



## Original Article

## Frequency of Fetal Central Nervous System Anomalies Detected on Ultrasonography

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## ABSTRACT

The most serious congenital abnormalities are those involving the central nervous system (CNS). Ultrasound (US) examination is a safe and noninvasive method for detecting these anomalies during pregnancy. **Objective:** To find out the frequency of the fetal central nervous system anomalies detected on ultrasonography. **Methods:** It was a cross sectional study conducted on 385 pregnant women using Convenient Sampling Technique. The patients were referred by obstetricians/gynecologists for routine obstetrical scan. Data was collected from two hospital settings, Allama Iqbal Memorial Teaching Hospital, Sialkot and Umer Diagnostics, Sialkot. The study used a high resolution ultrasound probe with a frequency of 3.5 MHz to identify CNS anomalies in 13 embryos. Data were analyzed using SPSS 26.0. **Results:** 13 fetuses with CNS anomalies were found after 385 pregnant women underwent transabdominal sonography. Anomalies included 2(0.5%) ventriculomegaly, 3(0.8%) acrania, 2(0.5%) encephalocele, 2(0.5%) bilaterally present choroid plexus cyst, 2(0.5%) hydrocephalus, 1(0.3%) agenesis of cerebellar vermis along with mega cisterna magna. **Conclusions:** Acrania is most common among all CNS anomalies. Disability and bed rest are the two most serious consequences of major CNS abnormalities, early detection of these conditions is now crucial. Another significant problem is family counseling.

## INTRODUCTION

Central nervous system (CNS) malformations are a common reason of developing delays and neurological impairments [1]. CNS anomalies are frequent and often fatal and develops between 3 and 20 weeks of intrauterine life [2, 3]. Almost all CNS anomalies are caused by an insult during embryogenesis at some point during development [4]. CNS abnormalities represent an extensive range of congenital birth defects, with a frequency of approximately 1% of all births [5]. Many CNS anomalies can be detected using ultrasound in the first and early second trimesters. Some appear or develop in late pregnancy [6, 7]. Solitary or multiple CNS malformations affect approximately 0.61

percent of kids enrolled in a pediatric clinic [8]. CNS anomalies account for perinatal autopsy series, CNS anomalies make up nearly 10% of all congenital malformations, with neural tube defects (NTDs) (45.5%), hydrocephaly (12.4%), and neuronal proliferation disorders (8.8%) being the most frequent. These chromosomal, cerebral, extracerebral, syndromal, and CNS malformations are frequently linked together [9]. Brazil's ideal second trimester ultrasound screening window is between 20 and 24 weeks of pregnancy [10, 11]. Even though second-trimester ultrasound is the gold standard for identifying structural defects, the number of referrals

