DANDY-WALKER SYNDROME VARIETY IN THE CONTEXT OF A POLINALFORMATIVE SYNDROME. THE LIMITS OF CONVENTIONAL ULTRASOUND EXAMINATION

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REZUMAT
O mare parte a malformațiilor cerebrale pot fi identificate încă din viața intrauterină, în special pe parcursul examinării morfologice de trimestrul al doilea, efectuată la 20-24 săptămâni de gestație. Sintomul Dandy-Walker este o anomalie foarte rară, care constă din asocierea hidrocefaliei de diverse grade, lârgirii cistei magnii și agenezia totală sau parțială a vermisului cerebelos, care poate fi vizualizată ca o imagine chistică ce face legătura între ventriculii III și IV. Această situație “clasăică” poate fi detectată sau doar suspiciune încă din viața intrauterină. De la această asociere pot exista diverse variante, majoritatea fără semnificații intrauterine. În urma introducerii examinării cerebrale tomografice (CT) sau prin rezonanță magnetică nucleară (RMN), această definiție a putut fi largită, ea cuprinzând la ora actuală: 1. malformatia Dandy-Walker clasică (lărgirea fosei posterioare, agenezia vermisului cerebelos, tentoriu înalt, hidrocefalie); 2. varianta Dandy-Walker (hipoplazia variabilă a vermisului cerebelos, cu sau fără lârgirea fosei posterioare); 3. megacisterna magna (cisterna magna lărgită, cu păstrarea integrității vermisului cerebelos și a ventriculului IV). Hidrocefalia a fost în mod clasic considerată ca un element esențial de diagnostic, ea putând apare în evoluție, chiar la câteva luni postpartum. Tehnica ecografică actuală, ce permite examinarea fetului intrauterin 3D, prin care examinatorul poate obține orice incidentă 2D, este necesară pentru evaluare, metoda cea mai valoroasă fiind TUI, cu posibilitatea prelucrării volumului achiziționat prin alegerea incidentei, în care se pot efectua o serie de sonograme 2D consecutive, tehnică similară RMN.

Cuvinte cheie: variația Dandy-Walker, hidrocefalie congenitală, ecografie fetălă, RMN

ABSTRACT
A significant number of brain malformations can be identified during intrauterine life using ultrasonography, especially doing a morphological scan at 20-24 weeks of gestational age. The Dandy-Walker syndrome is a very rare abnormality (its incidence is 1/25000-1/35000 newborns, which involves the association of: ventriculomegaly in various degrees, enlarged cysterna magna, partial or total agenesis of the cerebellar vermis, which can be visualized as a cystic image between the 3rd and 4th ventricles. This “classic” form can usually be detected or suspicioned from intrauterine life. The other forms cannot be diagnosed before delivery, all clinical signs starting to develop after birth. As a result of the introduction of CT examination or the MRI investigations, this definition can be enlarged, including the following: 1. the classic Dandy-Walker malformation (enlarged posterior fossa, partial or complete agenesis of the cerebellar vermis, the high position of the tentorium, hydrocephaly); 2). megacisterna magna (i.e. enlarged cystern, the cerebellar vermis and the fourth ventricle are normal); 3). Dandy Walker variant (variable cerebellar vermis hypoplasia, mild enlargement of the posterior fossa). Hydrocephaly was usually considered as an essential element to the diagnosis of this abnormality, and it can even appear in evolution a few month after delivery. Our case was an extremely rare variety, without any signs during intrauterine life, all the symptoms starting after delivery, the diagnosis being confirmed by MRI examination, which used some new incidences for examination of the cerebellum, some of them being extended to the intrauterine fetal ultrasound scanning. In 3D ultrasound examination the most valuable method for cerebellum seems to be TUI (Tomographic Ultrasound Imaging), a multislice method which offers the examiner the possibility to rotate the volume and choose the best incidence in which he can use different consecutive 2D slices.

Key Words: Dandy-Walker variant, congenital hydrocephaly, fetal ultrasonography, MRI

INTRODUCTION

The Dandy-Walker syndrome, first described in 1914, indicates the association of: 1,2
1. Ventriculomegaly in various degrees;
2. Enlargement of posterior cranial fossa;

3. Partial or total agenesis of the cerebellar vermis, which can be visualized as a cystic image between the 3rd and 4th ventricles.

As a result of the introduction of CT examination or using the MRI investigations this definition can be enlarged including the following: 3
1. The classic Dandy Walker malformation (dilated posterior fossa, agenesis of the cerebellar vermis, high position of the tentorium, hydrocephaly);
2. Megacisterna magna (i.e. enlarged cystern, the cerebellar vermis and the 4th ventricles remaining normal);
3. Dandy Walker variant (variable cerebellar vermis hypoplasia, with or without the dilation of the posterior fossa).
Hydrocephaly was usually considered as an essential element in the diagnosis of this abnormality, and it can even appear in evolution a few months after delivery.

The standard intrauterine fetal ultrasound examinations or the postnatal transphontanellar ultrasonography don't often reveal the Dandy-Walker variant, the assessment of these cases being obtained postnatal, only undergoing an MRI examination.4-9

To identify these cases, the medical literature suggests using a hinder incidence to the standard transcerebral incidence, yet determining only the severe anatomical variants of the syndrome. Other fetal malformations are often associated to determining the diagnosis of this particular syndrome.

CASE REPORT

A 32 years old primigravida, with a correct follow-up clinical and ultrasound assessment, with a Down syndrome risk score based on NT, embryologic biometry, serum level of βhCG and αFP of 1/1674 (at a normal value less than 1/250), was diagnosed with low site placenta implantation, observed at 12 weeks of gestation.

