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# Ultrasonographic evaluation of ventriculomegaly cases

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## Abstract

**Objective:** To evaluate retrospectively the demographic data of the patients diagnosed in or referred with the pre-diagnosis of ventriculomegaly to Perinatology Department of Adana and Ankara Hospitals of Başkent University.

**Methods:** In this study, 61 pregnant women with fetal ventriculomegaly diagnosed in our clinic or referred by other centers to the Perinatology Department of Ankara and Adana Hospitals of Başkent University between May 2008 and March 2013 were evaluated in terms of their demographic data, diagnosis weeks, concomitant anomalies and the course of their pregnancies. SPSS v. 16.0 was used for statistical calculations.

**Results:** The analysis of 61 pregnant women between 20 and 41 years old showed that it was the first pregnancy of 45.9% of them. Only 16.4% of them were at or over 35 years old. It was observed that 4.9% of them were conceived by assisted reproduction techniques. The rates of mild (10–12 mm), moderate (12.1–14.9 mm) and severe ventriculomegaly (≥15 mm) were 65.6%, 24.7% and 4.8%, respectively. Toxoplasma, Cytomegalovirus, Rubella or Herpes virus infections were not found in any case. Down syndrome was found in the karyotype analysis of 3 patients (4.9%). The ventriculomegaly was unilateral in 67.2% of the cases. The detection rate between 16 and 24 weeks which were also the weeks for detailed ultrasonography was 52.5%. There was an additional anomaly in 33.3% of the cases. The most frequent concomitant anomalies were found as increased nuchal thickness (13.3%), corpus callosum agenesis (11.1%) and nasal bone hypoplasia (8.9%). In their follow-ups, it was observed that the findings were regressed in 53.8% of the cases, progressed in 19.3% of the cases and remained unchanged in 26.9%.

**Conclusion:** When ventriculomegaly is detected, the presence of additional anomalies should be investigated by detailed ultrasonographic examination during etiological investigation. In the selective cases, the physicians may utilize the method of magnetic resonance imaging to evaluate additional cerebral anomalies. It is also necessary to recommend karyotype analysis and investigating Toxoplasma, Rubella, Cytomegalovirus and Herpes virus infections in the presence of additional anomaly and even in isolated cases regardless of the level of ventriculomegaly. Patients should be followed up regularly.

**Keywords:** Isolated ventriculomegaly, additional anomalies, follow-up.

## *Özet:* Ventrikülomegali vakalarının ultrasonografik değerlendirilmesi

**Amaç:** Başkent Üniversitesi Adana ve Ankara Hastaneleri Perinatoloji bilim dalında tanı almış veya ventrikülomegali ön tanısı ile refere edilmiş hastaların demografik verilerinin retrospektif olarak değerlendirilmesi.

**Yöntem:** Bu çalışmada Mayıs 2008 ve Mart 2013 tarihleri arasında Başkent Üniversitesi Ankara ve Adana Hastaneleri Perinatoloji Bilim Dalına dışarıdan gönderilen veya kliniğimizde tespit edilen fetal ventrikülomegalisi olan 61 gebe demografik verileri, tanı konulma haftaları, eşlik eden anomaliler ve gebeliklerin seyri açısından değerlendirildi. İstatistiksel hesaplamalarda SPSS v. 16.0'dan faydalanıldı.

**Bulgular:** 20–41 yaş arası 61 gebenin değerlendirmesinde, %45.9'unun ilk gebelikleriydi. %16.4'ü 35 yaş ve üzeriydi. %4.9'unun yardımcı üreme teknikleri ile gebe kaldığı izlendi. Hafif (10–12 mm), ılımlı (12.1–14.9 mm) ve ciddi ventrikülomegali (≥15 mm) oranları sırası ile %65.6, %24.7 ve %4.8 idi. Hiçbir vakada toksoplazma, sitomegalovirüs, rubella veya Herpes virüs enfeksiyonu saptanmadı. Karyotip analizinde 3 hastada Down sendromu tespit edildi (%4.9). Ventrikülomegalilerin %67.2'si tek taraflı idi. Ayrıntılı ultrasonografi haftası olan 16–24 haftaları arası tespit oranı %52.5 idi. %33.3 ek anomali mevcuttu. En sık eşlik eden anomaliler sıklık sırasına göre artmış nukal kalınlık (%13.3), korpus kallozum agenezisi (%11.1) ve nazal kemik hipoplazisi (%8.9) olarak izlendi. Takiplerde %53.8 vakada bulguların gerilediği, %19.3'ünde ilerlediği ve %26.9'unda değişmeden kaldığı izlendi.

