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Dandy-Walker Malformation

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INTRODUCTION

Dandy-Walker malformation is a rare congenital malformation and involves the cerebellum and fourth ventricle. The condition is characterized by agenesis or hypoplasia of the cerebellar vermis, cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa. A large number of concomitant problems may be present, but the syndrome exists whenever these three features are found. Approximately 70-90% of patients have hydrocephalus, which often develops postnatally. Dandy-Walker malformation may be associated with atresia of the foramen of Magendie and, possibly, the foramen of Luschka. Dandy-Walker malformation first was described by Dandy and Blackfan.[1]

Since the original description, additional studies have reported on the various morphologic features of the syndrome. Not until 1954 did Benda first emphasized that atresia of the cerebellar outlet foramina is not an essential feature of the condition and suggested the now widely accepted term Dandy-Walker malformation.[2]

Studies by Hart et al.[3] further defined the characteristic triad of Dandy-Walker malformation as consisting of:

1. Complete or partial agenesis of the vermis.
2. Cystic dilatation of the fourth ventricle.
3. An enlarged posterior fossa with upward displacement of lateral sinuses, tentorium, and torcular herophili. The triad typically is found in association with supratentorial hydrocephalus, which should be considered a complication rather than part of the malformation complex .

Key Words:
Cerebral Ventricles, cerebellum abnormalities, Dandy-Walker Syndrome.

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Dandy-Walker complex

Classically, posterior fossa cystic malformations have been divided into Dandy-Walker malformation, Dandy-Walker variant, mega cisterna magna, and posterior fossa arachnoid cyst. Precisely differentiating the malformations may not be possible using imaging methods. Dandy Walker malformation, variant, and mega cisterna magna currently are believed to represent a continuum of developmental anomalies on a spectrum that has been termed the Dandy Walker complex.[4]

Dandy-Walker variant

Dandy-Walker variant consist of vermian hypoplasia and cystic dilttion of the fourth ventricle without enlargement of the posterior fossa (Figure 1).

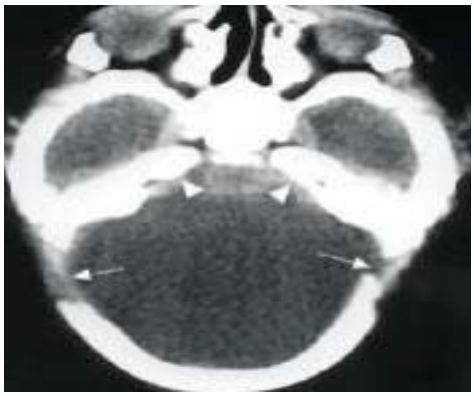

Fig. 1: Dandy-Walker malformation. Dandy-Walker variant in a 13-year-old girl with thoracal scoliosis. Sagittal T1-weighted MRI shows agenesis of the corpus callosum and a hypoplastic inferior vermis. The fourth ventricle is enlarged slightly, but the posterior fossa typically is normal in size.[6]

Mega cisterna magna :

The mega cisterna magna consists of an enlarged posterior fossa secondary to an enlarged cisterna magna, with a normal cerebellar vermis and fourth ventricle (Figure 2).

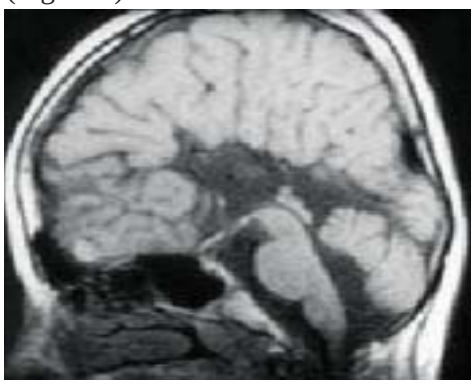

Fig. 2: Dandy-Walker malformation. Mega cisterna magna. Sagittal T1-weighted MRI shows a large retrocerebellar cerebrospinal fluid collection and a normal fourth ventricle and vermis.[6]

Dandy-Walker complex is characterized by an enlarged posterior fossa, high position of tentorium with upward displacement of the lateral sinuses, torcular herophili associated with varying degrees of vermian aplasia or hypoplasia, and a cystic dilatation of the fourth ventricle that nearly fills the entire posterior fossa. Since the vermis is present in posterior fossa arachnoid cyst, this is considered separately from Dandy-Walker malformation.[5]

Arachnoid cyst :

Retrocerebellar arachnoid cysts of developmental origin are uncommon but clinically important. True retrocerebellar arachnoid cysts displace the fourth ventricle and cerebellum anteriorly and show significant mass effect. Differentiation of posterior fossa arachnoid cyst from Dandy-Walker malformation is essential as surgical therapy differs between the two entities (Figure 3).

