DANDY WALKER MALFORMATION WITH UNUSUAL FINDINGS IN FETAL AUTOPSIES

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ABSTRACT

The Dandy-Walker malformation is a rare group of congenital abnormalities of brain, typically involving the fourth ventricle and the cerebellum. The Dandy-Walker malformation has an estimated prevalence of about 1 in 25,000 to 35000 live births with a slight female preponderance and is responsible for 4-12% of infantile hydrocephalus. Dandy-Walker malformation is frequently associated with other intracranial anomalies such as agenesis of the corpus callosum, holoprosencephaly, occipital encephaloceles and ocular abnormalities. Extra-cranial anomalies include polycystic kidneys, cardiovascular defects, polydactyly and cleft palate. Present study was conducted on 1200 fetuses obtained from department of Obstetrics and Gynecology, Government medical college and hospital, Chandigarh. Autopsy was done in department of Anatomy, GMCH Chandigarh. In our present study the incidence of Dandy walker syndrome was 0.4% (out of 1200 fetuses DWS was noted in 5 fetuses). Associated anomalies were noted in each case. Prenatal diagnosis of DWS is important to detect and avoid further complications in life.

INTRODUCTION

Dandy–Walker malformation is a rare congenital abnormality of brain, characterized by hypoplasia or absence of cerebellar vermis, cystic dilatation of fourth ventricle and hydrocephalus 1. Dandy-Walker malformations (DWM) accounts for about 1 in 25,000 to 35000 live births2. Sutton was the first one to described DWM, which was further described by Dandy and Black fan followed by Tagart and Walker. Finally in 1954 Benda labeled this disease as Dandy Walker3. Associated congenital anomalies such as cleft palate, micrognathia, hypertelorism, cardiac anomalies, renal and skeletal malformation are said to be present in 48% cases2. In the present cases DWS is associated with unusual associations such as agenesis of corpus callosum, agenesis of vermis, agenesis of spleen along with some associated anomalies such as microphthalmia, cardiac anomalies, duplicated ureter, diaphragmatic hernia. DWS is a frequent cause of termination of fetus diagnosed prenatally4.

MATERIALS AND METHODOLOGY

The present study was conducted on 1200 humans fetuses (since Jan 2008- July 2016) in the Department of Anatomy, Government Medical College & Hospital, Chandigarh.

CASE REPORTS

Case 1

First case reported was a male fetus of 19th 6 weeks. Mother of fetus was 22yrs old, primi gravisda. Medical history and past history of parents were not suggestive of any etiological factors responsible for the defect. The indication of MTP was DWS.

| External examination | Internal examination |
|----------------------|-----------------------|
| Large head           | Cerebrum had pachygyria |
| bilateral club foot with over | hypoplasia of cerebellum making |
| ridding of 4th toe on right | floor of fourth ventricle visible |
| side and 2nd toe on left side. |                  |
| Right side cerebral hemisphere was smaller than left | posterior cranial fossa appeared larger |
Dandy walker malformation with unusual findings in fetal autopsies

Case 2

22+1 week female fetus born to 20 yr old mother who had an obstetric history of G2P1A1L1. Her first child was born alive but died at the age of 2 1/2 yrs, due to bilateral retinoblastoma. In rest of the pregnancies spontaneous abortion occurred at 2-3 months. In present case indication for MTP was Dandy Walker Syndrome, Microphthalmia, pyelectasis, echogenic shadow in heart, VSD, club foot on ultra sonogram. Maternal history showed that she had unilateral retinoblastoma which got operated at age of 6yrs. Paternal history showed excessive use of drugs, tobacco, smoke and anti depressant drugs.

| Gross examination | Internal examination |
|-------------------|----------------------|
| Microphalma       | Bilateral presence of double ureter(fig 1 and fig 2) |
| polydactyl of rt.foot | enlarged kidney (fig 1 and 2) |
| post axial polydactyl on left hand | hypoplasia of vermis(fig 3) |

Figure 1

Figure 2

Figure 3 showing hypoplasia of vermis

Case 3

21+2 week female fetus born to 22yr old mother. Medical history and past history of parents were not suggestive of any etiological factors responsible for the defect. The indication of MTP was DWS

| Gross examination | Internal examination |
|-------------------|----------------------|
| Microphalma       | Bilateral long fingers, |
| polydactyl of rt.foot | camptodactyl, |
| post axial polydactyl on left hand | contractus elbow and wrist(fig 4) |
| hypoplasia of vermis(fig 3) | B/L CTEV, Poor muscle bulk(fig 4) |

Figure 4 showing contractus elbow and wrist B/L CTEV, Poor muscle bulk

Figure 5 Lateral and fourth ventricle small compared to normal

Case 4

22 weeks female fetus born to yr mother, who had an obstetric history of G2A1. Medical history and past history of parents were not suggestive of any etiological factors responsible for the defect. The indication of MTP was DWS

| Gross examination | Internal examination |
|-------------------|----------------------|
| Microphalma       | Bilateral long fingers, |
| polydactyl of rt.foot | camptodactyl, |
| post axial polydactyl on left hand | contractus elbow and wrist(fig 4) |

Figure 6 showing left side diaphragmatic hernia (fig 6) with herniation of intestine, stomach, liver to thoracic cavity (fig 7)

Figure 8 showing asplenia, horizontally placed left lung, left lung hypoplasia, right lung with abnormal lobulation, aorta and pulmonary trunk arising from right ventricle (fig 9 and fig 10).

