Observational Study

Dandy-Walker malformation : a review of cases diagnosed by prenatal ultrasonography

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Dandy-Walker (DW) malformation is characterised by agenesis of cerebellar vermis, enlarged posterior fossa and elevation of tentorium. It may be isolated or associated with other systemic anomalies. The purpose of this case series was to observe abnormalities diagnosed by prenatal ultrasonography (USG) and study the clinical outcomes of fetuses. We reviewed seven cases of Dandy-Walker malformation, all of whom presented differently. A proportion of parents were consanguineous (two cases). Four had severe early onset intrauterine growth restriction (UGR), two with associated oligohydramnios. Routine scan prior to 20 weeks enabled detection in two cases. Interestingly we had one case of recurrent DW. A posterior fossa cyst with defective or absent vermis was the sonographic feature in all cases. Two cases had extracranial malformations. Fetal karyotype was normal. Excluding three terminated pregnancies, two were neonatal deaths. Autopsy confirmation was possible in two cases. One surviving infant is being followed up for mild developmental delay. When diagnosed in utero, it can be extremely difficult to ascertain the extent of associated malformations and prognosticate the postnatal outcome. Fetal karyotype and echocardiogram (ECHO) can be useful. Diagnosis prior to 20 weeks enables termination of pregnancy since neurological outcome and prognosis remain guarded in such cases.

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Key words **: Dandy-Walker malformation, vermis defect, ultrasonography.**

Dandy-Walker malformation is characterized by agenesis of cerebellar vermis, enlarged posterior fossa and elevation of tentorium. It may be isolated or associated with other systemic anomalies[1]. About 15-30% of affected fetuses have chromosomal abnormalities[2]. Many infants die after birth, usually as a result of associated malformations. Majority of survivors have some degree of neurological handicap[2].

Dandy-Walker malformation is estimated to affect 1 in 10,000 to 30,000 newborns[3]. There were seven cases of Dandy-Walker malformations details of which we intend to present in this case review.

Dandy-Walker malformation affects brain development, primarily development of the cerebellum, which is the part of the brain that coordinates movement. In individuals with DW, various parts of cerebellum develop abnormally, resulting in malformations that can be observed with medical imaging. These abnormalities often result in problems with movement, coordination, intellect, mood, and other neurological functions[1].

In majority, signs and symptoms caused by abnormal brain development are present at birth or develop within the first year of life. Some children have hydrocephalus[4] that may cause increased head size. Up to half of affected individuals have intellectual disability that ranges from mild to severe, and those with normal intelligence may have learning disabilities.

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- **Dandy-Walker malformation is characterized by agenesis of cerebellar vermis, enlarged posterior fossa and elevation of tentorium.**
- **A posterior fossa cyst with defective or absent vermis is the sonographic feature. Fetal karyotype and fetal Echo can be useful.**
- **Diagnosis prior to 20 weeks enables termination of pregnancy.**

Children with Dandy-Walker malformation often have delayed development, particularly a delay in motor skills such as crawling, walking, and coordinating movements.

Less commonly, associated with DW, other CNS abnormalities have been reported such as agenesis of the corpus callosum, occipital encephalocoele etc which are associated with more severe signs and symptoms[5]. Associated extracranial anomalies include heart defects, malformations of the urogenital tract, extra fingers or toes (polydactyly) or fused fingers or toes (syndactyly), or abnormal facial features. Problems related to hydrocephalus are the most common cause of death in Dandy-Walker malformation[4,6].

Genetic Changes :

Researchers have found mutations in a few genes thought to cause Dandy Walker malformation, but these account for only a small number of cases[7].

Dandy-Walker malformation has also been associated with many chromosomal abnormalities such as trisomy 18 (most often), trisomy 13, 21 or 9, deletions or duplications. DW can also be a feature of certain genetic syndromes.

Research suggests that it could be environmental fac-

tors that affect early embryo development, such as exposure to teratogens. In addition, a mother with gestational diabetes is more likely to have a child with Dandy-Walker.

No clear inheritance pattern exists. Most cases are sporadic, whereas a small percentage of cases seem to run in families. First-degree relatives of Dandy-Walker malformation have an increased risk of developing the condition compared to general population[7].

Objective :

The purpose of this case series was to observe the associated abnormalities diagnosed by prenatal ultrasonography and the outcomes of fetuses with Dandy-Walker malformations.

Methods :

We reviewed seven cases of Dandy-Walker malformation, all of whom presented differently. Sonograms and medical reports of 7 cases were reviewed and information regarding each outcome was collected from fetal autopsy records, hospital charts and specialists caring for the surviving infants.

Description of Interesting Cases :

First case was a low risk primigravida at 19 weeks period of gestation referred with suspected posterior cranial fossa cyst on routine USG. A detailed level II scan showed defect in cerebellar vermis 6.5 x 6.7 mm. Fetal karyotype was normal. Patient requested for termination of pregnancy. Post delivery fetal autopsy done confirmed the vermian defect.

Second case was also a primigravida at 35 weeks period of gestation referred with severe intrauterine growth restriction (IUGR) and oligohydramnios. Anomaly scan at 19 weeks told to be normal but USG at 35 weeks showed defect in cerebellar vermis 1.2 x 1.2 cm. Fetal ECHO was normal. Vaginal delivery at 36 weeks was the outcome with low birth weight 1.2 kg. Postnatal neurosonogram of the surviving infant confirmed the cerebellar vermian defect (as isolated anomaly). Infant is under paediatric follow-up for mild neurodevelopmental delay.

Third case was Gravida abortion at 13 weeks period of gestation with history of consanguineous marriage and normal couple karyotype. Previous one fetus had shown DW at 14 weeks. USG done showed 17 weeks fetus with ventriculomegaly, absent vermis and posterior fossa cyst 6 mm. Fetal autopsy confirmed the findings. Risk of recurrence (1-5%) may be as high as 25%.

Observations and Results :

A proportion of parents were consanguineous (two cases).

Four had severe early onset intrauterine growth restriction, two with associated oligohydramnios and the other with shortened long bones such as short femur.

Interestingly we had one case of recurrent Dandy-Walker malformation, the risk of which is up to 1-5%[8].

Routine USG prior to 20 weeks enabled detection in two cases. A posterior fossa cyst with defective or absent vermis was the sonographic feature in all cases. Ventriculomegaly, though a common feature, was not seen in all. There were no other associated central nervous system (CNS) anomalies.

Two cases had extracranial malformations with multiple anomalies including cleft lip, single umbilical artery, absent kidneys, abnormal fetal echocardiogram (tricuspid atresia and transposition of great vessels). One case of DW variant was associated with multiple contractures and dysmorphic facies.

Chromosome abnormalities were found in none of the fetuses that underwent karyotype analysis. Fetal karyotype was available in 3 cases.

Excluding three terminated pregnancies, two were neonatal deaths (one was immediate and another late NND). Outcome of one fetus with multiple congenital anomalies was spontaneous vaginal expulsion at 29 weeks gestation. Only one DW infant survived (with isolated anomaly), was delivered by vaginal route and is currently under paediatric follow up for mild developmental delay.

Fetal autopsy confirmation was possible in two cases as in others either consent was not given or the autopsy was unsatisfactory due to maceration.

Conclusion :

When diagnosed in utero, it can be extremely difficult to ascertain the extent of associated malformations and prognosticate the postnatal outcome in DW. Fetal karyotype and fetal echocardiogram (ECHO) can be useful.

Diagnosis prior to 20 weeks gestation enables termination of pregnancy since neurological outcome and prognosis remain guarded in such cases[8,9,10].

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