for ultrasound exams in the first trimester (11–13 + 6 weeks) has increased in developed nations [12]. A previously unknown fetal congenital defect is discovered on the occasion of a regular third-trimester scan [13]. Structural abnormalities that appear or manifest only in late pregnancy and could not have been reliably detected earlier; congenital malformations that were present earlier but went unnoticed despite adherence to first- and second-trimester screening programmes [14]. Fetal cerebral ventriculomegaly is defined as an atrial diameter of more than 10 mm on prenatal ultrasound examination in the 13th to 40th week of gestation. Due to its non-obstructive causes, the ventricles are mildly increased in size. In spite of the fact that mild fetal ventriculomegaly is quite often unintentional and harmless, it can also be linked to genetic, structural, and neurocognitive abnormalities, with results that can range from standard to severely impaired. Normal classifications for fetal ventriculomegaly include slight (10 to 12 mm), modest (13 to 15 mm), and extreme (more than 15 mm) [15]. Acrania is defined by the nonappearance of flat bones in the cranial vault. The cerebral hemispheres are anomalous despite being fully developed. Acrania develops when an ectodermal and mesodermal growth abnormality following neural tube closure. Acrania must have perfect facial bones, a normal vertebral column devoid of fetal skull, and brain tissue capacity amounted to approximately one-third of the brain size corresponding to the gestational age. There are few effective treatments for acrania, which is a uniformly fatal condition [16]. The embryonic exencephalic brain's abnormal vascularization is caused by the cranial neuropore's breakdown to narrow during the 4th week of growth. Following that, a brain continues to be a spongy vascular clump with certain hind brain structures while the nervous tissue degenerates. Meroanencephaly (the existence of physiological neural tissue) is the current name for what was formerly known as anencephaly (brain complete lack). Meroanencephaly is a deadly birth defect [17]. A congenital NTD known as an encephalocele occurs when a bone defect causes a pouch comprising the brain, the meninges, and CSF to form outside of the cranium. Trauma, tumours, or iatrogenic injury can all cause acquired encephaloceles. Meningocele is the term for the capsule that results from the protuberance of the meninges and cerebral spinal fluid; however, when it contains brain tissue, it is called an encephalocele. However, both are commonly referred to as encephaloceles [18]. The prosencephalon, or embryo's forebrain, neglects to split into two distinct lobes in between the third and fourth weeks of pregnancy, resulting in a birth defect. This method causes the cerebral hemispheres to be separated to varying degrees [19].

## METHODS

It is a cross sectional study conducted on 385 pregnant women using Convenient Sampling Technique, who underwent a routine obstetrical scan after being referred by obstetricians or gynaecologists. Data was collected from two hospital settings, Allama Iqbal Memorial Teaching Hospital, Sialkot and Umer Diagnostics, Sialkot in the time period of January 2022 to June 2022. The CNS anomalies was diagnosed in 13 fetuses utilizing a high-resolution ultrasound probe of 3.5 MHz (Toshiba Xzario 500, Honda Electronics HS-2200). Data were analyzed using SPSS 26.0. Each ultrasound examination took about 15 to 20 minutes. The brains and spinal canals of fetuses were thoroughly scanned in all possible planes.

## RESULTS

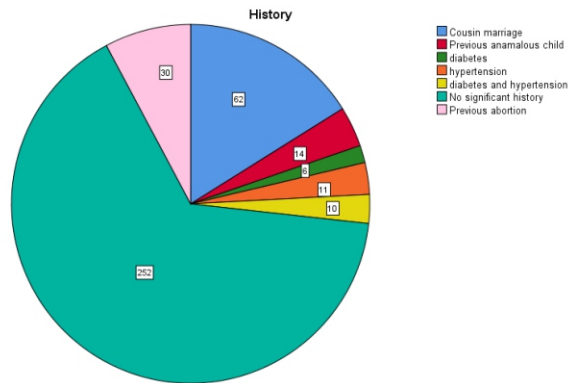
Following transabdominal sonography of 385 expecting females out of which 8 were twin pregnancies, 1 was triplet and 376 were single, 13 pregnancies were found to have CNS anomalous behavior, some fetuses exhibit multiple abnormalities and some fetuses have abnormalities other than CNS Anomalies. CNS Anomalies included 2 (0.5%) ventriculomegaly, 3 (0.8%) acrania, 2 (0.5%) encephalocele, 2 (0.5%) bilaterally present choroid plexus cyst, 2 (0.5%) hydrocephalus. Agenesis of cerebellar vermis along with mega cisterna magna was seen in 1 case (0.3%). Some other abnormalities were seen such as Multicystic kidney (right) in 1 case, Ovarian teratoma in 2 female fetuses, Cystic hygroma in 2 fetuses, Omphalocele in 1 fetus, Esophageal ectasia in 1 fetus. Hydrops fetalis along with cystic hygroma was seen in 1 case. 3 intrauterine demise (IUD) cases were seen. Out of 8 twin pregnancies 1 case was seen with IUD of fetus number 2.