**Sonuç:** Ventrikülomegali tespit edildiğinde etyoloji araştırmasında ayrıntılı ultrasonografik muayene ile ek anomalilerin varlığı araştırılmalıdır. Selektif vakalarda ek serebral anomalileri değerlendirmek için manyetik rezonans görüntüleme yönteminden faydalanılabilir. Karyotip analizi ve toksoplazma, rubella, sitomegalovirüs ve Herpes virüs enfeksiyonlarının araştırılması ek anomali varlığında ve hatta izole vakalarda ventrikülomegalinin derecesi ne olursa olsun önerilmelidir. Hastalar düzenli takibe alınmalıdır.

**Anahtar sözcükler:** İzole ventrikülomegali, ek anomaliler, takip.

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## Introduction

The term ventriculomegaly (VM) is used for the cases where anterior horns of ventricle, cavum septum pellucidum and choroid plexus can be seen in the axial plane and lateral ventricle measurement on the plane is 10 mm and above at the glomus level of choroid plexus (**Fig. 1**).[1,2] There are various definitions according to the width of lateral ventricle. When lateral ventricle measurements are 10–12 mm and 12.1–15 mm, the definitions of mild and moderate VM are used.[3,4] However, some authors deny 10-12 mm as mild VM and define mild VM as 10-15 mm.[5] In measurements which are 15 mm and above, severe VM term is used.[6] While mild VM prevalence is 7.9 out of 10,000 live births, severe VM prevalence is reported as 3.6 out of 10,000 live births.[7]

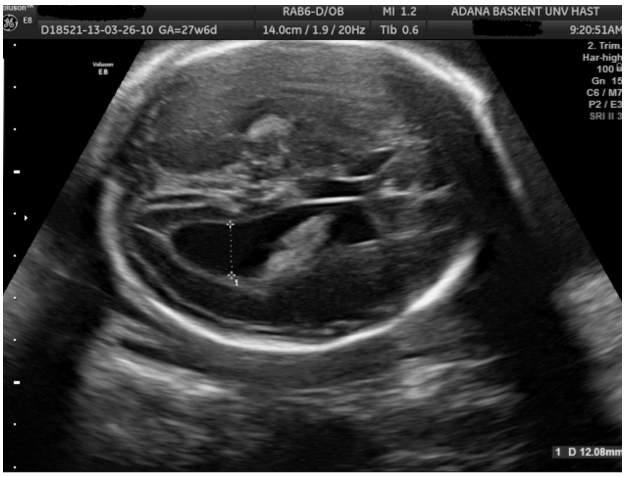


**Fig. 1.** Moderate ventriculomegaly.

Many factors are assumed in the etiology such as infections; cerebral atrophy induced by white matter injury and/or cases causing absorption of cerebrospinal fluid to decrease; obstructive causes such as Dandy-Walker malformation or aqueductal stenosis; developmental anomalies such as encephalocele, corpus callosum agenesis (**Figs. 2a** and **2b**); genetic disorders such as trisomy 13, 18 and 21; and cases such as choroid plexus papilloma which may cause excessive cerebrospinal fluid production.[8] This expansion in ventricles may be accompanied by anomalies such as hydrocephaly, gray matter migration anomalies, corpus callosum agenesis, trisomies and microcephaly.[5] The incidence of associated cerebral or extracerebral anomalies varies between 41% and 78%.[2] Many authors believe that the presence of additional malformations is directly associated with the prognosis. Prognosis is observed better in most of the isolated VM cases.[2,9] Therefore, prenatal diagnosis becomes crucial.

In this study, we aimed to analyze the demographic data, diagnosis weeks, concomitant anomalies and the course of pregnancy retrospectively in patients diagnosed with ventriculomegaly during intrauterine period.