Figure 11 showing left cerebral hemisphere larger than right (fig 12).
Case 5

16 +3 weeks male fetus born to 25 yr old mother. The indication of MTP was DWS, abberent rt subclavian artery complex cardiac malformation. she had a medical history of viral infection for 2 week.

| Gross examination | Internal examination |
|-------------------|----------------------|
| normal            | heart horizontally placed with right atrium and ventricle enlarged (fig 14) |

Out of 1220 autopsies, dandy walker malformation was found in 5 fetuses (0.4%)

| Incidence of dandy walker syndrome |
|-----------------------------------|
| Male | Female |
| 2(40%) | 3 (60%) |
Dandy walker malformation with unusual findings in fetal autopsies

DISCUSSION

The Dandy-Walker complex is a rare and group of congenital intracranial malformation which comprises abnormalities of the posterior fossa. It can be classified as

1. Dandy-Walker malformation – which includes cystic dilatation of the 4th ventricle, partial or complete agenesis of the cerebellar vermis and an enlarged posterior fossa.
2. Dandy-Walker variant - cystic posterior fossa mass with variable hypoplasia of the cerebellar vermis and no enlargement of the posterior fossa.
3. Megacisterna magna - enlarged cisterna magna with normal cerebellar vermis and fourth ventricle. DWM accounts for 12% of all cases of congenital hydrocephalus.

The Dandy-Walker malformation has a slight female preponderance. In present study also we came across female preponderance, out of 5 fetuses with DWS, 3 were female and 2 male. DWS accounts for about 1:30,000 live births and is responsible for 4-12% of infantile hydrocephalus.

Philips JJ et al. in his study on DWM noted extra CNS abnormalities in 30 out of 44 cases. These abnormalities included congenital diaphragmatic hernia, renal anomalies, congenital heart defects & extremity anomalies such as clubfoot. Agenesis of corpus callosum was reported in one case. Various other authors had mentioned about associated CNS anomalies such as ventriculomegaly, corpus callosum agenesis, abnormalities of gyri, microcephaly, occipital meningocoele and encephalocoele. Murray et al in his study noted other extra-cranial anomalies such as polycystic kidneys, cardiovascular defects, polydactyly and cleft palate. In our present study agenesis of corpus callosum was noted in one case, cardiac anomalies were noted in two cases, renal anomalies in one case, polydactyly in one case, CTEV in one case.

According to Dandy and Walker dilatation of ventricular system occurred due to atresia of the foramina of Luschka and Magendie. How ever Benda suggested that it’s difficult to understand how atresia of these foraminas would lead to cerebellar vermis hypoplasia. Gardner et al gave a more clarified theory regarding hypoplasia of vermis. He proposed that early dilatation and herniation of rhombencephalic roof can be due to overproduction of CSF at fourth ventricle, this dilatation due to overproduction at the level of fourth ventricle can cause compression and secondary hypoplasia of cerebellar vermis. Hirsch et al.1 suggested that malformation can be classified into communicating and non-communicating depending on whether the foramina of Luschka and Magendie are open or closed., Non communicating is related with varying type of hydrocephalus.

Kolb and philips reported a very rare case in which there was association of DWS with congenital absence of spleen. Congenital absence of spleen is rare and life threatening condition. In our present finding one of the case there was aspelenia associated with congenital unilateral diaphragmatic hernia.

Chowdareddy N. et al reported a case of DWS, in 26 week female fetus, associated with congenital diaphragmatic hernia with herniation of liver, stomach & spleen into left hemithorax, hypoplasia of lung, they also noted Hydrocephalus, Absent corpus callosum & vermis, Cyst. Congenital diaphragmatic hernia (CDH) is an uncommon birth defect with incidence of 1:2000 to1:3000 births, of which 96% are Bochdalek type. We had also got similar findings in one of our case -22 week female fetus with left diaphragmatic hernia with herniation of liver, stomach and intestine to left thoracic cavity, absent corpus callosum, absent left temporal pole of cerebral hemisphere, large left cerebral hemisphere along with CVS defects.

Murray et al in his study on etiologic heterogeneity of DWS suggested that environmental factors, such as viral infection, alcohol, and diabetes, have been playing a role in its etiology. In our study we came across two cases with such a history. In one of the case mother had medical history of viral infection. In another case paternal history showed excessive use of drugs, tobacco, smoke and anti depressant drugs.

