

M. Tahmasebi MD¹
N. Afsar MD²
M. Bastani MD, MPH²

Accuracy of Ultrasound in Detection of Gross Prenatal Central Nervous System Anomalies after the Eighteenth Week of Gestation

Background/Objective: Ultrasound (US) detection of prenatal central nervous system (CNS) anatomic anomalies is very important in making decision about therapeutic termination. In the present study, the accuracy of US in detection of gross prenatal CNS anatomic anomalies has been investigated.

Patients and Methods: 3012 pregnant women were scanned after 18 weeks of gestation by an expert operator in a referring center. All delivered fetuses were followed after birth through clinical examination and sonography.

Results: In this study, the accuracy of US in detection of gross CNS anatomic anomalies of fetuses after 18 weeks gestation was found to be 100%. The sensitivity, specificity, positive and negative predictive values of US were 100%. In sonographic examination of these 3012 pregnant women, 36 fetuses were detected with CNS anomalies, some of whom had more than one anomaly. Gross CNS anomalies observed included microcephaly, hydrocephaly, anencephaly, holoprosencephaly, ventriculomegaly, meningocele, encephalocele, lissencephaly, agenesis of corpus callosum, bilateral choroid plexus cysts and hypoplastic cerebellum.

Conclusion: US is highly operator dependent and operator experience may be the most determinant affecting the results. Sonographic scanning after 18 weeks of gestation is associated with the best results.

Keywords: ultrasonography, fetal abnormalities, congenital defects

Introduction

Prenatal diagnosis of central nervous system (CNS) anatomic anomalies is very important in making decision about therapeutic termination. In the past several decades, prenatal abnormalities were mainly detected by maternal serum analysis, amniocentesis, cordocentesis and chorionic villous sampling.¹⁻³

Ultrasound (US) detection of prenatal abnormalities is a non-invasive technique, which is more acceptable by patients. Several studies have shown an accuracy of 92% to 99.7% for US detection of CNS anatomic anomalies.⁴⁻⁹

This study was designed to assess the accuracy of US in detection of gross prenatal CNS anatomic anomalies. There are certain ethnic groups residing in Khuzestan such as Arabs and Bakhtiarians among which the familial marriage is common. So it may cause an increased rate of fetal structural anomalies. Also, since it was a frequent question of pregnant women in Khuzestan who referred to our imaging center that how much they can rely on sonography reports about anomalies in their fetus and fetus health, this study designed to reveal the fact.

Patients and Methods

This is a prospective study of diagnostic accuracy, sensitivity, specificity and predictive values of US in detecting prenatal gross CNS anomalies in pregnant

1. Assistant Professor, Department of Radiology, Golestan Hospital, Jondishapour University of Medical Sciences, Ahvaz, Iran.

2. Department of Radiology, Golestan Hospital, Jondishapour University of Medical Sciences, Ahvaz, Iran.

Corresponding author:
Mohammad Bastani
Address: Department of Radiology,
Golestan Hospital, Ahvaz, Iran.
Tel: +98611-3349298
Fax: +98611-2220432
Email:
Mohammadbastani2000@yahoo.com

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Table 1. Frequency of CNS anomalies

Type of anomaly	Number of fetuses
Microcephaly	3
Ventriculomegaly	6
Bilateral choroid plexus cysts	4
Holoprosencephaly	3
Anencephaly	5
Hydrocephaly	6
Meningocele	10
Encephalocele	4
Agenesis of corpus collosum	3
Hypoplastic Cerebellum	1
Lissencephaly	1
Total	46

women with gestational age of 18 weeks or more.