## Methods

In this study, 61 pregnant women with fetal ventriculomegaly diagnosed in our clinic or referred by other centers to the Perinatology Department of Ankara and

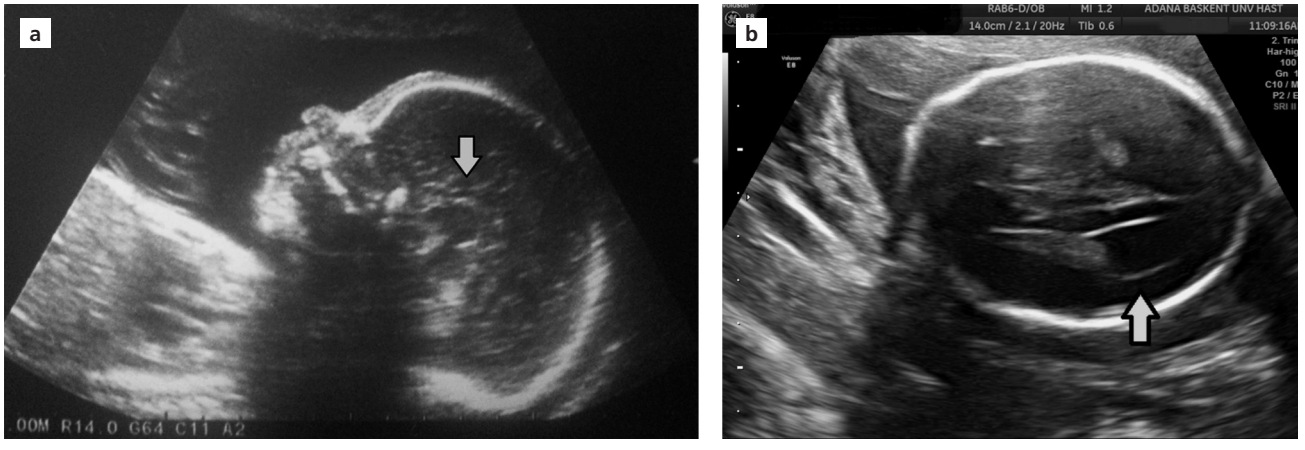


**Fig. 2.** (**a**) Corpus callosum agenesis (arrow). (**b**) Corpus callosum agenesis, colpocephaly, teardrop appearance (arrow).

Adana Hospitals of Başkent University between May 2008 and March 2013 were evaluated in terms of their demographic data, diagnosis weeks, concomitant anomalies and the course of their pregnancies. All fetuses were evaluated by detailed fetal biometric examination. Statistical data were analyzed by SPSS v. 16.0 (SPSS Inc., Chicago, IL, USA) software.

## Results

The analysis of 61 pregnant women between 20 and 41 years old showed that it was the first pregnancy of 45.9% of them. Only 16.4% of them were at or over 35 years old. It was observed that 4.9% of them were conceived by assisted reproduction techniques. The rates of mild (10–12 mm), moderate (12.1–14.9 mm) and severe ventriculomegaly (≥15 mm) were 65.6%, 24.7% and 4.8%, respectively. No Toxoplasma, Rubella, Cytomegalovirus (CMV) and Herpes virus infection was found in any of the pregnant women. Down syndrome was found in the karyotype analysis of 3 fetuses (4.9%). The ventriculomegaly was unilateral in 67.2% of the cases. The detection rate between 16 and 24 weeks which were also the weeks for detailed ultrasonography was 52.5%. The most frequent concomitant anomalies were observed as increased nuchal thickness (13.3%), corpus callosum agenesis (11.1%), and nasal bone hypoplasia (8.9%). Additionally, obstetric magnetic resonance was applied to 44.3% of the pregnant women. Of pregnant women, 72.5% of them delivered at 37 weeks or later. In their follow-ups, it was observed that the findings were regressed in 53.8% of the cases, progressed in 19.3% of the cases and remained unchanged in 26.9%.

## Discussion

In the detailed ultrasonographic examination performed on 18–22 weeks of gestation, it is routinely recommended to measure the width of lateral cerebral ventricles.[1,10,11] In the lateral ventricle measurements, 10–12 mm is defined as mild VM, 12.1–15 mm as moderate and 15 mm, and above as severe VM.[3,4]

Infections may also have a role in the etiology. In the study of Doğan et al., severe VM was found in 5 of 8 cases with CMV infection, increased periventricular echogenicity, intracranial calcification in 4 cases, thalamic hyperechogenicity in 3 cases, and mega cisterna magna.[12] Tijana et al. found VM, which did not display any finding on previous weeks, in patient with positive toxoplasmosis at 25 weeks of gestation.[13] Dommergues et al. detected CMV positivity in 29% of the cases in their study.[14] In various publications, infection positivity was observed as 10–20% in severe VM cases while it was 1–5% in mild VM cases.[9,15,16] Therefore, it is recommended to evaluate all cases diagnosed with VM in terms of infection.[6,15,17,18] In our study, no infection factor was found; the reason is the rate of severe VM seen in our patients as low as 4.8%.

