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Dandy walker syndrome- a CMF involving brain -case report

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Abstract

The Dandy Walker syndrome comprises of congenital brain mal formation involving the cerebellum and the fluid filled spaces around it. It is a disorder of ventral induction resulting in hindbrain abnormalities incorporating varying degrees of cerebellar vermis and/or enlargement of cisterna magna. A case was reported during routine fetal autopsy done in the Department of Anatomy at Government Medical College & Hospital, Chandigarh. It was a male fetus of 19^{+6} wks. The indication for MTP was Dandy Walker Syndrome detected on ultra sonogram at 27 weeks. In autopsy, there was hypoplasia of cerebellum making floor of fourth ventricle visible and also, right side cerebellar hemisphere was smaller than left. The syndrome is associated with fetal karyotype abnormalities, so amniocentesis can be offered on prenatal diagnosis.

Key words: Central nervous system (CNS), Dandy walker malformation (DWM), Dandy walker syndrome (DWS), medical termination of pregnancy (MTP)

Introduction

Hydrocephalus is found in 70-90% of patients, and in several cases DWM is a part of a syndromic condition variably associated with heart, face and ocular abnormalities .[1,2] The Dandy Walker syndrome comprises of congenital brain mal formation involving the cerebellum and the fluid filled spaces around it. It is a disorder of ventral induction resulting in hindbrain abnormalities incorporating varying degrees of cerebellar vermis and/or enlargement of cisterna magna .[3]Cerebellar anomalies are usually associated with chromosomal anomalies .[4] Dandy walker malformation represents one of the commonest congenital defects of cerebellar development, and a frequent cause of termination of fetus diagnosed prenatally .[5]

Case report

A case is reported during routine fetal autopsy done in the Department of Anatomy. It is a male fetus of 28 wks. The indication for MTP was Dandy Walker Syndrome on ultra sonogram.

The Mother of the fetus is 22 yrs old, primi gravida. Medical history of mother and father were normal. Past history and family history of parents were not suggestive of any etiological factors responsible for the defect. Autopsy performed as per routine procedure.

Observations

External examination showed large head, club foot on both side with over-riding of 4th toe on right side and 2nd toe on left side. On internal examination, after opening cranial cavity, Cerebrum was found to have pachygyria. There was hypoplasia of cerebellum making floor of fourth ventricle visible and also, right side cerebellar hemisphere was smaller than left. Posterior cranial fossa appeared larger which could be because of smaller size of cerebellum. Cerebrum was measuring 64.93x44.52x24.72 mm, cerebellum was measuring 15.88x11.30x7.47 mm.

Discussion

The Dandy–Walker complex is a genetically sporadic disorder that occurs one in every 30,000 live births. Genetic basis of DWM is complex and involved genes are largely unknown. Identification of 7 DWM patients with overlapping deletions at 3q first implicated the Z1C1 and Z1C4 genes as causative of the malformation .[6]Two cases of Trisomy 21 and an abnormality of chromosomes 5 amongst 40 patients with DWS have been reported .[7]Trisomy 18, 13, and partial Trisomy of chromosome 11q have also been associated with DWS. [4,7,8] A case is presented of a live born male with Dandy Walker malformation, agenesis of the corpus callosum, and Peter anomaly of the right eye who was exposed to Warfarin between the 8th and 12th weeks of gestation .[9] The DWS has been seen in siblings[10,11] but is rare in identical twins.[12,13]

Most of the patients with Dandy Walker syndrome, signs and symptoms caused by abnormal brain development appear within first year of life with hydrocephalus that causes macrocephaly . [14]The 2 essential features of the DW syndrome are hypoplasia or aplasia of the cerebellar vermis and cystic dilatation of the 4th ventricle. It was noted that measurements in our case (cerebrum -64.93x44.52x24.72 mm and

cerebellum -15.88x11.30x7.47 mm) was less than measurements in normal fetuses of same age, that was 69.16x45.67x24.05mm in cerebrum and 17.25x14.91x9.88 mm in cerebellum. The other 4 abnormalities include elevation of the tentorium cerebelli and lateral transverse sinuses and torcula; lack of patency of foramina of Megendie and Luschka, enlargement of the posterior fossa and hydrocephalus .[15]People with this syndrome frequently experience muscle stiffness and paralysis of lower limbs and they may also have seizures. The posterior cranial fossa is enlarged and tentorium is in high position. The diagnosis depends on radiological investigations and three main features of DWS which are bilateral cerebellar hypoplasia or agenesis, posterior fossa cyst, hydrocephalus .[14]

