Clinical case study

Dandy-Walker variant associated with Down syndrome

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Abstract

We report on a patient with Down syndrome and a multisystemic condition comprising congenital heart disease (atrioventricular canal), facial angioma and neurological malformation (Dandy Walker variant). The nosological status of posterior fossa cyst malformations is reviewed, as the present case is felt to support the utility of the Dandy-Walker complex, a concept which allows grouping of posterior fossa malformations (megacisterna magna, Dandy-Walker syndrome, and Dandy-Walker variant) within the same syndrome.

Keywords: Dandy-Walker complex, Down syndrome, cerebellar/vermian hypoplasia, ataxia

Introduction

Patients with Down syndrome (DS) often have malformations which may involve a number of organs and systems [1]. Frequent congenital malformations include cardiac defects, atresias of the gastro-intestinal tract (duodenal or anal), pulmonary abnormalities (pulmonary hypoplasia), and musculoskeletal and neurological malformations, particularly Moya-Moya syndrome and cerebellar/vermian hypoplasia. We report the case of a patient who has DS in association with a congenital malformation of the posterior fossa that matched a variant of Dandy-Walker syndrome.

Case report

A 28-year-old patient with DS and a history of congenital heart malformation (atrioventricular canal) and idiopathic thrombocytopenia presented with personality changes and a 6-month history of unstable gait with leftward lateropulsion. No headaches or signs of brainstem dysfunction were present, nor was there any associated cognitive deterioration. Neurological examination found bradipsychia with release of primary reflexes and bilateral grasping reflex.

Cranial pairs and motor and sensorial pathways were normal. A venous angioma was found on the patient’s lower lip. Cerebellar changes were not present, but gait was altered, slow and unstable, with no further specific features. A CT head scan showed swelling of the cisterna magna and hypoplasia of the lower cerebellum, which suggested a Dandy-Walker malformation.
MRI confirmed cerebellar hypoplasia associated with a small posterior fossa, which suggests Dandy-Walker variant (figs. 1 and 2). Concomitant hydrocephalus was not found. Given the unspecific symptoms and lack of hydrocephalus, watchful waiting was the option chosen. The patient improved and the process stabilized spontaneously.

Discussion

Cerebellar/vermian hypoplasia is a condition consisting of congenital malformations in association with a normal posterior fossa, varying degrees of hypoplasia of the vermis/cerebellum, and a normal or dilated fourth ventricle unobstructedly connected with a prominent CSF-filled space in the hindbrain [2]. Only a fuzzy line can be drawn between this process and Dandy-Walker syndrome (DWS). The classical definition of DWS rests on a triad: a) partial or total vermian agenesis; b) cystic dilatation of the fourth ventricle; c) an enlarged posterior fossa. This syndrome usually presents before late childhood, typically during infancy. It is often concomitant with hydrocephalus and usually requires surgery. It is also frequently concomitant with other malformations, such as facial angiomas, reported in 10% of patients [3], or cardiovascular abnormalities. As a result, it is believed that such malformations arise at some point after the neural crest cells develop in the early weeks of gestation and before they migrate. Other cases have been described in association with syringomelia [4] or Coffin-Siris syndrome [5]. Patients such as the one presented here, who have vermian hypoplasia associated with a dilated fourth ventricle but no enlargement of the posterior fossa, are categorized under the so-called Dandy-Walker variant. Among the cystic abnormalities of the posterior fossa, at present Dandy-Walker malformation, Dandy-Walker variant, and mega cisterna magna are considered different expressions of the same process. For this reason, they have been grouped for quite some time under a single heading, the Dandy-Walker complex [6].

Few cases have been reported of DS in conjunction with DWS [7], although cerebellar/vermian hypoplasia, which may be considered a DWS variant, is often associated with DS. An interesting study analyzing chromosomal abnormalities in patients with varying degrees of vermian agenesis [8] found...
a greater frequency of such abnormalities, including DS, in cases of inferior vermian agenesis (53%) than with complete vermian agenesis (45%).

The patient’s symptoms – changed personality and unstable gait with left lateropulsion, with no other neurological findings – cannot be linked with certainty to the cerebellar malformation, especially considering the patient’s age, which seems excessive for an onset of symptoms tied to a congenital defect. However, such a link may not be ruled out altogether, as there are cases of adult-onset DWS [9]; furthermore, the patient’s ataxia symptoms correlated well, both clinically and radiologically, with the hypoplastic area of the cerebellum (vermis).

Generally, DWS is treated with surgery, especially when there is concomitant hydrocephalus as well as signs of intracranial hypertension. There are a number of surgical options, ranging from a ventriculoperitoneal shunt to fenestration of the cystic membrane, a primary cystoperitoneal shunt or a ventriculocysto-peritoneal shunt [10]. With Dandy-Walker variant, surgery indications are hydrocephalus and signs of high intracranial pressure.

Finally, another noteworthy point in this case was multiorgan involvement, which extended to the central nervous system (DWS variant), cardiovascular system (atrioventricular canal), and skin (facial angioma). These associations are well documented in patients with DWS [3], so there is a good case for linking them, which may in turn be interpreted as yet another point supporting the nosological similarity of DWS and its variant.

References