Case Report

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Dandy Walker Syndrome

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Abstract

Dandy-Walker syndrome is characterized by a triad of complete or partial agenesis of the cerebellar vermis, cystic dilatation of the forth ventricle and enlarged posterior fossa with upward displacement of the transvers sinus, tentorium and torcular. The most striking abnormality is the presence of a huge dilated forth ventricle which acts as a cyst and is roofed by a neuroglial-vascular membrane lined with epandyma. This cyst herniates caudally and separates the cerebellar anteriorly and choroid plexus are rudimentary the formation of the fourth ventricle are often occluded by membranes or are atretic. A varient form, in which is cystic dilatation of the fourth ventricle and hypoplasia of the cerebellar vermis without enlargement of the posterior fossa is more common than the classic Dandy-Walker malformation and account for one-third of posterior fossa malformations. Hydrocephalus is precent in 90% of patients. Some of the patients have other malformations associated with this syndrome include, Occipital Encephalocel, facial angioma, midline cleft palate, cardiovascular malformations and polycystic kidney. Hydrocephalus is not present at birth. It often appears by 3 months of age. In some instance, hydrocephalus fails to develop and this condition remains Asymptomatic throughout life. Prenatal diagnosis with ultrasonography and Postnatal diagnosis with CT scan and MRI in progressive hydrocephalus Treatment is surgical. Befor birth in 10 embrio, dandy walker syndrome was diagnosed with ultrasound, that after delivery, diagnosed and confirmed in eight cases. The case was male and 2nd product of non consanguinous marriage.This study is a case report of Dandy-Walker syndrome in a newborn.

Keywords: Dandy-Walker syndrome; Hydrocephalus; Vermis Aplasia.

Introduction

Dandy–Walker syndrome (DWS), or Dandy–Walker complex, is a congenital brain malformation involving the cerebellum and the fluid filled spaces around it. A key feature of this syndrome is the partial or even complete absence of the part of the brain located between the two cerebellar hemispheres (cerebellar vermis).[1] The Dandy–Walker complex is a genetically sporadic disorder that occurs one in every 30,000 live births.[2] Prenatal diagnosis and prognosis of outcomes associated with Dandy-Walker can be difficult.[3] It is named for Walter Dandy and Arthur Earl Walker.[4,5] This case is on male baby diagnosed having Dandy walker syndrome and focuses on nomenclature, the consecutive classification, and steps for diagnosis. Furthermore, we discuss the approach to the family for comprehensive counselling in decision making for their child.

Case summary

2 month 11 days old male baby was admitted in our department with birth weight (birth weight-2.76 kgs) with complaints of repeated vommiting after feed, irritability. He is 36 weeks 9 day by gestational age delivered by vaginal route in civil hospital. Mother is 23 yr old not registered case ,no antenatal USG

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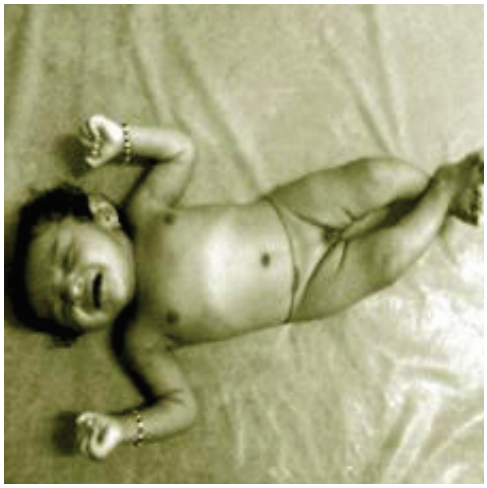
a) Baby showing crossed over legs With hypertonia



done. Baby cried immediately after birth, cry was good and birth weight is 2760 gms. Our case is 2th issue of non consanguinous marriage of his parents. First is male child 2 yrs old without any similar history or any congenital anomoly.

On examination, his pulse was 120 beats/ minute & respiratory rate of 32 cycles/min. Anthropometric measurements shows weight -2.96 kg , length - 49 cms, Head circumference – 42 cms, Chest circumference-36 cms

His investigations were as follows:

Hb: 11.3 gm%

TLC: 7500/cmm

P – 55, L – 39, E – 02, M – 03, B – 01.

Platelet count: 2.49 lacs/cmm.