This study was conducted in Ahvaz, center of Khuzestan county, southeast of Iran, in a referral center for obstetrics sonography between July 2005 and June 2006. Three thousand and forty eight pregnant women were scanned after 18 weeks of gestation; 3012 cases were found eligible and entered the study. They were all referred by obstetricians/gynecologists for routine workup of the second trimester of pregnancy, suspicion of fetal CNS anomalies, a previous US report implying a fetal CNS anomalies to be confirmed, or for a positive familial history of fetal CNS anomalies. All patients were scanned by an expert operator in a referral center, by a Hitachi 525 sonography machine. Every US scan took about 15 minutes. Fetuses' brains were scanned in axial, coronal and sagittal sections through most important anatomic areas; *i.e.*, transventricular, transthalamic and transcerebellar planes.^{2,3,5,10,11}

Exclusion criteria consisted of gestational age under 18 weeks and ambiguous appearance in scanned planes. Ambiguous appearance might be due to upper gestational age (often >24 weeks of gestation) that include lower mobility of fetus and diminished amniotic fluid, and also oligohydramnium.

All delivered fetuses were followed by calling their mothers and asking about the child health. Alive and still birth newborns were followed by obstetrician/gynecologist experts and pediatricians visit reports. Those fetuses suspected with anomalies in the fetal period were scanned by sonography after birth to confirm the prenatal diagnosis.

Since the pregnant women were referred to so-

nographer by obstetricians/gynecologists, these physicians were not blind to the results of US scans. However, they made their diagnosis after the birth. So it seems that blindness is not helpful in this study. Data were analyzed by a 2×2 contingency table and test specifications were calculated.

Results

In US examination of these 3012 pregnant women, 36 fetuses had CNS anomalies, 26 (72.2%) had one and 10 (27.8%) had two anomalies (Table 1) (Figs. 1 and 2).

It was found that the accuracy of US in detection of CNS anatomic anomalies of fetuses after 18 weeks of gestation was 100% (95% CI: 99.88%–100%). Furthermore, the test had a sensitivity of 100% (95% CI: 90.26%–100%), a specificity of 100% (95% CI: 99.88%–100%), and positive predictive and negative predictive values of 100%.

Discussion

Care of handicap and disable persons is a serious healthcare burden on communities. Antecedent diagnosis of congenital defects is of great help to health economy.

Several studies have shown US as an accurate and non-invasive diagnostic test for determining prenatal CNS anomalies. The calculated accuracy for US varies from 70% to 100%.^{4,7-9} In one study, the accuracy of US in detection of anencephaly in fetus was 100%.^{2,8} In another study from New Zealand, 7880 pregnant women were scanned by US in 16–20 weeks of gestation and the accuracy of US in detecting fetal CNS anatomic anomalies was found to be 92%.⁴ Also, in another study, an accuracy of 96% was reported for US in detection of fetal CNS anatomic anomalies.³ Finally, in a case-control study, the accuracy of US in detecting neural tube defects (NTD) was found to be 90%.⁴ The difference in the reported values may arise in proficiency of sonographer and gestation week in which the US scan was carried out.

US scan for detecting fetal CNS anomalies is best to be done after 16–22 weeks of gestation, according to different references—though, some anomalies such as anencephaly can be detected earlier. In this study,



Fig. 1. A fetus of 18 weeks of gestation which shows bilateral choroid plexus cysts. The cysts disappeared in follow up scans.

women with gestational age of 18 weeks or more were scanned for detecting prenatal CNS anomalies according to team consensus and experience.

Before 18–22 weeks of gestation, the normal and more recognizable posterior fossa structures of the fourth ventricle, cisterna magna, cerebellar vermis, and cerebellar hemispheres are not present. Because the vermis is not fully developed early in gestation, the caudal portion of the fourth ventricle is covered only by a thin ventricular roof, and thus, sonographically, it may appear as though there is communication between the fourth ventricle and cisterna magna. This finding later in gestation suggests the presence of Dandy-Walker malformation.^{8,10}

