CASE REPORT

Dandy-Walker malformation with meningoia and lissencephaly: A rare case report

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ABSTRACT

Dandy-Walker malformation is a rare congenital defect characterized by agenesis or hypoplasia of the cerebellar vermis with cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa. We present a rare case of Dandy-Walker malformation associated with a meningocele and lissencephaly, in a female baby born alive but died soon after. There was agenesis of the cerebellar vermis and lateral displacement of cerebellar hemispheres exposing the dilated fourth ventricle, which communicated with a posterior cranial fossa cyst. Cerebral hemispheres with incompletely formed sulci and gyri were observed. A midline foramen in the occipital bone was seen through which the meningeal layer herniated from posterior cranial fossa. Complete ante-natal examination, karyotyping & ultrasonography may help in early diagnosis and decision making.

Key words: Dandy-Walker malformation, congenital anomaly, fourth ventricle, lissencephaly, vermis

INTRODUCTION

The term Dandy-Walker Malformation (DWM) encompasses a wide variety of congenital malformations of the brain involving the fourth ventricle and cerebellum. These are frequently associated with other extra and intra-cranial developmental anomalies.11 Antenatal ultrasonography allows a definitive diagnosis only in cases with severe anomalies. Treatment and prognosis depend on the severity of the defect and milder forms may not need any treatment at all. The condition is considered as having a multifactorial origin. DWM may appear in pure form or may be associated with several other syndrome complexes. We present a case of DWM associated with a meningoia and lissencephaly in a new born female baby.

CASE REPORT

A full term 25 year old primi gravida with a non-consanguineous marriage presented to the hospital with onset of labour. She had regular ante-natal checks, was immunized according to prescribed schedule and family history was unremarkable. The latest antenatal USG showed a female fetus with a posterior fossa dilatation and abnormality [Figure 1]. She delivered a female baby by emergency caesarean section, indicated by foetal distress. The baby appeared drowsy, weighed 1.47 kg and had a poor cry; muscle tone was weak and reflexes were sluggish. Breathing was labored with in-drawing of intercostal spaces, and suckling was poor. Mouth was large with down-turned lips. The baby had a large globose nose and had hypertelorism. A fluctuant swelling measuring about 6×8 cm was seen on the occiput about five cm left of midline [Figure 2A]. All the fontanellae appeared enlarged and bulging. The baby stopped breathing and was declared dead two hours after birth. The body was embalmed and further dissection of cranial cavity carried out according to Cunningham’s Practical Manual. Cerebral hemispheres had very few widely spaced gyri; the sulci were very shallow, Lissencephaly [Figure 2D]. The tentorium cerebelli was bulging upwards. A meningocele was seen on the occipital region which extended through an occipital skull defect [Figures 1and 2C]. Cerebellum was underdeveloped. The cerebellar vermis was almost nonexistent with only a small remnant seen on the left side [Figure 2B] leaving behind a wide empty space. The fourth ventricle was enlarged and communicated with the meningocele. The cerebellar hemispheres appeared displaced laterally by the cyst. The floor of the fourth ventricle appeared normal. Institutional ethical committee clearance and parents’ consent was duly obtained for the case.
Sain, et al.: Dandy-Walker malformation

DWM is a rare developmental disorder with a reported incidence of 1 in 25000 to 35000. It is said to be one of the commonest congenital defects affecting the embryology of cerebellum. The syndrome complex associated with malformations of the fourth ventricle and cerebellum were first recognized by Dandy and Blackfan in 1914, was described in detail by Taggart and Walker in 1942 and named as Dandy-Walker Malformation by Benda in 1954. Some researchers used other terms like Dandy-Walker Variant, Dandy-Walker Complex, Dandy-Walker Continuum, Vermian-Cerebellar Hypoplasia, etc. Disagreement still exists amongst researchers as to what exactly constitutes the syndrome. Whatever the name, there is agreement that the syndrome complex mainly involves malformation of the cerebellum/ cerebellar hypoplasia, underdevelopment or absence of vermis along with its upward rotation and cystic dilatation of the fourth ventricle due to obstruction in its outflow.

Clinical features may not be obvious in some; Belfquih et al have reported a case of DWM in an asymptomatic individual, diagnosed incidentally. At the other end of the range, the affected individual may not survive for long, as was in the present case reported by us. Hydrocephalus may be seen in up to 90% of cases of DWM. In a case series by Sasaki-Adams et al, DWM was most commonly associated with cardiac abnormalities, followed by neurological, gastrointestinal, orthopaedic and genitourinary abnormalities. In a few cases it was associated with pulmonary abnormalities and psychiatric effects. The morphological features associated with DWM are- hydrocephalus, hypotonia, mental retardation, epilepsy. Cerebellar signs are also seen (ataxia, oculomotor apraxia). Hydrocephaly, though seen in 80 to 90% of patients of DWM is not to be included as a criterion for diagnosis as it is said to be a postnatal complication arising out of the malformation and not the cause of it. The cause of hydrocephalus is said to be the compression of aqueduct by the posterior fossa cyst, because it is seen that hydrocephalus always gets resolved by shunt surgeries. The association of DWM with a meningocoele though not rare, is an uncommon occurrence. The case which we present here has such a combination. Similarly, agenesia of corpus callosum, cardiac defects and facial anomalies are also seen in association with DWM. Shuto et al and Hirsch et al state that such co-existing conditions suggest that the time of origin of DWM may be around 7th to 8th week of intrauterine life, which correlates with the formation and migration of neural crest cells. Skeletal abnormalities too are associated with DWM.

The cerebellum develops from alar plate of the first two rhombomeres under the influence of several genes which exert their effect through hormones. The vermis develops from two independent primordia- independent of each other and also from the developing cerebellar hemispheres. Some authors suggest that development of posterior fossa cyst in DWM is the result of two processes occurring, either independent of each other or together, viz; arrest of development of the vermis and failure to establish flow of CSF through the Foramina of Magendie and Luschka.

The genetic inheritance of DWM is complex and not clear as yet. Though DWM is said to have a sporadic appearance, chromosomes 3, 6 and 7 have been implicated in the appearance of DWM. Chromosomes 9, 13 and 18 have also been implicated, but inconclusively.
CONCLUSION

DWM is an uncommon congenital anomaly of the development of hindbrain with a very wide spectrum of clinical symptoms. We have presented a case of DWM associated with a meningocele and lissencephaly which is rare. It is associated with several other syndrome complexes. Prognosis depends on the degree of mal-development and in select cases shunt surgeries are helpful in relieving the hydrocephalus.

REFERENCES


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