

## CASE REPORT

**Dandy Walker Syndrome with Unusual Associated Findings in a Fetal Autopsy Study***Surekha U Arakeri<sup>1\*</sup>, Himanshu Mulay<sup>1</sup>**<sup>1</sup>Department of Pathology, BLDEU's Shri B M Patil Medical College, Hospital & Research Centre, Bijapur-586101(Karnataka) India***Abstract:**

Dandy Walker Syndrome (DWS) is a congenital brain malformation characterized by hypoplasia or absence of cerebellar vermis, cystic dilatation of fourth ventricle and hydrocephalus. It is frequently associated with other congenital anomalies. Associated central nervous system anomalies such as agenesis of corpus callosum and vermis are associated with poor prognosis. Association of DWS with congenital absence of spleen is life threatening condition and has been reported very rarely. Autopsy findings of DWS with association of congenital absence of corpus callosum, vermis and spleen are reported in a stillborn fetus of 28 weeks gestation and review of relevant literature was done.

**Keywords:** Agenesis of spleen, Agenesis of vermis, Corpus callosum agenesis, Dandy walker syndrome

**Introduction:**

Dandy Walker Syndrome (DWS) is a congenital brain malformation characterized by hypoplasia or absence of cerebellar vermis, cystic dilatation of fourth ventricle and hydrocephalus [1, 2]. Prevalence of Dandy-Walker malformations (DWM) among live birth varies from 1 in 25,000 to 35000 [3]. DWM was described originally by Sutton. Further it was described by Dandy and Black fan followed by Tagart and Walker. Benda finally labelled this disease as Dandy Walker in 1954 [4]. DWM is frequently associated with other congenital anomalies and is reported in about 47 to 81 % cases of DWS [2]. In the present case DWS is associated with unusual associations such as agenesis of corpus callosum, agenesis of vermis and agenesis of spleen.

**Case Report:**

A female aged 30 year in the 28<sup>th</sup> weeks of gestation with obstetric history of 4<sup>th</sup> gravida, Para 3 & live 3 was admitted to the obstetric services. Her antenatal ultrasound check-up at 28 weeks, revealed dilatation of all ventricles in fetal brain and displacement of posterior fossa suggestive of DWS. Based on radiological diagnosis termination of pregnancy was done. A stillborn male fetus was delivered which was sent for autopsy study.

A male fetus with umbilical cord weighing 1.0 kg was received for autopsy. Umbilical cord was 10 cm in length. Two cystic lesions measuring 2 and 0.5 cm in diameter containing clear fluid were noted on the wall of umbilical cord which showed features of benign cystic lesions on microscopy. On external examination crown-to-heel length was 40 cm & head circumference was 28.5 cm. Enlarged head, club feet and low set ears were noted on external examination. More than 70 ml serosanguinous cerebrospinal fluid was drained from cranial cavity after opening the skull cavity (Fig 1, 2). Cerebrum was hypoplastic measuring 7x5x0.5cm. Cerebellum was hypoplastic and was measuring 3x2x1 cm. One of the cerebellar hemisphere, corpus callosum and vermis were absent. Cerebrum and cerebellum together weighed 10 gm. Fourth and lateral ventricles were dilated (Fig. 3)



**Fig. 1: Gross Photograph of Cut Open Skull Showing Excess Cerebrospinal Fluid Suggestive of Hydrocephalus.**



**Fig. 2: Gross Photograph of Cut Open Skull Showing Hypo plastic Cerebrum and Cerebellum**



**Fig 3: Gross Photograph Showing Absence of Vermis & Corpus Callosum**

On cut open thorax & abdominal cavity showed right & left lung weighing 10 & 25 gm respectively. Heart was 10 gm & liver was 40 gm. Spleen was not found. Right & left kidneys were 10 gm each. No specific pathology noted in left lung, liver, heart & kidneys on gross examination. Microscopy of brain tissue showed dilated ventricles & hypoplastic brain tissue. Extramedullary hematopoiesis was noted in liver. No specific microscopic findings in heart, kidney & lungs. Cystic lesions from the wall of umbilical cord showed benign cystic lesions. Based on these findings this case was concluded as DWS with hydrocephalus & agenesis of spleen.