Extra cranial malformations are also very commonly associated with DWS.Among those most frequently noted are facial hemangiomas, cardiovascular defects, and digital anomalies .[3]Hypertelorism,flat nose, broad root, low set deformed ears,microstomia ,palmar flexion of hand, dorsal flexion of phalanges ,pes calcaneovalgus , were also reported. [16]Association of DWS with congenital absence of spleen is also been reported . [16,17]In a study, fusion between spleen & pancreatic tissue was found in one case of DWS . [16]There were 3 cases of DWM variant with cystic dysplastic kidney & hepatic fibrosis, out of which absence of spleen was noted in one case. [18] Poor fetal outcome is related to extra CNS anomalies found relationship to be direct.[4,19]

The lesion has been regarded by some as a dorsal neural tube defect similar to the Arnold-Chiari malformation. Others have considered it a true dysraphic disorder because of the relatively high (17%) incidence of coincident agenesis of the corpus callosum.[20-22] 4 out of 10 cases confirmed frequency of corpus callosum dysgenesis in association with DWM. [23]A case of DWS was reported in identical twins, in association with Neurofibromatosis type 1 (NF 1) .[15] Association between dandy walker malformation and congenital diaphragmatic hernia was found on autopsy in a stillborn fetus.[3]

1st clinical description of Dandy Walker syndrome (DWS) was published by Dandy and Blackfan in 1914.[15]The pathogenic theory of atresia at the foramina of Luschka and Megendie was introduced at that time and was later elaborated by

Taggart and Walker. A second theory was proposed by Benda who believed that the syndrome represented maldevelopment in region of fourth ventricle but not limited to foramina .[14]Embryologically DWM is believed to result from an insult to alar plate at an early stage of organ development and growth.[24]Cerebellar vermis forms in 9th gestational week from midline fusion of developing cerebellar hemisphere, beginning superiorly and continuing inferiorly ,until entire vermis is closed by end of 15th week . [25]

The Dandy-Walker complex is a rare congenital intracranial malformation that comprises a spectrum of abnormalities of the posterior fossa which are classified as (a) Dandy-Walker malformation (cystic dilatation of the 4th ventricle, complete or partial agenesis of the cerebellar vermis and an enlarged posterior fossa) (b) Dandy-Walker variant (cystic posterior fossa mass with variable hypoplasia of the cerebellar vermis and no enlargement of the posterior fossa) and (c) Mega cisterna magna (enlarged cisterna magna with normal cerebellar vermis and fourth ventricle).[26]An adult onset presentation of DWS in siblings was reported recently. [10]Prognosis is poor for diagnosis made before 21 weeks [27]and markedly better for postnatal diagnosis.[7]

This defect is diagnosed by USG prenatally and MTP is advised. Treatment for individuals born with DWS generally consists of treating the associated problems, if needed. Shunt operation is done to reduce intracranial pressure. Endoscopic third ventriculostomy is also an option. Treatment may also consist of various therapies such as occupational therapy, physiotherapy, speech therapy or specialized education.[14]

Conclusion

The reported case of dandy walker malformation was associated with club foot of both side with overriding of 4th toe on right side and 2nd toe on left side .DWS is not limited to a mechanical disturbance of CSF circulation but rather represent a more generalized disorder of neural development. Prenatal diagnosis is possible with ultrasound and amniocentesis. The defect represented here resembled as variety (a), consisting of Dandy Walker Malformation.

