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Role of 3D / 4D Ultrasound in Assessment of Fetal CNS Congenital Anomalies

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Abstract: Background: Congenital abnormalities account for 20-25% of perinatal deaths. Central nervous system anomalies are often severe and are the most common indications for therapeutic abortions. Objectives: The aim of this study is to verify the role of 3D, 4D ultrasonography in prenatal assessment of anatomical structure of central nervous system and early diagnosis of the CNS congenital anomalies. Patients and Methods: From January 2018 to April 2019, 30 pregnant women range from 18 to 40 years old which suspected to have CNS congenital anomalies during routine prenatal obstetric examination, then referred to do 3D / 4D detailed ultrasonography after an initial detailed diagnostic 2D ultrasonography to confirm the diagnosis. 30 pregnant women had undergone detailed anomaly scan in an outpatient private clinics and Ghamra hospital using Chison i3 ultrasound machine and Voluson p8 with both curvilinear 3.5 MHz 60-mm curved probe and 3D real time volumetric 4.5 MHz probe. Results: CNS anomalies occur in significant frequency and some of them are incompatible with life whereas others are life limiting. Ultrasound is an effective investigation for in-utero screening for anomalies including CNS. Ultrasound imaging in antenatal period practically gives an anatomical record of the developing fetus. Confirmation of the anomalies by 3D / 4D ultrasonography definitely helps in increasing accuracy of diagnosis and better counseling of the patient. Additionally, it provides very useful educational information. This also provides psychological benefits for some patients by confirming the reality of fetal anomalies. Conclusion: Three dimensional ultrasonography play an important role in detection and diagnosis of some fetal central nervous system anomalies which may be difficult to be detected by two dimensional ultrasonography due to position of the fetus. For example, the acquisition of the median plane may be impossible with 2D transabdominal ultrasound and require special expertise in transvaginal-transfontanelle scanning, as well as prolonged examination time.
[Fatma Salah El-Dein Mohammed, Wafaa Raafat Abdel Hamid, Ahmed Bassiony Bassiony Elsayed. **Role of 3D / 4D Ultrasound in Assessment of Fetal CNS Congenital Anomalies.** *Nat Sci* 2019;17(11):154-157]. ISSN 1545-0740 (print); ISSN 2375-7167 (online). http://www.sciencepub.net/nature. 18. doi:10.7537/marsnsj171119.18.

Keywords: 3D / 4D Ultrasound, Fetal CNS Congenital Anomalies

1. Introduction

Congenital abnormalities account for 20-25% of perinatal deaths.

Central nervous system anomalies are often severe and are the most common indications for therapeutic abortions ***(Bornstein et al., 2014).***

Central nervous system (CNS) malformations are the second most frequent category of congenital anomaly, after congenital heart disease ***(Monteagudo et al., 2017).*** Approximately 21% of congenital malformations involve the CNS, constituting one of the most common congenital defects and may occur either isolated or in association with other malformations of the CNS itself or other systems ***(Barros et al., 2012).***

The CNS develops from 3 to 20 weeks of intrauterine life. Almost all CNS anomalies are a result of the insult in embryogenesis at some point of development. Ultrasound can diagnose many CNS anomalies in first and early second trimester. Some develop or become apparent in late pregnancy. The earlier the detection, the more time available for the clinician and parents to plan the outcome of pregnancy. Lethal and severely life limiting disorders warrant early termination of pregnancy, whereas detection of minor anomalies helps everybody to be prepared for postnatal management ***(Bornstein et al., 2008).***

Prenatal diagnosis uses various noninvasive and invasive techniques to determine the health condition of the fetus or any abnormality in an unborn fetus. Techniques of fetal visualization are:

a) Non invasive techniques; Ultrasound (US), fetal echocardiography, Magnetic resonance imaging (MRI).

b) Invasive techniques; Embryoscopy, Fetoscopy.

US examination is an effective modality for prenatal diagnosis of these anomalies. It 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 ***(Monteagudo et al., 2017).***

The current study advocates performing a CNS targeted 3D / 4D ultrasonography after an initial diagnostic 2D ultrasonography.

