Case 15544

Pontocerebellar hypoplasia type 1
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Section: Paediatric radiology
Area of Interest: Neuroradiology brain
Procedure: Diagnostic procedure
Imaging Technique: Ultrasound
Imaging Technique: MR
Special Focus: Congenital Case Type: Clinical Cases
Authors: Picado A, Llorens Salvador R
Patient: 1 days, female

Clinical History:

A male newborn presents hypotonia, bradycardia and significant respiratory distress. No particular incidences were documented during pregnancy. There was no family background of congenital disease.

Imaging Findings:

Transfontanellar ultrasound showed a small cerebellum with excessive CSF in the posterior fossa (Figs. 1, 2). MRI was then performed without sedation. Mid-line sagittal T2 weighted sequences (Fig. 3) showed striking cerebellar hypoplasia, with reduced cerebellar volume and vermian biometry, which corresponded to a gestational age of 20-22 weeks. The tegmentovermian angle was also abnormal and a malformative cyst was identified in the posterior fossa. In the coronal plane this abnormal appearance of the cerebellum has been described as a “dragonfly”, with the flattened cerebellar hemispheres representing the “wings” and the vermis the “body” (Fig. 4). Brainstem volume was also decreased, with a mild reduction of pontine volume. Axial T2-weighted MR sequences demonstrated flattening of the cerebellar hemispheres and a small pons and vermis (Fig. 5).

Discussion:

Pontocerebellar hypoplasia (PCH) is a group of neurodegenerative disorders with autosomal recessive inheritance characterised by hypoplasia of the cerebellum and pons with associated progressive microcephaly. Ten PCH subtypes (PCH1-10) have been described. Genes involved in the pathogenesis are essential for protein synthesis and tRNA processing. These genes have been well studied and families in which there are known mutations can be tested prenatally. Prognosis is poor. Most patients will die during infancy or childhood, and treatment during the disease is symptomatic. [1, 2]

PCH type 1 represents the most prevalent subtype, and is characterised by a loss of anterior horn spinal cord neurons similar to that in the spinal muscular atrophies (SMA).

Diagnosis will be based on a combination of radiological, neurological and neuropathological findings. [3]

The radiological findings include hypoplasia and/or atrophy of the cerebellum and pons. The cerebellar vermis and cerebellar hemispheres are equally affected. Involvement of the pons and cerebrum is variable. [1] Supratentorial abnormalities, such as widened extracerebellar CSF spaces and widened lateral ventricles due to small basal ganglia can be seen.

In patients with PCH type 1, MRI shows a variable degree of cerebellar hemisphere involvement. Involvement of the pons and cerebrum can be variable. A dragonfly appearance has been described on coronal MR imaging. [1, 2]

The main radiological differential diagnosis are the congenital disorders of glycosylation type 1A, where the key finding is a progressive cerebellar atrophy, usually without pontine involvement.

Several mutations have been associated with PCH type 1, genes implicated are VRK1, RARS2, and TSEN54, and
more recently the EXOSC3 gene has been defined as a major cause. [4]
It is important to note that this is a hereditary disease with an autosomal recessive pattern of inheritance, so that parents have a 25% risk of having more children with the same disease.
The most important differential diagnosis of PCH1 from the genetic point of view is infantile SMA, which is caused by mutations in the SMN1 gene, and which shares involvement of spinal motor neurons. [5]
Management is symptomatic since there is no cure for the disease. Some of the care provided includes nutritional support (PEG feeding), treatment of seizures, dyskinesia and dystonia. Respiratory support sometimes is necessary. [1]

**Differential Diagnosis List:** Pontocerebellar hypoplasia type 1, Spinal muscular atrophy type 1 (SMA I), Congenital disorders of glycosylation type 1A (CDG1A)

**Final Diagnosis:** Pontocerebellar hypoplasia type 1

**References:**

Namavar Y et al. (2011) Classification, diagnosis and potential mechanisms in Pontocerebellar Hypoplasia. Orphanet Journal of Rare Diseases Vol.6, p 50 (PMID: 21749694)
Description: Sagittal mid-line view of the brain. Globally small cerebellum (blue arrow) and increased CSF in posterior fossa (white arrow). Origin: HUP La Fe, Valencia, Spain
Description: Coronal view of the brain US. Small vermis is identified (blue arrow) and abnormal amount of CSF in the posterior fossa. Origin: HUP La FE
Figure 3

Description: Sagittal MR T2-weighted sequence. Small cerebellum (blue arrow) and pons. Vermian biometry (double head arrows) corresponds to 20 weeks gestational age. Abnormal Tegmentovermian angle (black lines) and a posterior fossa cyst (red arrow) Origin: HUP La Fe
**Description:** Coronal MR T2-weighted sequence. Global decrease of cerebellar volume. This appearance has been described as a “dragonfly” (blue arrow). Increased CSF in the posterior fossa (black arrow). **Origin:** HUP La FE
Description: Axial T2w MRI of the posterior fossa showing the abnormally small pons and cerebellum (blue arrow). Increased CSF in the posterior fossa. Origin: HUP La Fe