II. NEONATOLOGY

POSITIVE DIAGNOSIS OF DANDY - WALKER SYNDROME

Marioara Boia[1], Aniko Manea[2], Daniela Iacob[1], Dana Mihut[2], ES Boia[1], RE Iacob[1]
[1]University of Medicine and Pharmacy "Victor Babes" Timisoara
[2]Neonatology and Health Care Clinic Timisoara

Abstracts

Dandy - Walker syndrome is a rare affection in newborn pathology and due to its neuropathologic implications and central nervous system associated anomalies early diagnosis is very important. The most important complication is evolving hydrocephalus, characterized by occipital prominence of the skull.

The authors would like, in the present work, to highlight the importance of prenatal and neonatal ultrasonography in early diagnosis of Dandy Walker syndrome and of central nervous system associated anomalies.

Key words: Dandy-Walker Syndrome, ultrasound scan.

Introduction

The Dandy- Walker complex includes the Dandy-Walker malformation and the variant of Dandy - Walker syndrome. Dandy- Walker malformation consists of enlargement of the posterior fossa as a result of the cystic dilatation of the fourth ventricle in partial or total agenesis of the cerebellar vermis. In approximate 5 - 10 % of the cases congenital hydrocephalus is determined. The Dandy - Walker variant is characterized by light enlargement of a posterior fossa, a cyst of the posterior fossa continuing the fourth ventricle which is united with through a narrow duct and a disgenetic cerebellar vermis.

In Dandy- Walker malformation several associated anomalies occur (70 % of cases); most frequent are: agenesis of the corpus callosum and the variant of neuronal migration disorders.

Neuropathology

Fundamental anomaly of the posterior brain is related with the wrong forming of the cerebellar vermis and the roof of the fourth ventricle. The start point of the malformation seems to be, mainly, a delay or a failure in opening Magendie's orifice which leads CSF storage and cystic dilatation of the fourth ventricle. Despite the subsequent opening of the foramina of Luschka (usually opened in Dandy- Walker malformation) the cystic dilatation of the fourth ventricle and CSF leaking persists (fig.1 and fig.2).

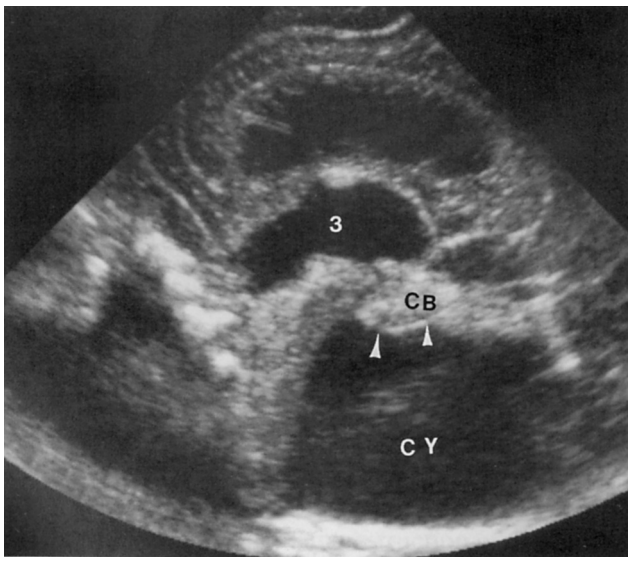

Fig. 1. Dandy- Walker malformation. A cyst of posterior fossa.

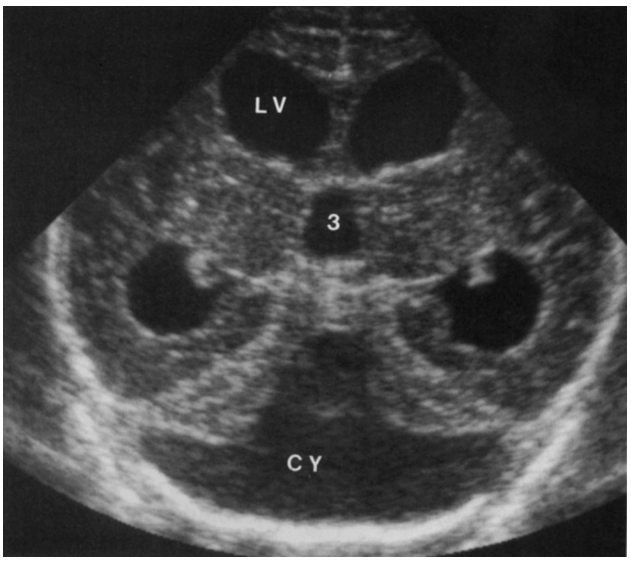

Fig. 2. Coronal posterior scan. A large cystic structure; behind - the compressed cerebellar vermis.

Appearance of the Dandy- Walker malformation is not cleared yet; nevertheless, the period of maximum development of the orifices is the second and the third month of gestation, during the prosencephalic development. This period coincides with neuronal migration explaining in this way the association of Dandy- Walker malformation with agenesis of the corpus callosum and neuronal migration disorders.

