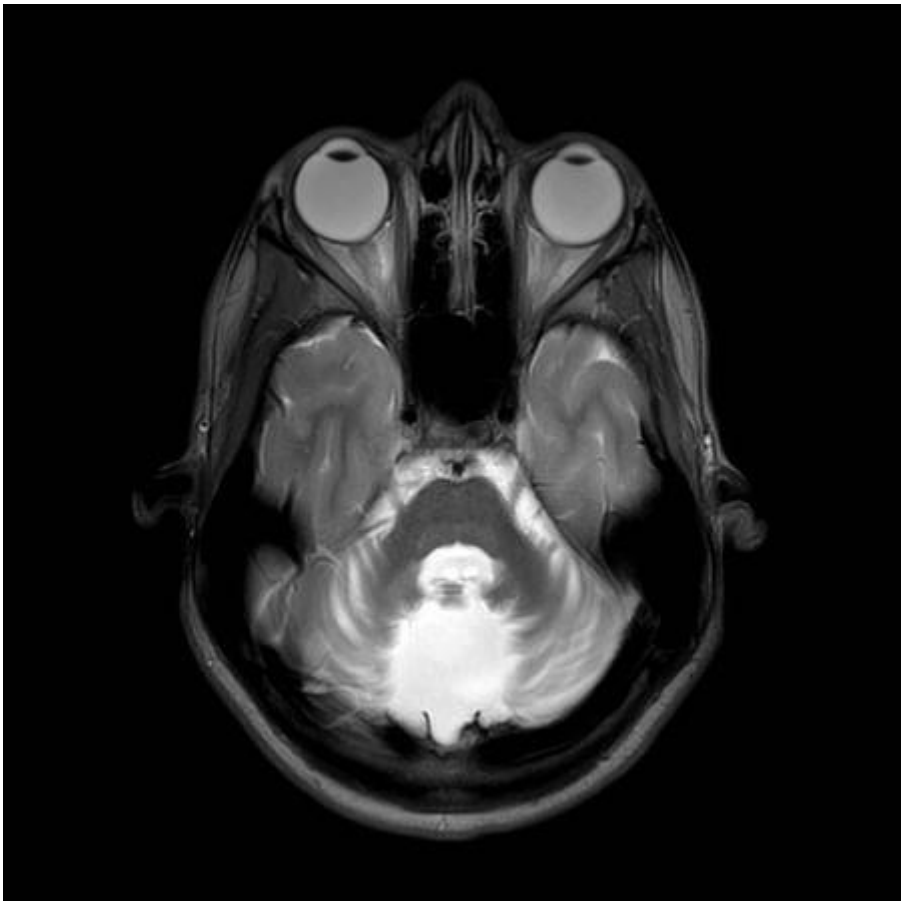
Description: A coronal FLAIR-weighted image showed an increase signal-intensity of the cerebellar parenchyma, when compared with the cerebral parenchyma. **Origin:** Radiologic department of the university hospital of Leuven, Belgium.

b



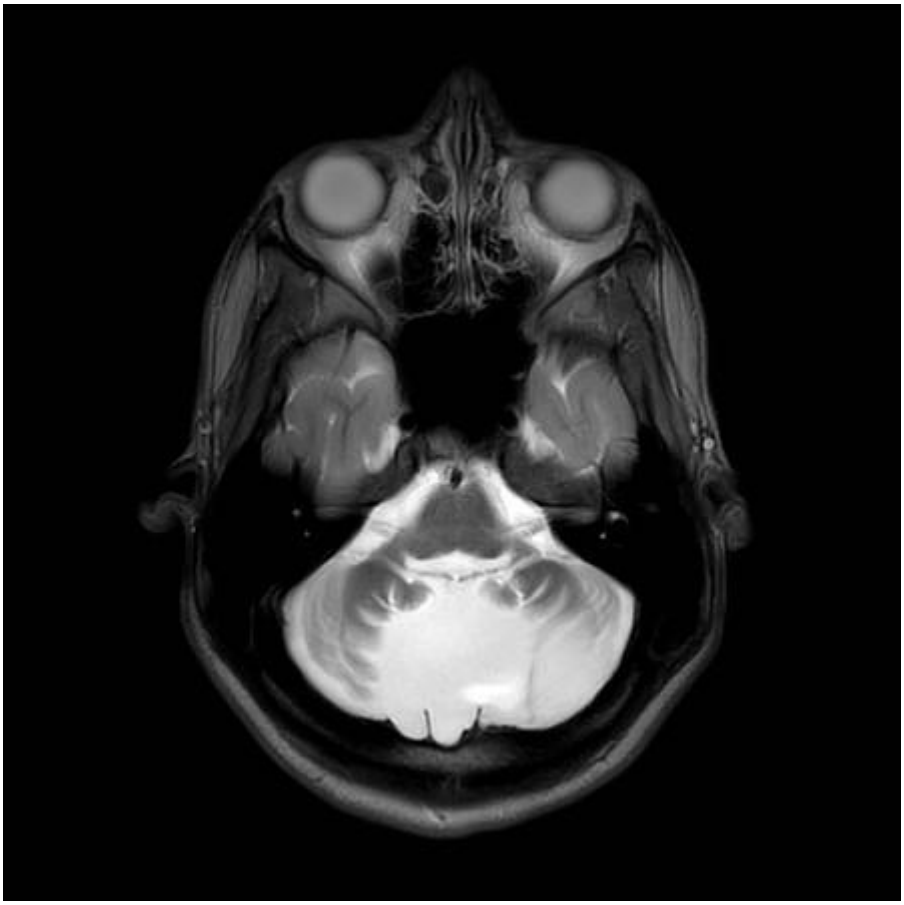
Description: Axial T2-weighted image shows the hypoplastic and atrophic cerebellum, involving the vermis and both cerebellar hemispheres. **Origin:** Radiologic department of the university hospital Leuven, Belgium.