Various authors had mentioned about DWS association with cardiovascular anomalies. According to Hirsch JF et al anomalies associated with DWS includes encephaloceles, polycystic kidneys, and cardiovascular defects (mainly ventricular septal defects). According to Golden JA et al 26-38% of cases with DWS cases had extracranial manifestation including ventricular septal defect, patent ductus arteriosus, ASD, pulmonary stenosis, intestinal anomalies and renal defects. In our case, we came across a fetus with CVS defect we noted that aorta and pulmonary trunk were arising from right ventricle and left ventricle and left atrium were...
hypoplastic, same case had a left sided congenital diaphragmatic hernia with liver, stomach, intestines herniating into thoracic cavity, asplenia.

Eventhough hydrocephalus is considered as a major diagnostic element of DWS, recent research had suggested that in most cases hydrocephalus is not present at the time of birth and they had noted that hydrocephalus develops usually at first month of life. In such cases the diagnosis can only be made prenatally depending on the posterior cranial fossa abnormalities. Positive diagnosis before viability gives a option for pregnancy termination.

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 ventriculo peritoneal shunt to fenestration of the cystic membrane, a primary cystoperitoneal shunt or a ventriculocysto-peritoneal shunt.

CONCLUSION

Agenesis of corpus callosum interference with medullary control of respiration which often results in respiratory failure. DWS association with congenital diaphragmatic hernia and asplenia is very rare ultrasound & pathologic examination are indicated in DWS for screening of concurrent cranial and extra cranial malformation. Proper diagnosis of DWS is necessary for further treatment. Eventhough hydrocephalus is considered as a diagnostic element in DWS in most cases it appears after birth. So the posterior cranial fossa abnormalities should be noted prenatally as a diagnostic tool.

References

1. Surekha U Arakeri, Himanshu Mulay. Dandy Walker Syndrome with Unusual Associated Findings in a Fetal Autopsy Study. JKIMSU, Vol.4, No.1, jan-march 2015
2. Lavanya T, Cohen M, Gandhi SV, Farrell T, Whitby EH. A case of a Dandy-Walker variant: the importance of a multidisciplinary team approach using complementary techniques to obtain accurate diagnostic information. Br J Radiol 2008; 81: 242-245
3. Tadakamdia J, Kumar S, Mamatha GP. Dandy-Walker Malformation: An Incidental Finding. Indian Journal of Human Genetics 2010; 16: 33-35
4. Dr Shweta Mane, Dr Shruti Rao, Dr S.D Ladi, Dr S.S Aphale. Innovative DANDY WALKER SYNDROME: CASE REPORT. Journal of Medical and Health Science 4 : 1 Jan - Feb(2014) 309-311
5. Alessandro et al. dandy walker malformation and Wisconsin syndrome: novel case add further insight into the genotype-phenotype correlation of 3q23q25deletions. Orphanet journal of rare diseases .2013,8:75
6. Fischer EG: Dandy-Walker syndrome: An evaluation of surgical treatment. J Neurosurg 39:615, 1973. 13.
7. Osenbach RK, Menezes AH. Diagnosis and management of the Dandy-Walker malformation: 30 years of experience. Pediatr Neurosurg 1991; 18:179-85.
8. Philips JJ, Mahony BS, Siebert JR, Lalani T, Flinger T, Kapur RP. Dandy-Walker malformation complex: correlation between ultrasonographic diagnosis and postmortem neuropathology. Obstet Gynecol 2006; 107: 685-693
9. Murray JC, Johnson JA, Bird TD. Dandy-Walker malformation: etiologic heterogeneity and empiric recurrence risk.Clin Genet 1985;28:272-6
10. Dandy WE: The diagnosis and treatment of hydrocephalus due to occlusion of the foramina of Magendie and Luschka. Surg Gynecol Obstet 32:112, 1921. 8
11. Taggart JK, Walker AE: Congenital atresia of the foramen of Luschka and Magendie.AMA Arch Neurol Psychiatr 48:583, 1942.
12. Gardner E, O’Rahilly R, Prolo D: The Dandy~Walker, and Arnold-Chiari malformations: Clinical, developmental and teratological considerations. Arch Neurol 32:393, 1975
13. Hirsch JF, Pierre-Kahn A, Renier D, et al.: The DandyWalker malformation: A review of 40 cases. J Neurosurg 61:515, 1984.
14. Kolkhe N, Wisser J, Kurmanavicius J, Boltshauser E, Stallmach T, Huch A et al. Dandy-Walker Malformation: prenatal diagnosis and outcome. Prenat Diagn 2000; 20: 318-327.
15. Chowdareddy n.1, anil kumar y.c.1, nihalal1, mano j1, gopal k.1, ravichander1 and neetha r. A case of dandy walker malformation with congenital diaphragmatic hernia - a rare variant- case report. Indian journal of medical case reports 2013 vol.2 (4) october-december, pp.63-64
16. Golden JA, Rorke LB, Bruce DA (1987) Dandy-Walker syndrome and associated anomalies. Pediatr Neurosci 13: 38-44
17. Kumar R, Jain MK, Chhabra DK. DandyWalker syndrome: different modalities of treatment and outcome in 2 cases. Childs Nerv Syst 2001; 17: 348-52.

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