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CENTRAL NERVOUS SYSTEM ABNORMALİTİES DİAGNOSED BY PRENATAL SONOGRAPHY İN THE EARLY SECOND TRİMESTER

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SUMMARY

Ultrasound is an efficient method for the prenatal diagnosis of central nervous system abnormalities. We present a case of Dandy Walker malformation and a case of mild ventriculomegaly diagnosed by fetal sonography in the early second trimester over the past 6 months.

Key words: *dandy Walker malformation, fetal sonography, mild ventriculomegaly, prenatal diagnosis*

ÖZET

Erken İkinci Trimesterde Prenatal Ultrasonografi ile Tanı Konulan Merkezi Sinir Sistemi Anomalileri

Ultrasonografi merkezi sinir sistemi anomalilerinin prenatal tanısı için etkin bir yöntemdir. Bu yazıda, son 6 ay içerisinde fetal sonografi ile erken ikinci trimesterde tanı konulan bir Dandy Walker malformasyonu ve bir hafif ventrikülomegali vakası sunulmaktadır.

Anahtar sözcükler: *dandy- Walker malformasyonu, fetal sonografi, hafif ventrikülomegali, prenatal tanı*

INTRODUCTION

Central nervous system (CNS) anomalies are frequent and often have an unfavorable outcome. Modern, high-resolution ultrasound equipment has a unique potential in evaluating normal and abnormal anatomy of the fetal neural axis from the very early stages of development. However, identification of selected anomalies remains a challenge in many cases. In this report, we describe two cases of CNS anomalies identified by antenatal sonography in the early second trimester over the past 6 months at the Department of Obstetrics and Gynecology, Baskent University Ankara Hospital.

CASE 1

A 22-year-old, primigravida woman started to visit our antenatal clinic in the first trimester. At the 163/7th week of gestation, ultrasound showed a cyst communicating with the fourth ventricle between the two cerebellar hemispheres in the posterior cranial fossa (Figure 1a). The cerebellar vermis was

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hypoplastic. Ultrasound findings suggested the diagnosis of Dandy-Walker malformation. One week later, we demonstrated the characteristic ultrasound findings of Dandy-Walker malformation including a small cerebellar vermis, an enlarged posterior fossa where the width of cisterna magna was 10,6 mm, and antero-laterally displaced but apparently normal cerebellar hemispheres (Figure 1b and Figure 1c). The lateral ventricles were enlarged and had an atrial width of 10,6 mm at this gestational age (Figure 1d). Fetal cranial magnetic resonance imaging (MRI) also confirmed the diagnosis. The fetal karyotype and maternal serology for the TORCH group of infections were normal. Specialist fetal echocardiography showed a structural normally connected heart with no obvious structural defect. The family decided to interrupt the pregnancy after counseling with a neurosurgeon and a pediatrician. After termination of the pregnancy, postmortem findings were also compatible with Dandy-Walker malformation.

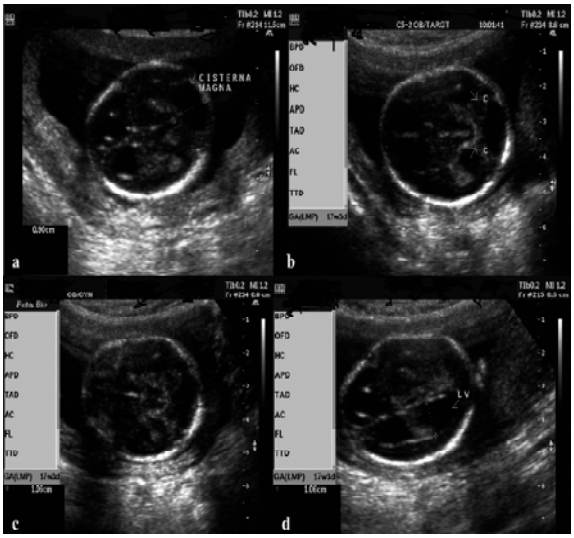

Figure 1 a-d: *Dandy-Walker Malformation. a. Cystic appearance of posterior cranial fossa at the 16th week of gestation.* ***b.*** *The typical ultrasonographic features including a small cerebellar vermis, an enlarged posterior fossa and antero-laterally displaced normal cerebellar hemispheres at the 173/7 week of gestation.* ***c.*** *Enlarged cisterna magna measured 10,6 mm.* ***d.*** *Enlarged lateral ventricle.*

CASE 2

A 33-year-old, primiparous woman, attended our antenatal clinic from the first trimester of her second pregnancy. The atrial width of the right lateral ventricle was measured 11 mm by fetal sonography at the 16th week of gestation (Figure 2). There were no any cerebral or extracerebral structural abnormalities. The fetal karyotype and maternal serology for the TORCH group of infections were normal. The couple was informed that the majority of babies with such a mild ventriculomegaly have a completely normal outcome if there are no other associated anomalies and a normal karyotype exists. However, the couple opted for termination of the pregnancy.

