Case 17212

Eurorad

Dandy Walker Malformation

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Section: Paediatric radiology
Area of Interest: Neuroradiology brain Paediatric
Imaging Technique: CT
Imaging Technique: MR
Case Type: Clinical Cases
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Patient: Term baby, male

Clinical History:

Term baby with antenatally diagnosed schizencephaly. On postnatal ultrasound, extensive schizencephaly saw with dilatation of lateral and fourth ventricles. Patient was seen to suffer from hyperventilation and experienced seizures.

Imaging Findings:

MRI brain showed enlargement of the posterior fossa with torcular-lambdoid inversion and wide communication between the fourth ventricle and the posterior subarachnoid enlarged posterior fossa (Figure 1). Dysgenesis of the cerebellar vermis was seen. A wide CSF cleft lined by grey matter, which is communicating the medial aspect of the left lateral ventricle with the superomedial subarachnoid space, is seen as a dorsal cyst. There is also dysgenesis of the corpus callosum, the body and splenium cannot be identified (Figure 2). In addition, atresia of the cerebral aqueduct can also be seen. MR Renal was also performed and showed nephronophthisis. MR brain was repeated. Interval ballooning of the tentorium cerebelli upwards into the interhemispheric space was seen. A ventriculoperitoneal (VP) shunt was inserted. The size of the supratentorial ventricles and extra-axial spaces otherwise remained stable. Patient was followed up with brain CT to monitor positioning of VP shunt (Figure 3).

Discussion:

Background

Dandy-Walker malformation (DWM) is a rare intracranial congenital abnormality that affects the cerebellum and some of its components; particularly cerebellar vermis, fourth ventricle and is characterized by an enlarged posterior fossa [1]. In DWM, the fourth ventricle opens into and is continuous with almost the entire posterior fossa
DWM occurs as an autosomal dominant inherited disorder and occurs in one in 25000 – 35000 pregnancies [2]. The most frequent and prominent symptoms of DWM are those associated with hydrocephalus in the postnatal period. Hydrocephalus occurs in an estimated 80% of patients with classic DWM. This usually presents within the first year of life most often within the first 3 months [3].

Clinical perspective

The clinical presentation is nonspecific, subject to multiple factors including the severity of hydrocephalus, intracranial hypertension, and associated comorbidities. Most patients are in their first year of life, with signs and symptoms of increased intracranial pressure [2]. The most common manifestation is macrocephaly, affecting 90 to 100% of patients during their first months of life [4]. The syndromic form of DMW may also have malformations of the heart, face, limbs and gastrointestinal or genitourinary systems that could draw the initial medical attention [5]. Furthermore, apart from medical assessments, diagnostic imaging techniques such as ultrasound and MRI are useful to diagnose this syndrome.

Imaging perspective

MRI brain revealed dysgenesis of the cerebellar vermis and corpus callosum, with an enlarged posterior fossa and right lateral ventricle. These were the main findings that indicated Dandy-Walker malformation. Furthermore, the association of renal cystic diseases (nephronophthisis) seen on both US abdomen and MR renal supported a diagnosis of ciliopathic genetic condition such as Joubert Syndrome. However, other genetic conditions such as Goldston syndrome cannot be excluded. The diagnosis of DWM is based on the presence of MRI findings described above. Associated syndromes such as Joubert and Goldston can be diagnosed using genetic counselling.

Since hydrocephalus was present in this case of DWM, as it commonly is, a VP shunt was needed to drain the excess CSF present and hence reduce the intracranial pressure and improve symptoms. Imaging was important to see that the VP shunt was positioned in situ and hence, non-enhanced CT (NECT) was performed. Other management would include occupational therapy and physiotherapy.

Take-home message/Teaching points

Timely prenatal and antenatal scans are key to diagnose DWM so that the appropriate treatment can be administered.

Differential Diagnosis List: Dandy-Walker Malformation , Schizencephaly, Joubert syndrome, Goldston syndrome

Final Diagnosis: Dandy-Walker Malformation

References:

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Figure 1

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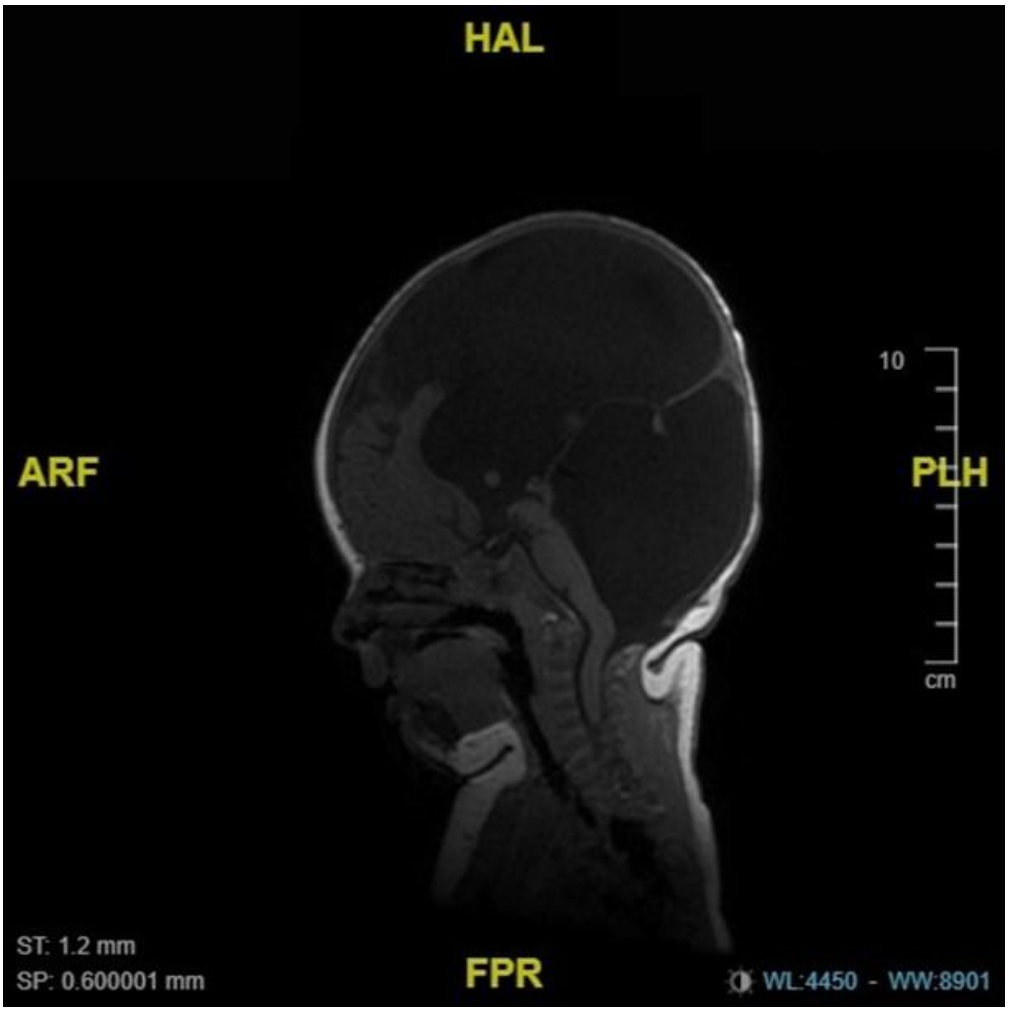

Description: Sagittal T1W MR image showing enlargement of the posterior fossa with torcular-lambdoid inversion, and wide communication between the fourth ventricle and the posterior subarachnoid enlarged posterior fossa. **Origin:** Medical Imaging Department at Mater Dei Hospital, Malta.

Figure 2

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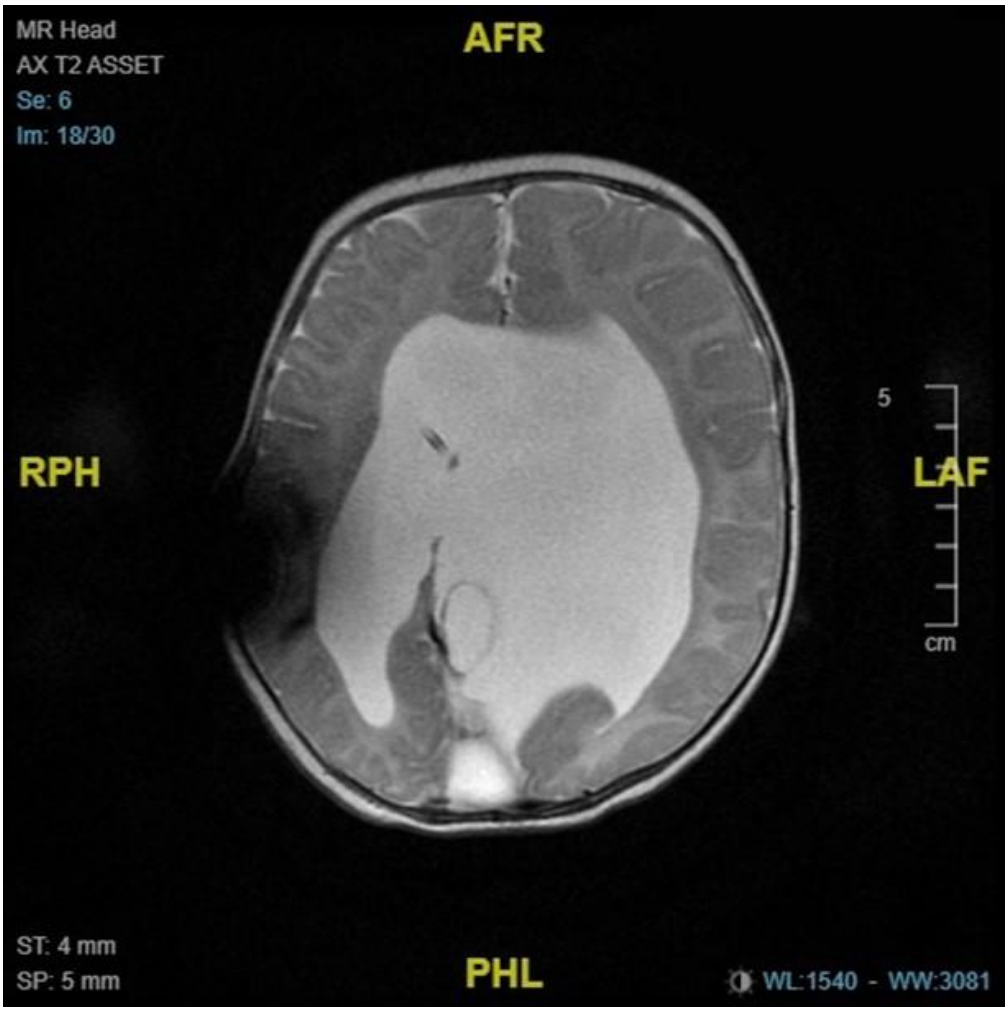