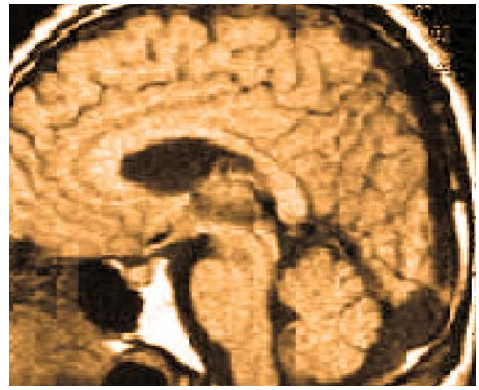

Fig. 3: Dandy-Walker malformation. Posterior fossa arachnoid cyst in a 15-month-old girl with a lumbar pilonidal sinus. Sagittal T1-weighted MRI shows a large posterior fossa cyst that is compressing the cerebellar hemispheres, vermis, fourth ventricle, and brainstem.[6]

Pathophysiology:
Dandy Walker malformations are formed during embryogenesis. Insults of varying severity to both the developing cerebellar hemispheres and fourth ventricle currently are believed to be the genesis of the anomaly. The nature and cause of the insult are unknown. Multiple diverse theories have been offered to explain the diffuse manifestations of Dandy-Walker malformation. To date, no single theory has proven satisfactory; therefore none has been accepted widely.[7]

Dandy and Blackfan[1] and Taggart and Walker[8] believed that the massive dilatation of the fourth ventricle originates from a congenital obstruction of the outlets of Luschka and Magendie. This theory includes the presence of a developmental cerebellar defect that begins before the embryologic differentiation of the fourth ventricle foramina, and results in blockage or atresia of the foramina of Magendie and Luschka. This, in turn, results in cystic transformation of the roof of the fourth ventricle and in an obstructive (Noncommunicating) hydrocephalus, in which a cyst arises from compromised absorption of CSF. The most comprehensive theory concerns dysembryogenesis involving the hindbrain. An insult that leads to developmental arrest in formation of the hindbrain, with lack of fusion of the cerebellum in the midline, can be localized temporally between the 7th and 10th gestational weeks. This results in persistence of the anterior membranous area, which extends and herniates posteriorly. Simultaneous formation of the foramen of Magendie, tentorium, superior longitudinal sinus, straight sinus, torcular herophili, and lateral sinuses helps to explain their association with Dandy-Walker malformation.

Inheritance:
The etiology is heterogeneous, and familial occurrence also has been reported. A few cases resulting from autosomal recessive genes have been reported, although in most patients, the cause of Dandy-Walker malformation is not known. Genetic counseling is critical to estimate the risk of recurrence of genetic disorders in family members.[9]

Etiologic heterogeneity and low recurrence risk in siblings (15%) for Dandy-Walker malformation have been reported. Increased frequency of an association with congenital heart disease, cleft palate, and neural tube defects appears to exist. An unusual case of an infant with both Ellisvan Creveld and Dandy-Walker syndromes and with homozygosity for an unusually long heterochromatic segment of the long arm of chromosome 9 (9qh+) was reported. An extensive tabulation of single gene disorders, chromosomal aberrations, teratogeninduced conditions, sporadic forms, or forms with undetermined inheritance associated with Dandy-Walker malformation also were reported.[9]

Predisposing factors:
Predisposing factors include gestational (First trimester) exposure to rubella, cytomegalovirus, toxoplasmosis, warfarin, alcohol, and isotretinoin.

Frequency:
In the US: The incidence of Dandy-Walker malformation is 1 case per 25,00035,000 live births. Dandy-Walker malformation accounts for

approximately 14% of hydrocephalus cases.

Mortality/Morbidity:
Overall mortality rates of 1250% have been reported in Dandy-Walker malformation in the pediatric neurosurgical literature. Associated congenital anomalies contributed to 83% of postnatal deaths. Mortality rates have improved significantly over the last 30 years as a result of better anesthesia and shunting devices and the reduction of posterior fossa exploration. Sudden and unexpected death is an uncommon but wellrecognized occurence in patients with Dandy-Walker malformation.