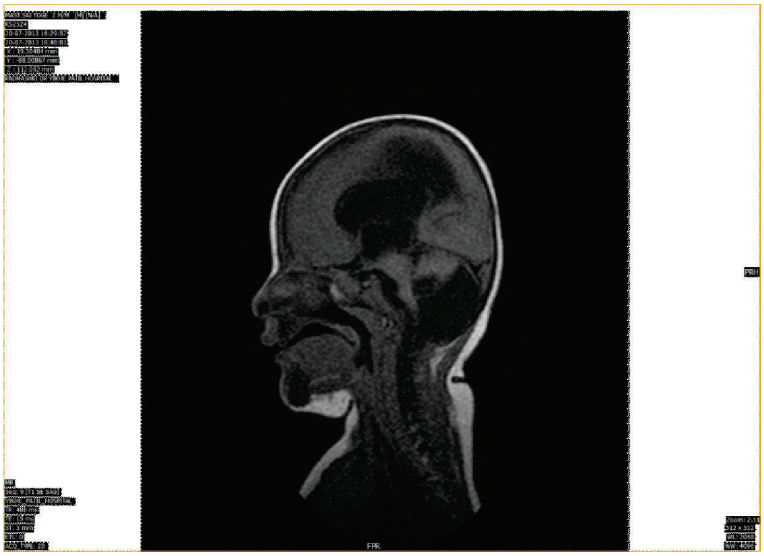
Chest x-ray is normal

MRI brain shows complete or partial agenesis of the vermis, cystic dilatation of the fourth ventricle, An enlarged posterior fossa with upward displacement of lateral sinuses, tentorium, and torcular herophili.

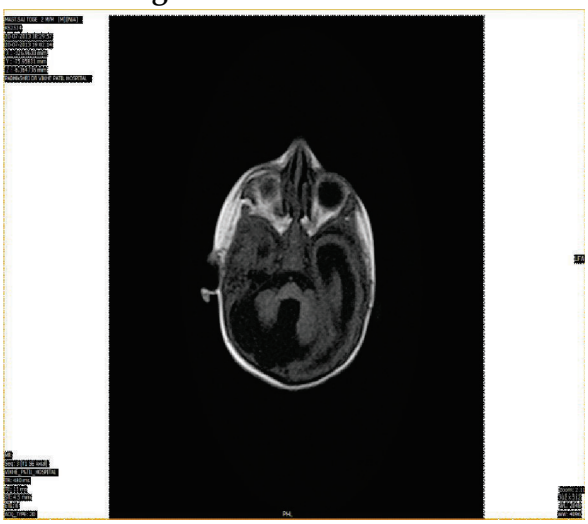
Discussion

Dandy-Walker malformation is a rare congenital brain malformation characterised by hypoplasia and upward rotation of the cerebellar vermis, cystic dilation of the fourth ventricle and enlargement of the posterior fossa.[6] It may also form part of the PHACES syndrome (= **P** osterior fossa anomalies as Dandy-Walker malformation; **H** aemangioma; **A** rterial lesions of the head and neck; **C** ardiac abnormalities as aortic coarctation; **E** ye abnormalities and **S** ternal defect).[7] A large number of associated problems may also be present, such as hydrocephalus (often develops postnatally), atresia of the foramen of Magendie and atresia of the foramen of Luschka. Incidence is approximately 1 in 25-

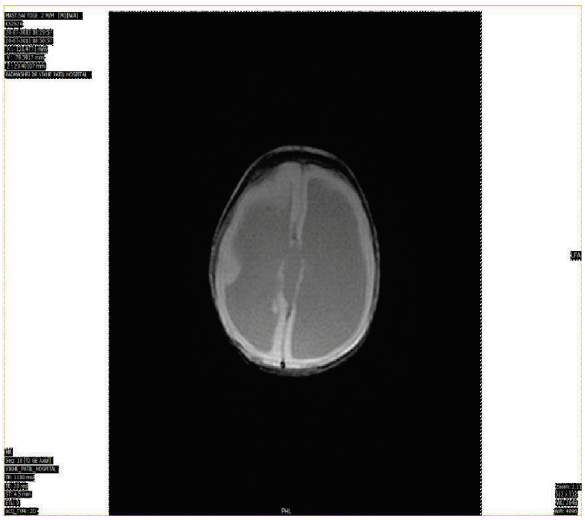
b) MRI showing enlarged posterior fossa with upward displacement of lateral sinuses, dilatation of third and forth vetricle



c) MRI brain shows Complete or partial agenesis of the vermis



d) MRI brain showing cortical atropy



35,000 live births.[8] It occurs slightly more frequently in females than in males.Marked variation has been shown in the genetics and aetiology.[9] Recurrence risk for siblings may be high when there is an association with a single gene disorder.Dandy-Walker syndrome may result from chromosomal anomalies or environmental factors.[10] Associated environmental factors include first trimester exposure to rubella, cytomegalovirus, toxoplasmosis or warfarin. When the evidence suggests that there is no association with a Mendelian or chromosomal disorder then the recurrence risk is relatively low at between 1 and 5%.[11]