Description: Axial T2W MR image showing Dysgenesis of the cerebellar vermis and corpus callosum, a wide CSF cleft lined by grey matter, communicating with the medial aspect of the left lateral ventricle with the supero-medial subarachnoid space. **Origin:** Medical Imaging Department at Mater Dei Hospital, Malta.

Figure 3

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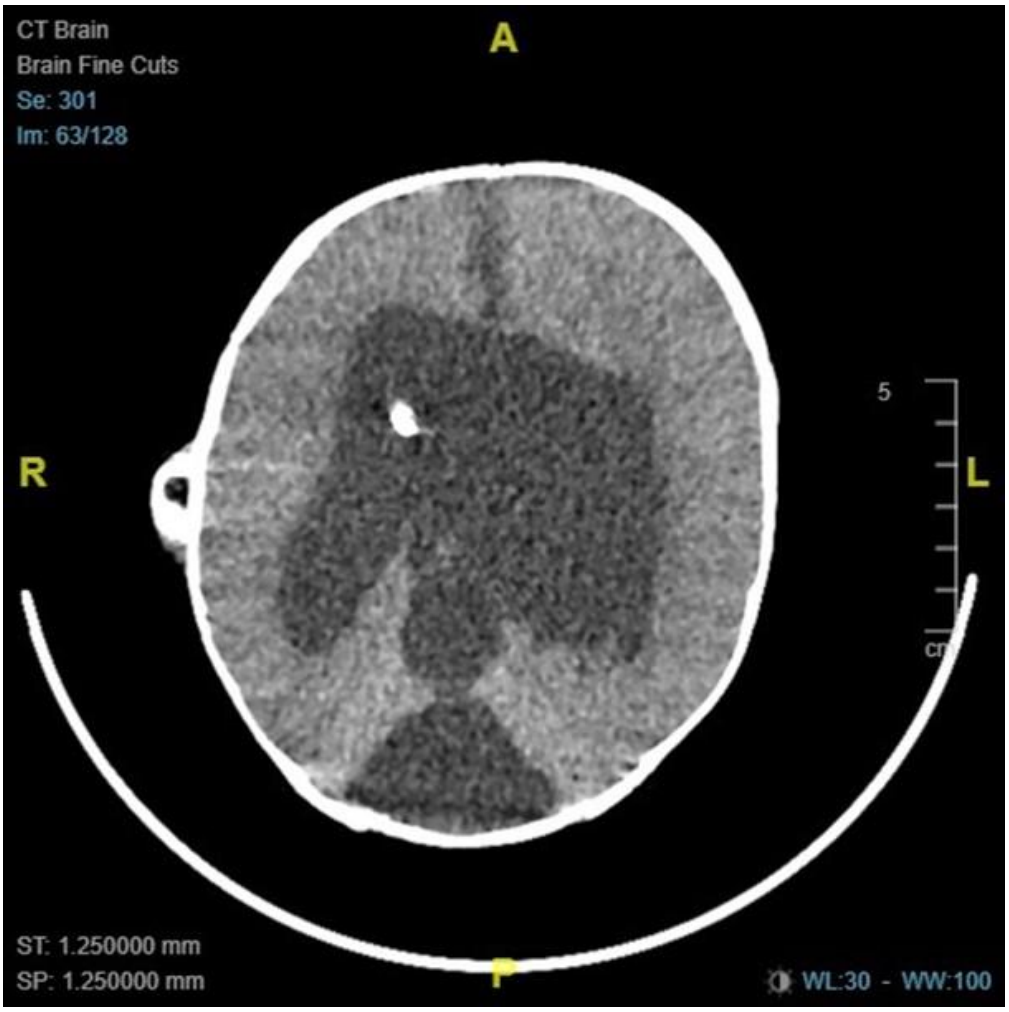

Description: Axial CT image showing positioning of the VP shunt. **Origin:** Medical Imaging Department at Mater Dei Hospital, Malta.