The prognosis is difficult to formulate. The prognosis is only moderately favorable, even when hydrocephalus is treated early and correctly. In one study, 3 patients with isolated Dandy-Walker cysts with hydrocephalus diagnosed in utero were treated at birth with shunting, and all 3 had normal outcomes. An extreme range of severity is seen in this malformation. The presence of multiple congenital defects may affect survival adversely. Some people have Dandy-Walker variant during their entire lives without any symptoms. Some infants may have it in association with other syndromes, resulting in severe complications or death.[10]

Sex:
Dandy-Walker malformation occurs more frequently in females than in males. The maletofemale ratio was 1:3 in one Spanish series.

Age:
Depending on the time of onset and degree of hydrocephalus, the age at diagnosis varies from birth to older childhood. Presentation in adulthood has been reported but is unusual. Patients with Dandy-Walker variant are more likely to present in adulthood than in infancy or childhood.

Clinical Details:
Patients with Dandy-Walker malformation present with developmental delay, enlarged head circumference, or signs and symptoms of hydrocephalus. The clinical presentation depends to some extent on the combination of the developpmental anomalies in the infant. An estimated 80% of patients had normal ventricles at birth, and by age 1 year, 80% had ventriculomegaly. Hydrocephalus is present in approximately 90% of patients at the time of diagnosis. If no other anomalies are present, the only symptom can be an abnormal enlargement of the head. Typical signs of increased intracranial pressure seen in older children and adults may be absent in infants secondary to the ability of the head to increase in size. Macrocrania usually is the consequence of hydrocephalus, but in some patients, it results from massive enlargement of the posterior fossa by the posterior fossa cyst. In this situation, macrocrania precedes development of hydrocephalus, giving the skull a characteristic dolichocephalic shape with bulging of the occiput. Difficulty with balance, spasticity, and poor fine motor control are common. The degree of developmental delay appears to be related to the level of control of hydrocephalus and to the extent of supratentorial anomalies. Interference with respiratory control centers in the brainstem may cause respiratory failure. Seizures occur in 15,30% of patients. Hearing or visual difficulties,

systemic abnormalities, and CNS abnormalities are associated with poor intellectual development. Subnormal intelligence (Intelligence quotient <83) is manifested in 41,71% of patients. More severe intellectual impairment has been observed in patients with agenesis of the corpus callosum.[11]

Preferred Examination:
Dandy-Walker malformation is diagnosed best with the help of US and MRI. US may be the initial examination performed since it can be done portably, without sedation, and allows multiplanar imaging.

The introduction of modern imaging techniques, specifically MRI, has radically changed the evaluation of symptoms related to the posterior fossa.

MRI usually is performed for detailed evaluation of Dandy-Walker malformation lesions and complications after the diagnosis is suspected using computed tomography (CT) and US. MRI can best define the relationship between the cyst and the fourth ventricle and can detect vermian rotation and the presence of signs of vermian dysgenesis.

MRI allows surgeons to view the cerebellum and associated structures accurately and to determine which form the malformation has taken and to what extent the malformation has progressed. MRI also demonstrates which space should be shunted first. Recently, MRI has been used frequently for diagnosis of fetal craniospinal anomalies.

CT scanning also is useful in Dandy-Walker malformation, since it can distinguish between hydrocephalus associated with Dandy-Walker and hydrocephalus with other etiologies.

Classic abnormal findings of Dandy-Walker malformation described on cranial CT and MRI also can be demonstrated on cranial sonography. US is used routinely during the antenatal period as a screening method and is used in particular for postnatal followup studies of hydrocephalus. US evaluation of posterior fossa cystic abnormalities in the newborn is best accomplished via a posterolateral fontanelle approach or through the cisterna magna posteriorly.

In recent years, plain radiography has been used primarily in the evaluation of shunt malfunction and for diagnosis of associated anomalies.[12,13]

Limitations of Techniques:
Plain radiographs have diagnostic importance in the evaluation of shunt malfunction and for evaluation of bone abnormalities.

CT is an effective diagnostic method but exposes the infant to ionizing radiation. Clearly distinguishing subtypes of the Dandy-Walker complex on axial CT images is difficult. In addition, evaluating subtle supratentorial pathologies and associated abnormalities on CT images may not be easy because its routine use is constrained by the axial plane.

MRI is relatively expensive. Highquality MRI images require patient cooperation or sedation.

US is limited because it is heavily operator dependent. US does not image well such abnormalities as the gyral, dural, tentorial, and skull anomalies

accompanying Dandy-Walker malformations.[13]

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