At 20 weeks gestational age, at a morphological ultrasound scan, no fetal malformation was observed, the intracranial structures were normal, the cerebellar vermis was present in standard examination and the ventricular cavities were also normal for this gestational age. Marginal placenta praevia was confirmed, without any clinical signs (uterine bleeding).

At 31 weeks gestational age the pregnant woman presented false premature labor that was treated with tocolytic, uterosedative and antispastic agents. The prevention of hyaline membrane disease was performed by administrating corticosteroids (Celestone). Marginal placenta praevia was reconfirmed at an ultrasound examination.

At 33 weeks gestational age the pregnant woman presented a minor uterine bleeding episode followed by massive hemorrhage at a few hours. An emergency cesarean section was performed. During the cesarean section marginal accreta placenta praevia was confirmed.

The newborn, a female of 2140 gr. with Apgar index of 6 at one minutes and 7 at five minutes presented transitory apnea. She had PPV with O2 100% and later with O2 40%. Cardiopulmonary radiography showed a fixed elevation of the right diaphragm suggesting a congenital right phrenic nerve paresis with a peribronchovascular trauma. At that moment, the newborn presented transient tachipnea of newborn ("wet lungs", with prominent vascular markings and fluid in the fissures).

24 hours postnatally the status of the newborn became deteriorated with cyanosis, hypotonia, low blood pressure and low O2 saturation (Silvermann score 5/6). CPR showed right pneumothorax with pneumomediastine. The symptomatology was
improved with orotracheal intubation and continuous mechanical ventilation associated with continuous pleural drainage. After four weeks complete remission was noticed.

Starting with the second week of life, the cranial perimeter increased progressively (starting from 31.5 cm at birth to 34 cm at two weeks, 35 cm at three weeks, 40 cm at eight weeks).

Once the hydrocephaly had been acknowledged a transfontanellar ultrasonography was performed, showing the following: well differentiated central tissue, LV = 19 mm, RV = 21 mm with non homogeneous transsonic content, dilation of anterior horns with hiperechogenic walls, moderate dilation of the third ventricle, intermediary mass present. The aspect pleaded for medium form evolutive bicavity hydrocephaly.

Most often, hydrocephaly is caused by obstruction of Sylvius aqueduct, Lushka or Magendie holes.
Rarely, the hydrocephaly appears through increased of CRL production or decreased of absorption capacity in subarachnoid space, arterio-venous malformations.

Isolated hydrocephaly is frequently caused by congenital obstruction of Sylvius aqueduct which makes the connection between third and fourth ventricles.

The most frequent anomaly with hydrocephaly is the meningocele with Arnold-Chiari malformation. After birth, hydrocephaly can be caused by cerebral bleeding or cerebral infections.

The studied case was of a premature newborn (gestational age of 33 weeks) born through Caesarean Section (marginal placenta praevia) with fetal distress with transitory tachipnea of the newborn and pneumothorax secondary to therapy (CPAP endonasal) and phrenic nerve paralysis.

Starting from the second week of life the newborn presented progressive hydrocephaly initially confirmed as being posthemorrhagic (CRL Pandy reaction positive, transfontanellar US), any other possible causes of hydrocephaly being excluded at the moment.

Still, the association of progressive hydrocephaly was installed early postnatal with phrenic nerve paresis, which raised the suspicion of a complex malformation syndrome, with indication for MRI. The MRI report confirmed important bilateral dilation of intracerebral liquid cavities, with important subsequent compression of cerebral parenchyma; it also revealed the presence of a cerebral arterio-venous malformation at cortical level, in front of anterior horn of right lateral ventricle (3/3/2 cm), with presence of drainage vessels and regional blood accumulation (hyposignal T1 and hypersignal T2), without fixing contrast substance. MRI also revealed partial agenesis (inferior and anterior) of the cerebellar vermis with moderate enlargement of the fourth ventricle and wide communication with cysterna magna, described as a variety of Dandy-Walker syndrome. Pituitary gland and orbits were normal.

MRI conclusions:
1. Triventricular massive hydrocephaly;
2. Cerebral arterio-venous right malformation, with cerebral bleeding in subacute stage;
3. A variant of Dandy-Walker syndrome.

CONCLUSIONS

1. The studied case is a variety of Dandy-Walker syndrome with partial agenesis of the anterior and inferior cerebellar vermis.
2. Positive diagnosis was based on MRI report.

3. Standard incidence of prenatal ultrasound examination of the cerebellum does not allow the imaging of this variety of the syndrome.
4. Conventional transfontanellary ultrasound has limitations in the diagnosis of this particular malformation.
5. Diagnosis is possible using prenatal US examination of the cerebellum in new, unconventional slides, taken from the MRI technique, using even the transversal and oblique subcerebellum incidence.
6. Using the newest ultrasound technique (3D and 4D scanning of the head) the obstetrician can obtain almost the same slices as MRI examination using the specific methods such as Magic Cut, TUI, niche mode,
performing some new incidences for the examination of the cerebellum. Some of them can be extended to the ultrasound intrauterine fetal scanning, especially when using 3D ultrasound technique the examiner can select and obtain any 2D slice necessary for optimal assessment. In 3D ultrasound examination the most valuable method for cerebellum seems to be TUI (Tomographic Ultrasound Imaging), a multislice method, similar to MRI, which offers to the examiner the possibility to rotate the volume and choose the best incidence, in which he can use different consecutive 2D slices.10

REFERENCES