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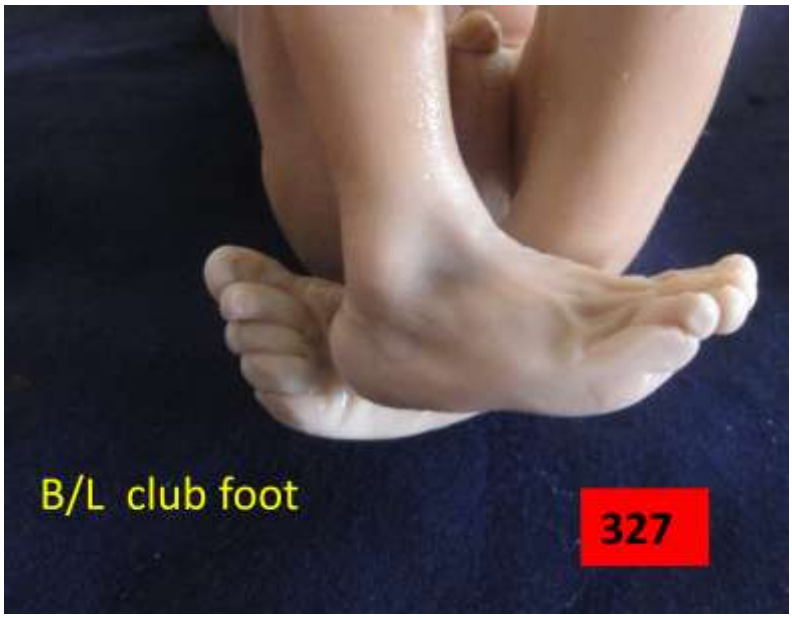
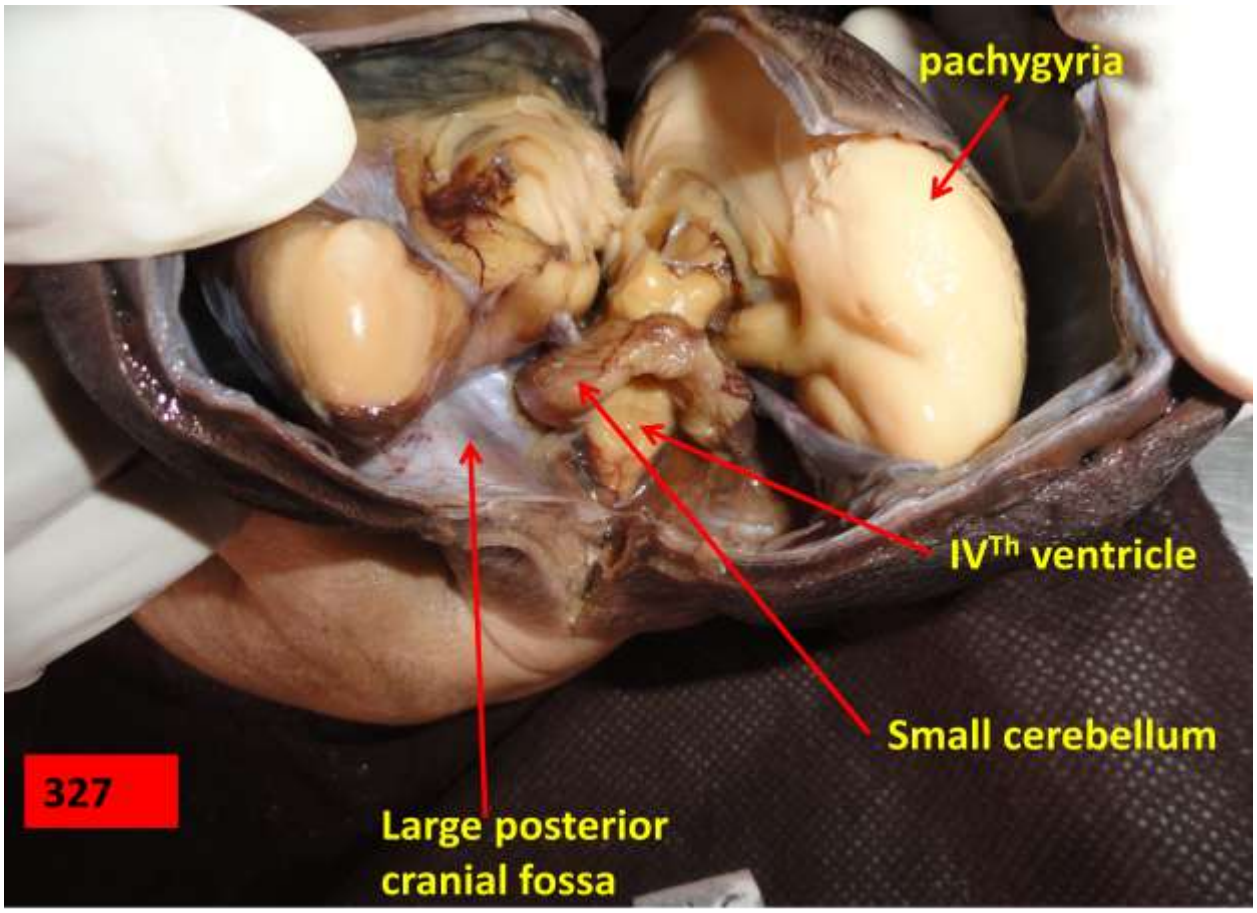
Figures

Fig no 1

Fig no 2





Captions to figures

Fig no 1- External examination showed large head, club foot on both side

Fig no 2- club foot on both side with over-riding of 4^{th} toe on right side and 2^{nd} toe on left side

Fig no 3- Cerebrum was found to have pachygyria .Hypoplasia of cerebellum with smaller right side cerebellar hemisphere than left. Posterior cranial fossa appeared large.

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