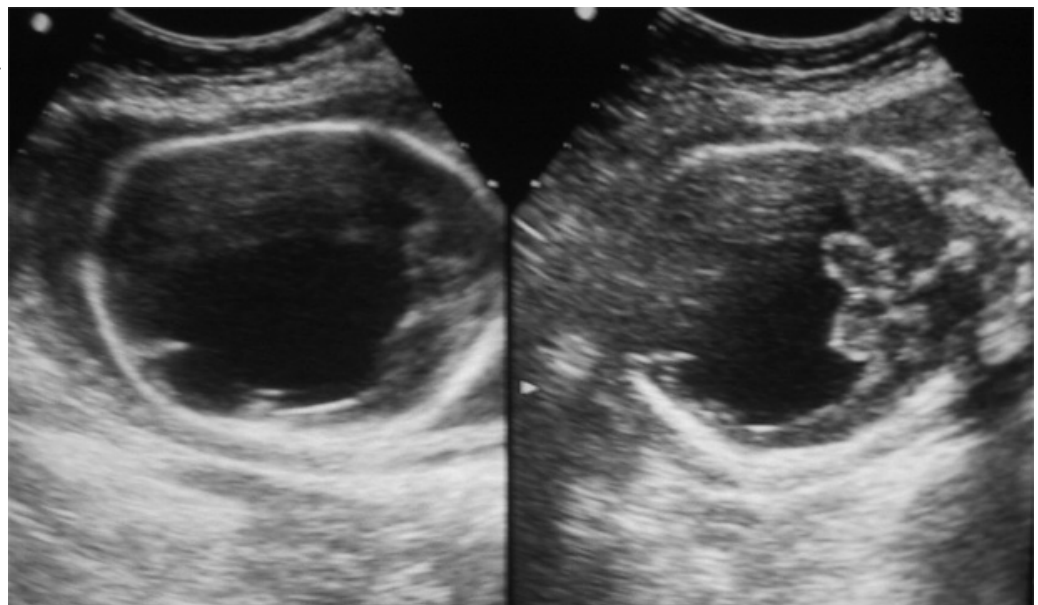
Before the 13th week of gestation, the choroid plexus normally fills the entire lateral ventricles. At

approximately 13 to 15 weeks of gestation, as the choroid plexus assumes its more normal posterior location, the anterior horns of the lateral ventricles appear quite prominent. This normal development and the prominence of the anterior horns of the lateral ventricles, now devoid of choroid plexus, may simulate ventriculomegaly, if one is not aware of this developmental process.

Likewise, the corpus callosum first begins to develop at 12th week of gestation but it is not complete until the 18th to 20th week of gestation when the cavum septum pellucidum and the course of pericallosal artery can be demonstrated sonographically, confirming the presence of the corpus callosum. Scans obtained before this time might wrongly suggest the diagnosis of agenesis of the corpus callosum.^{6,7,9}

Restricted fetal movement due to enlargement of fetus and decreased volume of amniotic fluid in late pregnancy may diminish the accuracy of US assessment in detection of CNS anomalies. Moreover, since anomalies in other organs are often associated with CNS anomalies, it is strongly advised to do fetal CNS US scan in case other anomalies were detected. Although in some studies, 16–24 weeks of gestation is mentioned as the best period for US scan, in this study, there we set no upper time limit for US scan. This decision was made for some cultural poorness entangled with some patients who did not respect pregnancy care protocols and their obstetric US scan was deferred to 24 weeks of gestation or later. Thus, more prominent anomalies that are detectable after

Fig. 2. A fetus with 22 weeks gestational age with alobar holoprosencephaly.



the 24th week of gestation were also reported in our study and ambiguous features were excluded according to the exclusion criteria.

As the pregnant women were not followed at the time of delivery, the still birth newborns were not autopsied, hence, the diagnosis was made solely on the prenatal obstetrics US report. It should be noted that in this study, only existence of gross prenatal CNS anomalies was confirmed and the exact characteristics of the anomaly and its associated anomalies could not be detected because of limited follow-up.

Another important point is that US is highly operator-dependent and the operator experience and proficiency may be the most important determinant affecting the results. In addition, the US machine technology is important in detection of abnormalities.

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