Presentation

- The syndrome can appear dramatically or develop unnoticed.
- Symptoms often occur in early infancy and include slow motor development and abnormally rapid increase in head circumference with bulging at the back of the skull.
- In older children there may be signs of increased intracranial pressure, such as irritability, vomiting, and convulsions. There may also be signs of cerebellar dysfunction, such as unsteadiness, lack of muscle co-ordination, or jerky movements of the eyes. One case report describes conduct disorder, hyperactivity, stereotypical movements and nocturnal enuresis in a 14 year-old.[12]
- There may also be jerky movements of the face and neck and abnormal breathing patterns.

Investigations

- The diagnosis of Dandy-Walker malformation can be made by antenatal ultrasound.[13]
- MRI scan allows a detailed evaluation of Dandy-Walker malformation lesions and complications. MRI evaluation can then be used antenatally to confirm the diagnosis and to gain more detailed information.

Associated diseases

Dandy-Walker syndrome is frequently associated with disorders of other areas of the central nervous system, including absence of the corpus callosum, and malformations of the heart, face, limbs, fingers and toes.

Management

- Treatment involves managing the associated problems, eg seizure

management.

- A shunt to treat associated hydrocephalus can be inserted. This may be to shunt the cyst (cystoperitoneal), the ventricles (ventriculoperitoneal), or both.
- Parents of children with Dandy Walker syndrome will benefit from genetic counselling if they intend to have more children.

Prognosis

- Children with Dandy-Walker syndrome may never have normal intellectual development, even when the hydrocephalus is treated early.
- Prognosis otherwise depends on the severity of the syndrome and associated malformations.
- The presence of multiple congenital defects may shorten lifespan.

References

1. National Institute of Neurological Disorders and Stroke. NINDS Dandy–Walker Syndrome Information Page. http://www.ninds.nih.gov/disorders/dandywalker/dandywalker.htm. Last updated September 16, 2008. Last accessed July 6, 2009.
2. Osenbach RK, Menezes AH. Diagnosis and management of the Dandy-Walker malformation: 30 years of experience. *Pediatr Neurosurg.* 1992; 18(4): 179–89. PMID 1472430.
3. Guibaud L, Larroque A, Ville D, Sanlaville D, Till M, Gaucherand P *et al*. Prenatal diagnosis of 'isolated' Dandy-Walker malformation: imaging findings and prenatal counselling. *Prenat Diagn.* 2012; 32(2): 185–93. doi:10.1002/pd.3828. PMID 22418964.
4. Incesu L *et al*. Imaging in Dandy-Walker Malformation. *eMedicine.* 2009.
5. Lopez Gutierrez JC. PHACES syndrome and ectopia cordis. *Interact Cardiovasc Thorac Surg*. 12(4):642-4 2011.
6. Lavanya T, Cohen M, Gandhi SV, *et al.* A case of a Dandy-Walker variant: the importance of a multidisciplinary team. *Br J Radiol*. 2008; 81(970): e242-5.
7. Dandy-Walker Syndrome. Online Mendelian Inheritance in Man (OMIM).
8. Imataka G, Yamanouchi H, Arisaka O. Dandy-Walker syndrome and chromosomal abnormalities. *Congenit Anom (Kyoto)*. 2007; 47(4): 113-8.
9. Murray JC, Johnson JA, Bird TD. Dandy-Walker malformation: etiologic heterogeneity and empiric recurrence risks. *Clin Genet*. 1985 ; 28(4): 272-83.
10. Prakash R, Singh LK, Kour J, *et al.* Psychiatric comorbidities in Dandy-Walker variant disorder. *J Neuropsychiatry Clin Neurosci*. 2009; 21(4): 477-9.
11. Has R, Ermis H, Yuksel A, *et al.* Dandy-Walker malformation: a review of 78 cases diagnosed by prenatal sonography. *Fetal Diagn Ther*. 2004; 19(4): 342-7.
12. Klein O, Pierre-Kahn A, Boddaert N, *et al.* Dandy-Walker malformation: prenatal diagnosis and prognosis. *Childs Nerv Syst*. 2003; 19(7-8): 484-9. Epub 2003 Jul 16.
13. Zimmerman RA, Bilaniuk LT. Magnetic resonance evaluation of fetal ventriculomegaly-associated congenital malformations and lesions. *Semin Fetal Neonatal Med*. 2005; 10(5): 429-43.