| CNS Abnormalities             | Frequency (%) |
|-------------------------------|---------------|
| Ventriculomegaly              | 2 (0.5%)      |
| Acrania                       | 3 (0.8%)      |
| Encephalocele                 | 2 (0.5%)      |
| Agenesis of Cerebellar Vermis | 1 (0.3%)      |
| Choroid Plexus cyst           | 2 (0.5%)      |
| Hydrocephalus                 | 2 (0.5%)      |
| Mega cisterna magna           | 1 (0.3%)      |
| Other findings                | 16 (4.2%)     |
| Normal Cases                  | 356 (92.5%)   |
| Total                         | 385 (100%)    |

**Table 1:** Frequency of fetal CNS Anomalies

| No. of fetuses | Frequency (%) |
|----------------|---------------|
| Single         | 376 (97.7%)   |
| Twin           | 8 (2.1%)      |
| Triplet        | 1 (0.3%)      |
| Total          | 385 (100%)    |

**Table 2:** Number of fetuses



**Figure 1:** History of Patients

## DISCUSSION

The most prevalent inherited conditions are CNS malformations. The most common central nervous system malformation is neural tube defects, which occur in about 1-2 cases out of every 1000 births [20]. Because of folic acid supplementation, between developed and developing nations, there are differences in the prevalence of CNS anomalies. Furthermore, in one nation, it may follow a different pattern depending on regional food and dietary customs [21]. For example, in our study, 385 pregnancies were evaluated and 13 cases were seen with CNS anomalies. In our study, Acrania was the most common CNS abnormality whereas in a study conducted in Iran on 22500 pregnancies, 112 (0.5 percent) pregnancies were found to have central nervous system defects. They concluded that the most common central nervous system congenital anomalies in Iran, East Azarbaijan, were Chiari malformation and hydrocephalus [22]. In our study we found that fetal brain develops throughout the pregnancy, Fetal CNS irregularities can be detected in late second and third trimester whereas in a study conducted in Israel on 840 pregnant females and 47 fetuses were diagnosed with CNS anomalies. They concluded that some CNS anomalies can only be detected in the end of second and third trimesters so, fetal CNS assessment should be considered when a third trimester scan is performed on patients for any reason [1]. While in another study in India on 7485 pregnant females and 24 cases were diagnosed with Central nervous system inconsistencies, the incidence of CNS anomalies detected by ultrasound was 0.31 percent. It has been decreasing over the years [6]. In China 2571 pregnant women were scanned and 14 fetuses were diagnosed with CNS anomalies [23]. In current study detected CNS anomalies included 2(0.5%) ventriculomegaly, 3(0.8%)acrania, 2(0.5%)encephalocele, 2(0.5%) bilaterally present choroid plexus cyst, 2(0.5%) hydrocephalus, 1(0.3%) agenesis of cerebellar vermis along with mega cisterna magna, whereas the study conducted in Saudi Arabia on 22880 pregnant women. They

diagnose CNS anomalies in 181 fetuses. There were 95 cases of ventriculomegaly (52.2%), 31 cases of neural tube defects (17.03%), 17 cases of cisterna magna (9.34%), and 12 cases of acrania (6.6 %) [24]. In the current study we concluded that early diagnoses of CNS abnormalities is the critical issue due to the most severe complications, Family counseling is really very important in such cases. In Turkey (Istanbul) 15000 expectant mothers in total were scanned, they diagnose CNS anomalies in 41 fetuses. They concluded that disability and bed rest are the two most serious side effects of major CNS anomalies, prognosis and early detection of these conditions have become crucial issues. On the other hand, fetal prognosis and family therapy are significant issues. Parents must decide whether or not to keep having children [25].

## CONCLUSIONS

Deficiency of folic acid, intake of drugs/medication during pregnancy can lead to the fetal CNS abnormalities. In our study, Acrania was the most common among all other CNS anomalies. Important issue is the early detection of CNS abnormalities due to the severe complications, such as disability and bed rest. Another significant problem is family counseling. Parents must decide whether or not to continue their pregnancies.

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