

Dandy Walker Malformation A Rare Case with Review of Literature. Embryological and Clinical Perspective

Dr Ashfaq Ul Hassan, Dr Zahida rasool, Dr Shifan Khandey, Dr Riyaz Malik, Dr Sajid Shafi

MS Lecturer Anatomy SKIMS Medical College Bemina
ashhassan@rediffmail.com

Medical Consultant IUST
Zahida rasool12@rediffmail.com

MS Tutor/Demonstrator Anatomy SKIMS Medical College Bemina

Assistant Professor Pediatrics SKIMS Medical College Bemina

Tutor Microbiology SKIMS Medical College Bemina
sajidshafi@yahoo.com

Abstract:

We present a case of a 12 month old female baby who was brought to a pediatric clinic with increasing head size and delayed milestones in her development. She did not have any history of trauma or any significant maternal medication or infections acquired during pregnancy. She was born full term and a result of non consanguineous marriage. There were no other physical abnormalities except the increased head size and delayed milestones.

Key Words:

Cerebellum, Hypoplasia, ataxia, transillumination, cleft

Introduction:

Dandy Walker Malformation is a rare clinical entity usually detected incidentally in the first few months of life. Early recognition of this syndrome is of prime importance as children with this abnormality have severe developmental and intellectual problems.

Discussion

The malformation was first described by Sutton in 1887.¹ It belongs to the group of disorders related to abnormal development of hind brain and posterior cranial fossa but can also be associated with other defects of nervous system. The Development of nervous system begins at 17 days of gestation when the neural tube begins to

form which is subsequently followed by development of three brain vesicles namely the Prosencephalon, mesencephalon and rhombencephalon. At the same time cavities form within these three vesicles.

The Rhombencephalon is concerned with the development of hind brain. The Cerebellar vermis begins to develop in the ninth week of gestation. There is disproportionate growth between cerebrum and cerebellum and the rate of growth of cerebellar hemispheres lags behind the cerebral hemispheres. The primary mechanism is excessive increase in production of CSF leading to hydrocephalus with dilatation of ventricles and consequent hypoplasia of cerebellar vermis. The association between hydrocephalus and Dandy Walker Malformation is of immense significance as the severity of hydrocephalus is related to the agenesis of cerebellar vermis. Congenital

anomalies of the posterior fossa, including the Dandy-Walker syndrome, the Chiari malformation, and encephalocele, are prominently associated with ataxia because of their destruction or replacement of the cerebellum. The Dandy-Walker malformation comprises of a cystic expansion of the fourth ventricle in the posterior cranial fossa, which results from a developmental failure of the roof of the fourth ventricle during embryogenesis. **2**

The condition is more common in females.

Approximately 90% of patients have hydrocephalus, and a significant number of children have associated anomalies, including agenesis of the posterior cerebellar vermis and corpus callosum. In this clinical syndrome Infants usually present with a rapid increase in head size and a prominent occiput. Transillumination test of the of the skull may be positive. Most have a prominent cerebellar ataxia, and delayed motor and cognitive milestones, probably owing to the associated structural anomalies. It may be associated with cleft palate, high arched palate, clinodactyly, hypertelorism, myopia, brachycephaly, lobulated ears. Fortunately our case did not have any of these associations. **3** The intelligence quotient is usually reduced with Most children having moderate to severe abnormalities in memory functions, Visual problems like strabismus, visuospatial abnormalities, visual field defects, and optic atrophy with decreased acuity secondary to increased intracranial pressure.

Usually the investigation of a child with hydrocephalus begins with the history. Familial cases suggest X-linked hydrocephalus secondary to aqueductal stenosis. A past history of prematurity with intracranial hemorrhage, meningitis, or mumps encephalitis is important to ascertain. Transillumination of the skull may be positive with massive dilatation of the ventricular system or in the Dandy-Walker syndrome. Inspection of the eyes is mandatory because the finding of chorioretinitis suggests an intrauterine infection such as toxoplasmosis as a cause of the hydrocephalus. Papilledema is observed in older children but is rarely present in infants because the cranial sutures separate as a result of the

increased pressure. Plain skull films typically show separation of the sutures, erosion of the posterior clinoids in the older child, and an increase in convolutional markings with longstanding increased intracranial pressure.

The CT scan and/or MRI along with ultrasound in the infant are the most important studies to identify the specific cause of hydrocephalus. The MRI Scans done suggest a large Posterior Cranial fossa with Cystic enlargement, hypoplasia of cerebellar hemispheres and vermian hypoplasia in Particular. It has been proven beyond doubt that MRI is the diagnostic modality of choice in detecting Dandy Walker Malformation.

Therapy for hydrocephalus depends on the cause. Medical management, including the use of acetazolamide and furosemide, may provide temporary relief by reducing the rate of CSF production, but long-term results have been disappointing. Most cases of hydrocephalus require extracranial shunts, particularly a ventriculoperitoneal shunt. **4, 5, 6**

The major complication of shunts is bacterial infection, usually due to Staphylococcus epidermidis. Prognosis depends on the cause of the dilated ventricles. Hydrocephalic children are at increased risk for a variety of developmental disabilities. In view of this fact Early recognition of this condition and effective means to control the development of hydrocephalus are the Primary modalities in treatment strategy.



Fig: A female baby with Cystic Posterior Cranial Fossa



Fig: A female baby with Cystic Posterior Cranial Fossa and Hypoplasia of vermis



Fig: A female baby with Cystic Posterior Cranial Fossa

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