c



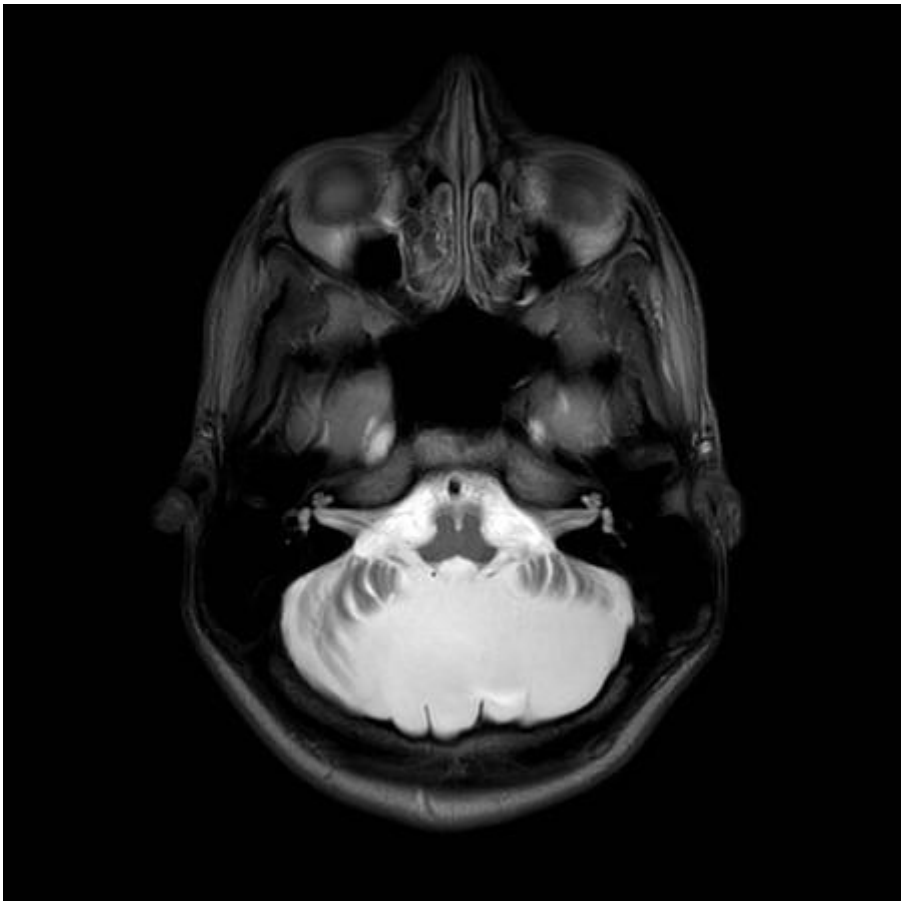
Description: Axial T2-weighted image shows the hypoplastic and atrophic cerebellum, involving the vermis and both cerebellar hemispheres. **Origin:** Radiologic department of the university hospital Leuven, Belgium.

d



Description: Axial T2-weighted images shows the hypoplastic and atrophic cerebellum, involving the vermis and both cerebellar hemispheres. **Origin:** Radiologic department of the university hospital Leuven, Belgium.

e



Description: Axial T2-weighted image shows the hypoplastic and atrophic cerebellum, involving the vermis and both cerebellar hemispheres. **Origin:** Radiologic department of the university hospital Leuven, Belgium.