



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.

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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 hydrocephalus¹. 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% cases⁴. 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 prenatally⁵.

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.

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Fetuses were sent by the Department of Obstetrics & Gynaecology of the same institute for routine autopsy. These included the spontaneous abortions, stillbirths, intra-uterine deaths and therapeutic abortions after recognition of congenital malformations on ultrasonography or by karyotyping. The consent was taken from the parents. Before autopsy relevant maternal and family history along with antenatal USG findings were recorded. The sex, gestational age of fetus were noted. External features were noted and photographed. Radiography was also done in relevant cases. Internal examination of the fetus was done after performing routine autopsy procedure and photographed. The associated anomalies, found, if any, was also noted

CASE REPORTS

Case 1

First case reported was a male fetus of 19⁺6 weeks. Mother of fetus was 22yrs old, primi gravida. 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
<ul style="list-style-type: none">• Large head• bilateral club foot with over riding of 4th toe on right side and 2nd toe on left side.	<ul style="list-style-type: none">• Cerebrum had pachygyria• hypoplasia of cerebellum making floor of fourth ventricle visible• Right side cerebral hemisphere was smaller than left• posterior cranial fossa appeared larger

Case 2

22⁺¹ week female fetus born to 20 yr old mother who had a obstetric history of G₅P₁A₄L₁. 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
<ul style="list-style-type: none"> • Microphthalmia • polydactyly of rt.foot • post axial polydactyly on left hand 	<ul style="list-style-type: none"> • Bilateral presence of double ureter(fig 1 and fig 2) • enlarged kidney (fig 1 and 2) • hypoplasia of vermis(fig 3)

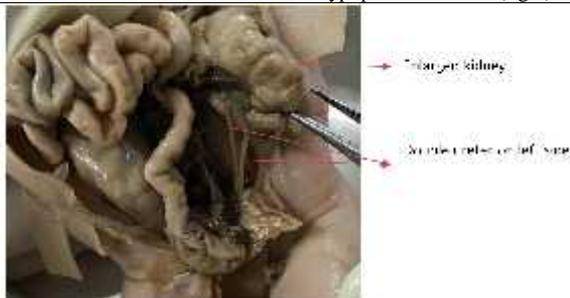


Figure 1

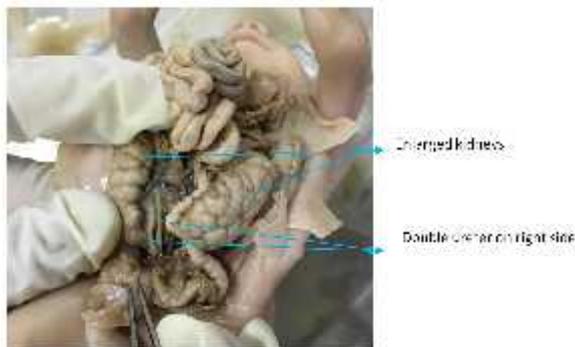


Figure 2



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, enlarged cistern magna, hypoplasia of b/l cerebellar hemisphere, aplasia of vermis

Gross examination	Internal examination
<ul style="list-style-type: none"> • Bilateral long fingers, • camptodactyly, • contractus elbow and wrist(fig -4) • B/L CTEV, Poor muscle bulk(fig 4) 	<ul style="list-style-type: none"> • Lateral and fourth ventricle small compared to normal (fig 5)



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 a obstetric history of G₄A₃. 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
<ul style="list-style-type: none"> • Short neck, • flat occiput, • low set ears, • retro microngnatia • ear dysmorphic, • rt.ear shows pre auricular tag. 	<ul style="list-style-type: none"> • Left side diaphragmatic hernia (fig 6)with herniation with herniation of intestine, stomach, liver to thoracic cavity (fig -7) • Asplenia, • Horizontally placed left lung-left lung hypoplasia,(fig 8) • rt lung with abnormal lobulation. • CVS-horizontally placed heart (fig 8). • aorta and pulmonary trunk arising from rt.ventricle.(fig 9 and fig 10) • Lt.ventricle and atrium hypoplastic • CNS-asymmetric posterior and middle cranial fossa • asymmetric cerebral hemisphere(fig 11) • left cerebral hemisphere larger than right(fig 12) • left temporal pole of cerebrum absent • Absence of corpus callosum(fig 13) • Small hind brain • Vermis too small to demarcate on inferior surface of cerebellum • Dilated lateral ventricle



Left sided diaphragmatic hernia with
in thoracic cavity

Figure 6 showing left sided diaphragmatic hernia



Small size of intestines and stomach in
thoracic cavity

Figure -7



left lung hypoplasia
horizontally placed heart.

Figure 8



Aorta arising from right ventricle.

Figure 9



Pulmonary trunk arising from right
ventricle

Figure 10



Figure 11 asymmetric cerebral hemisphere



Figure 12 left cerebral hemisphere larger than right



Figure 13 absence of corpus callosum

Case 5

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

Gross examination	Internal examination
<ul style="list-style-type: none"> normal 	<ul style="list-style-type: none"> 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%)



Figure 14 showing horizontally placed heart with right atrium and ventricle enlarged

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 hydrocephalus⁶.

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 was female and 2 male. DWS accounts for about 1:30,000 live births and is responsible for 4-12% of infantile hydrocephalus⁷.

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 meningocele and encephalocele⁸. Murray *et al* in his study noted other extra-cranial anomalies such as polycystic kidneys, cardiovascular defects, polydactyly and cleft palate⁹. 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. However Benda suggested that it's difficult to understand how atresia of these foramina would lead to cerebellar vermis hypoplasia^{10,11}. 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 vermis¹². Hirsch *et al* 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 hydrocephalus¹³

Kolble 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 asplenia associated with congenital unilateral diaphragmatic hernia^{8,14}.

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 to 1:3000 births, of which 96% are Bochdalek type¹⁵. 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 etiology⁹. 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)¹³. 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 defects¹⁶. 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 abnormalities¹³. 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 shunt¹⁷.

CONCLUSION

Agensis of corpus callosum interference with medullary control of respiration which often results in respiratory failure¹⁴. 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 prenataly as a diagnostic tool.

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