#### **Discussion:**

Dandy Walker Syndrome (DWS) has several variants, out of which DWS malformation, DWS mega cisterna magna and DWS variant are better identified variants. DWS malformation encompasses cystic dilatation of fourth ventricle, complete or partial agenesis of cerebellar vermis and enlarged posterior fossa, while DWS variant comprises cystic posterior mass with hypoplasia of cerebellar vermis without enlargement of posterior fossa. DWS malformation is the most severe presentation of the DWS whereas DWS variant is less severe form of DWS [4]. In present case dilatation of 4<sup>th</sup> ventricle, complete agenesis of cerebellar vermis, absence of one cerebellar hemisphere, absence of corpus callosum & hydrocephalous was found with enlarged posterior fossa. Hence this case was reported as DWS malformation which is fatal variant of DWS. Dilatation of lateral ventricles was also noted in the present case on radiology.

In a study done by Philips JJ *et al* [5] extra CNS abnormalities were identified in 30 out of 44 cases. These abnormalities were congenital diaphragmatic hernia, renal anomalies, congenital heart defects & extremity anomalies such as

clubfoot. Agenesis of corpus callosum was reported in one case. Frequently associated CNS anomalies reported by other authors were ventriculomegaly, corpus callosum agenesis, abnormalities of gyri, microcephaly, occipital meningocele and encephalocele. Commonly associated extra cranial malformations are cardiac, renal and skeletal malformations and congenital diaphragmatic hernia.

Facial anomalies such as cleft palate, facial hemangiomas are also commonly reported [2,4]. Association of DWS with congenital absence of spleen has been reported very rarely [2, 5]. Congenital absence of spleen is rare and life threatening condition. It is commonly associated with cardiac malformation. The association between cardiac malformation & congenital absence of spleen has been reported by various authors [6]. In a study done by Kolble *et al* [2] fusion between spleen & pancreatic tissue was found in one case of DWS. Walpole *et al* [7] described 3 cases of DWM variant with cystic

dysplastic kidney & hepatic fibrosis. Out of which absence of spleen was noted in one case. In the present case spleen, corpus callosum & vermis were absent. DWS is a brain malformation of unknown etiology. Predisposing factors reported by various authors are infections, cranial trauma, and chronic disturbance in cerebrospinal fluid pressure, persistence of embryonic tissue, vascular lesions, teratogens, rubella, alcohol and maternal diabetes [5]. In the present case etiology was not known.

### Conclusion:

Agenesis of corpus callosum is associated with poor intelligence and interference with medullary control of respiration which often results in respiratory failure [2]. Congenital absence of spleen is rare and life threatening condition [6]. Hence ultrasound & pathologic examination are indicated in DWS for screening of concurrent cranial and extra cranial malformation.

### References:

1. Adams LR, Larroche JC, Vires LD. Central nervous system malformations. In: Barness EG, editors. *Potter's pathology of the fetus, infant and child*. 2<sup>nd</sup> ed. China: Mosby-Elsevier; 2007: 1999-2003.
2. Kolble N, Wisser J, Kurmanavicius J, Bolthausen E, Stallmach T, Huch A *et al*. Dandy-Walker Malformation: prenatal diagnosis and outcome. *Prenat Diagn* 2000; 20: 318-327.
3. 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.
4. Tadakamdia J, Kumar S, Mamatha GP. Dandy-Walker Malformation: An Incidental Finding. *Indian Journal of Human Genetics* 2010; 16: 33-35.
5. Philips JJ, Mahony BS, Siebert JR, Lalani T, Flinger CL, Kapur RP. Dandy-Walker malformation complex: correlation between ultrasonographic diagnosis and postmortem neuropathology. *Obstet Gynecol* 2006; 107: 685-693.
6. Plowman DEM. Congenital absence of spleen associated with cardiac abnormalities. *Br Med J* 1957; 1:147-148.
7. Walpole IR, Goldblatt J, Hockey A, Knowles S. Dandy Walker malformation (variant), cystic dysplastic kidney & hepatic fibrosis; a distinct entity or Meckel syndrome? *Am J Med Genet* 1991; 39(3):294-298.

\***Author for Correspondence:** Dr Surekha U. Arakeri, Department of Pathology, BLDEU's Shri B M Patil Medical College & R C, Bijapur-586101, Karnataka, India, Cell: +91984576844 E-mail: drsuarakeri@hotmail.com