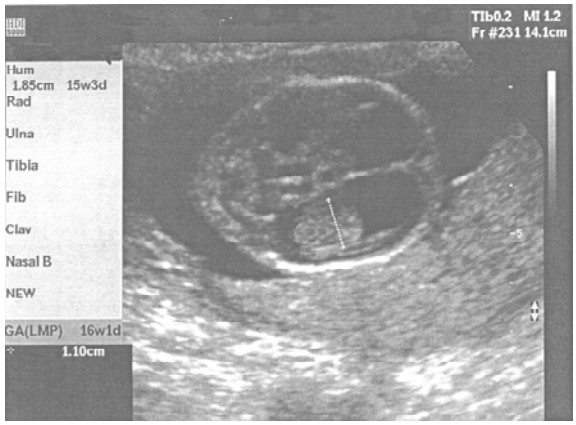

Figure 2: *The atrial width of the right lateral ventricle was measured 11 mm at the 16th week of gestation.*

DISCUSSION

The term Dandy-Walker complex is used to indicate a spectrum of anomalies of the posterior fossa that are classified by axial computed tomography as follows: classic Dandy Walker malformation (enlarged posterior fossa, complete or partial agenesis of the cerebellar vermis, elevated tentorium), Dandy-Walker variant (variable hypoplasia of the cerebellar vermis with or without enlargement of the posterior fossa), and megacisterna magna (enlarged cisterna magna with integrity of both cerebellar vermis and fourth ventricle). This classification was challenged after the introduction of MRI. The excellent resolution of sagittal planes made possible by MRI has demonstrated that the classification based on computed tomography axial planes is inadequate to describe the anatomic derangement encountered in the Dandy- Walker complex. Some degree of vermian dysgenesis can be found in all cases, even with megacisterna magna, whereas Dandy-Walker malformation and Dandy-Walker variant have so many similarities that a clear-

cut distinction is often impossible[1]. According to the available evidence, the antenatal sonography can make a certain diagnosis only of the most severe anatomic varities of the Dandy-Walker complex, that is commonly referred to as classic Dandy-Walker malformation. The Dandy-Walker malformation is a very rare congenital malformation of posterior fossa with an incidence of 1/25,000 to 1/35,000 births, representing about 4% of hydrocephalic cases[2]. It is frequently associated with other neural defects, usually ventriculomegaly, and other midline anomalies such as agenesis of the corpus callosum, holoprosencephaly, and cephaloceles. The frequency of associated malformations range between 50 and 70%. Chromosomal abnormalities are found in 15-30% of the cases. Other extracranial anomalies include polycystic kidneys, cardiovascular defects, and facial clefting.

The sonographic diagnosis of classic Dandy-Walker malformation is easy from midgestation[3] and has been reported as early as 14 weeks by using vaginal sonography[4]. The enlarged cisterna magna connected with the fourth ventricle through a defect in the cerebellar vermis can be easily shown in the transcerebellar view.

In most infants with absence of intrauterine hydrocephalus, development of symptoms of hydrocephalus or other neurologic symptoms occurs within the first year of life. The postnatal mortality rates as high as 24% have been reported in the first neurosurgical series. Subnormal intelligence has been reported in 40 to 70% of cases[2, 5,6]. The clinical significance of Dandy-Walker variant and megacisterna magna is uncertain.

In the prenatal evaluation of of classic Dandy-Walker malformation, a detailed fetal sonography including fetal echocardiography, fetal karyotyping and maternal serology for the TORCH group of infections (especially for cytomegalovirus infection and toxoplasmosis) should be offered. Counseling with a neurosurgeon, neuropediatrician, or neuropathologist would seem necessary. If the diagnosis is uncertain, fetal cranial MRI can be useful in deciding the clinical management. If the mother decides to carry on with the pregnancy, regular sonographic monitoring should be recommended.

The sonographic assessment of the cerebral ventricles has been the object of many studies and many different approaches to the definition and diagnosis of fetal ventriculomegaly have been proposed. The measurement of the transverse diameter of the ventricular atrium at the level of the glomus of the choroid plexus is easily obtained and has been found to be highly reproducible[7]. The atrial diameter is constant at 7,6 ± 0,6 mm (mean ± SD) from 14 to 38 weeks gestation[8]. The normal measurement of the atrium should be less than 10 mm at any time during gestation. Mild ventriculomegaly is commonly used to define the cases with an atrial width of 10 to 15 mm. It is associated with an increased risk of heterogeneous nervous and non-CNS anomalies. When the atrial width of lateral ventricle is greater than 15 mm, it is more likely to represent hydrocephalus. Approximately 15% of fetuses with ventriculomegaly will have an aneuploidy, only 2% if the hydrocephalus is isolated, but 17% if other findings are noted.

Although the prognosis is favorable in the majority of mild ventriculomegaly cases, the development of hydrocephalus has also been observed. When isolated, it has no consequences, in most cases. The risk of an abnormal neurologic outcome is increased when the atrial width is 12 mm or more, and when the diagnosis is made in the second trimester rather than later in gestation[9]. Ventriculomegaly may also be unilateral; it has been suggested that in this case it is usually a benign finding.

Despite the fact that it is unclear whether or not mild ventriculomegaly is an independent risk factor for chromosomal abnormalities, it would seem necessary to offer fetal karyotyping. Prenatal evaluation also should consist of detailed sonographic examination including fetal echocardiography and maternal serology for the TORCH group of infections (especially for cytomegalovirus infection and toxoplasmosis). Additionally, fetal MRI may be used to diagnose subtle cerebral maldevelopment in fetuses with mild ventriculomegaly[10].

In conclusion, when the Dandy-Walker malformation is diagnosed prenatally, clinical management should include further sonographic screening including fetal echocardiography, fetal karyotyping and search for the TORCH group of infections (especially for cytomegalovirus infection and toxoplasmosis) in the maternal blood. If the diagnosis is uncertain, fetal MRI can be useful in deciding the clinical management. Although the prognosis is favorable in the majority of mild

ventriculomegaly cases, the development of hydrocephalus has also been observed. Targeted sonographic examination including fetal echocardiography, fetal karyotyping and search for the TORCH group infections in the maternal blood should be performed. Fetal cranial MRI may be adjunctive to the antenatal sonography to confirm the diagnosis or identify subtle cerebral maldevelopment in fetuses with mild ventriculomegaly.

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