Chromosomal anomaly incidence varies in VM cases between 0% and 14%.[5,6,15] In our study, Down syndrome was found in 4.9% (3) cases. While Nicolaides et al. reported chromosomal anomaly incidence as 3% in isolated VM cases, they showed this rate as 36% in the presence of additional anomaly. Also, aneuploidy was reported at a low rate in isolated severe VM cases compared to isolated mild VM cases.[19] Similarly, Melchiorre et al. reported the rate of chromosomal anomaly in isolated cases as 2.8%.[20] Gaglioti et al. did not find any chromosomal anomaly in severe VM cases while aneuploidy rate was 3.5% in mild and moderate VM cases.[16] Gezer et al. reported chromosomal anomaly incidence (6.8%) in fetuses with severe ventriculomegaly higher than the fetuses with mild ventriculomegaly (4.2%). Chromosomal anomaly incidence (8.6%) in fetuses with isolated ventriculomegaly was also found higher than those with additional anomaly (3.8%).[21] Sezik observed Type 2 triploidy case with VM accompanied by atrioventricular septal defect.[22] Kara described the association of VM and 47 XXY syndrome in a case report.[23]

In ventriculomegaly cases, it is possible to observe both cerebral and extracerebral malformations as an additional anomaly. They especially accompany with severe VM. The most frequent concomitant anomalies of severe VM cases are corpus callosum agenesis and bifid spine.[24,25] In mild and moderate VM cases, this rate varies between 10% and 76%.[17,26] Various studies reported additional anomaly incidence at rates reaching up to 50%.[6,11,15] Gaglioti et al. found additional anomaly in 60% of severe VM cases. In 88% of these cases, families preferred to terminate the pregnancy.[16] Tatlı et al. found additional anomaly in 9% of cases with 10-15 mm ventricle width.[27] In our study, the most frequent additional anomalies (33.3%) were increased nuchal thickness (13.3%), corpus callosum agenesis (11.1%) and nasal bone hypoplasia (8.9%).

We observed in our study that the findings were regressed in 53.8% of the cases, progressed in 19.3% of the cases and remained unchanged in 26.9%. Ouahba et al. reported regression in 11% of 167 mild VM cases, and they observed more regression in these cases in terms of neurological development.[9] Melchiorre et al. found a progression at a rate of 15.7%. In those with progression, there was poor prognosis in terms of neurological development and association with chromosomal anomalies.[20]

During magnetic resonance imaging, Levine et al. found additional findings at a rate of 13.5% which may change patient management in cases found to have anomaly by ultrasonography.[28] Gezer et al. asserted that ventricle width and brain parenchyma volume rate may be helpful to determine prognosis in magnetic resonance imaging. Parenchyma volumes of those with poor prognosis were found to be low.[29] We applied obstetric magnetic resonance imaging to 44.3% of our cases. We confirmed the diagnosis of corpus callosum agenesis in four fetuses by ultrasonography, cortical atrophy in one case, and encephalomalacia in one case. In line with these findings, magnetic resonance imaging is significant to confirm the suspected diagnoses established by ultrasonography. The use of magnetic resonance imaging is useful for pathologies such as neuronal migration disorders, delayed sulcation, gyrus formation and heterotopias which can be detected at late second and third trimesters and overlooked by ultrasonography.[30,31]

After the 11 years of follow-up of 101 children with isolated VM, normal psychomotor development was observed in 89 of them, and neurological disorder in the spectrum reaching out from language delay up to the severe mental retardation.[12]

Vergani et al. found neurological development retardation clearly lower in groups with 12 mm and lower widths than the group with 12 mm and above (3% vs. 23%).[6] Devaseelan et al. reported neurological development retardation as 14% in children with progressed VM during intrauterine period.[32] In another study, scores below the normal levels were reported for fine motor skills and language development in children with persisting ventricle width during prenatal period.[33]

## Conclusion

When ventriculomegaly is detected, the presence of additional anomalies should be investigated by detailed ultrasonographic examination during etiological investigation. In the selected cases, the physicians may utilize the method of magnetic resonance imaging to evaluate additional cerebral anomalies. In line with the current information we have, we recommend performing karyotype analysis and investigating Toxoplasma, CMV and infections such as Rubella regardless of the level of ventriculomegaly and even in the isolated ventriculomegaly cases without any additional anomaly. Patients should be followed up regularly. The parents should be informed in detail for the neuropsychiatric conditions that may arise during postpartum period.

**Conflicts of Interest:** No conflicts declared.

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