Dandy- Walker syndrome – neuropathology

Primary injuries	Cystic dilatation of the fourth ventricle Agenesis of the vermis Hydrocephalus
Associated injuries	Agenesis of the corpus callosum (20-30%) Cerebral neuronal heterotopia (aprox. 15%) Aqueductal stenosis (aprox.5-10%) Cerebral gyral anomaly (aprox.10%) Syringomyelia (aprox. 5-10%) Occipital encephalocele (aprox.10-15%) Inferior olivary anomalies or dentate nuclei (aprox.20%) One or more associated (70%)

Positive diagnosis

Clinical dominant feature is hydrocephalus, characterized by occipital prominence of the skull and large dilatation of the fourth ventricle with enlargement of the posterior fossa. Marked hydrocephalus is present at few cases in neonatal period. Use on a large scale of prenatal and neonatal ultrasonography allowed the track down of several cases since intrauterine and early neonatal period, despite the absence of the quickly head growing and intracranial hypertension signs, which usually appear after first year of life. In some situations hydrocephalus can appear only in maturity period. Association of other anomalies of central nervous system and systemic malformations represents another clinical aspect. Amongst these cardiac disorders and renals are the most frequent, 20-30% from the cases with postnatal diagnosis and 60-80% from the cases with prenatal ultrasonography. Dandy - Walker variant results from a limited disorder of cerebellar hemispheres. Patients with both anomalies can present a development delay and an increasing of head circumference. The neurological development delay depends on the severity of the associated supratentorial anomalies: hydrocephalus, agenesis of the corpus callosum, gray matter heterotopia, polymicrogyria and occipital encephalocele. Hydrocephalus could be present at birth but more frequently it develops later, 75% of patients appearing after three month from birth. Regarding systemic anomalies the most frequent are cardiac anomalies and polydactyly.

Echographic manifestations of the disorder are better seen in sagital sections, especially medium sagital and they are represented by:

- a cyst of posterior fossa, big, with liquid, homogeneous, which is in the baloon-shape fourth ventricle;
- partial or complete absence of vermis;
- cerebellar hemispheres hypoplasia ;
- tentorium raising.

In the coronar section is confirmed the presence of a posterior fossa cyst and high position of the tentorium (fig. 3).

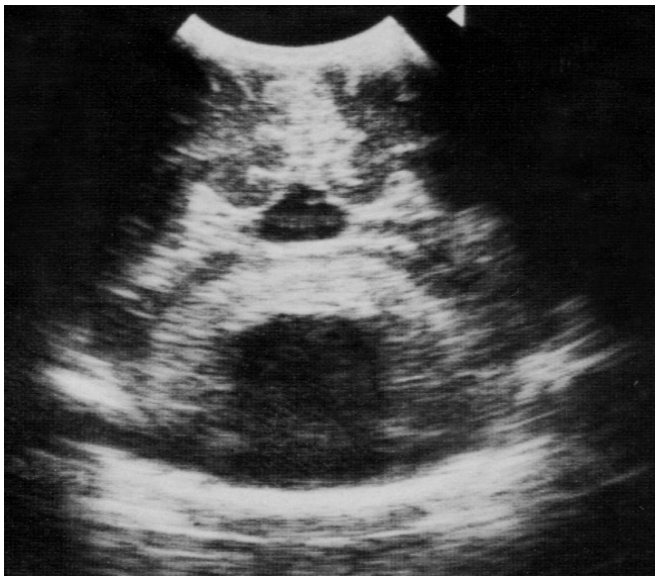

Fig. 3. Coronal posterior scan. Premature baby with congenital hydrocephalus. A large cystic structure is present.

Differential diagnosis:

Arachnoid cyst of posterior fossa and dilatation of the large cisterna, situation when cerebellar vermis and the fourth ventricle are not affected. In the Dandy-Walker variant the two cerebellar hemispheres, hypoplasic, in sagital section, could give the impression of a normal vermis. The frontal sections and the CT through posterior fossa will mark a narrow duct between anterior aria of the fourth ventricle and the rest of the posterior fossa, absence of vermis respectively.

Conclusions

1. Dandy Walker syndrome is a rare disease in medical practice, but fortunately can be easily detected by ultrasound exam.
2. Associated malformations are common especially neurological malformations: agenesis of corpus calos, disorders in the process of neural migration, occipital encephalocel.
3. Evolutive hidrocephaly is the most severe complication, it can occur at birth, but in 75 % the anomaly appears after the age of 3 months.

References:

1. Babcock D.S., Sonography of congenital malformations of the brain. *Neuroradiology* 1986; 28: 428-439
2. Counsell S.J., Rutherford M.A., Cowan F.M., Edwards A.D. ,Magneti cresonance imaging ofpreterm brain injury*, Arch.Dis.in Childhood Fetal and Neonatal* Ed, 2003, 88, 269-274
3. Grand EG, Schellinger D, Richardson JD, Real time ultrasonography of the posterior fosa, *J Ultrasound Med*, 1983, 2, 73-87
4. Funk KC, Siegel MJ, Sonography of congenital midline brain malformations, *Radiographics*, 1988, 8, 11-25
5. Hill LM, Breckle R, Gehrking WC – *Prenatal Detection of Congenital Malformations by Ultrasonography* – Am. J. Obstet. Gynecol., 151:44-50, 1985
6. Volpe JJ – *Human Brain Development* – in JJ Volpe (Ed.), Neurology of the Newborn, 3rd Ed., Pholadelphia: Saunders, p. 3, 1995

Correspondence to:

Marioara Boia,
Gospodarilor Street, No. 42,
Timisoara 300778,
Romania
E-mail: eemboia@rdslink.ro