Aim of the Work

The aim of this study is to verify the role of 3D, 4D ultrasonography in prenatal assessment of anatomical structure of central nervous system and early diagnosis of the CNS congenital anomalies.

2. Patients and Methods

From January 2018 to April 2019, 30 pregnant women range from 18 to 40 years old which suspected to have CNS congenital anomalies during routine prenatal obstetric examination, then referred to do 3D / 4D detailed ultrasonography after an initial detailed diagnostic 2D ultrasonography to confirm the diagnosis.

30 pregnant women had undergone detailed anomaly scan in an outpatient private clinics and Ghamra hospital using Chison i3 ultrasound machine and Voluson p8 with both curvilinear 3.5 MHz 60-mm curved probe and 3D real time volumetric 4.5 MHz probe.

Inclusion criteria:

- Age of 18 to 40 years old pregnant women.
- Suspected pregnant women to have CNS congenital anomalies during routine prenatal obstetric evaluation.

Exclusion criteria

- No women with suspected fetal neurological anomalies were excluded from the study.

All pregnant women underwent a detailed 2D diagnostic ultrasonography followed with 3D / 4D ultrasonography including:

1. A complete obstetric medical history

Includes personal history: name, age, special habits, history of previous miscarriage, history of previous CNS fetal anomalies, number of previous conceptions, medical illness (HTN or Diabetic), drug intake or radiation exposure during the first trimester.

2. A full detailed 2D diagnostic ultrasonography including fetal biometry and multiple 2D views for the fetal head as detailed before.

3. 3D / 4D real time ultrasonography to confirm the diagnosis.

Statistical Analysis

Data were collected, revised, coded and entered to the Statistical Package for Social Science (IBM SPSS) version 23. The quantitative data were presented as mean, standard deviations and ranges when parametric. Also qualitative variables were presented as number and percentages. So, the p-value was considered significant as the following: $P > 0.05$: Non significant (NS), $P < 0.05$: Significant (S), $P < 0.01$: Highly significant (HS).

3. Results

The results are shown in Tables 1, 2, 3.

Table 1: Distribution of cases as per 3D detailed ultrasound diagnosis:

Normal cases	**3**
Positive cases with CNS fetal anomalies	**27**

Table 2: Distribution of cases with CNS congenital anomalies

Anomalies detected by US	**Number**
Anencephaly	3
Occipital encephalocele	2
Spina bifida (meningocele)	1
Congenital Scoliosis	1
Agenesis of the Corpus Callosum	2
Holoprosencephaly	4
Dandy-Walker malformation	3
Chiari malformation	1
Schizencephaly	1
Hydranencephaly	1
Microcephaly	2
Hydrocephalus	2
Choroid plexus cyst	2
Aneurysm of the vein of Galen	1
Arachnoid cyst	1
Total	**27**

Table 3: Trimester wise distribution

Trimester	**Number**
First	0
Second	22
Third	8
Total	**30**

4. Discussion

Many studies were performed to evaluate the role of 3D ultrasound in assessment of fetal CNS congenital anomalies.

As regarding previous studies demonstrated that 3D ultrasonography is effective in assessment of fetal CNS anomalies, In our present study the detection of CNS anomalies on ultrasound was 90 % which is more than other studies in the literature ***(Aiyappan et al., 2014)*** [Table 4].

The increasing incidence of detection of CNS congenital anomalies in recent study may be explained by increased awareness amongst treating physicians and the progress and evolution of three-dimensional (3D) ultrasound technology over the last years, although our study is concised only on 30 pregnant women.

Table 4: Detection of CNS anomalies by 3D detailed anomaly scan

Study	Year	Percentage
Carroll et al.,	2000	77 %
Yeo et al.,	2002	80 %
Szigeti et al.,	2007	85 %
Present study	2019	90 %

Early detection of anomalies especially in first and early second trimester helps in planning interventions and further management. In the present study, average gestational age for ultrasound diagnosis of CNS anomalies was 23 weeks. It was probably due to late visit of the pregnant women to the hospital, as majority of the patients under this study was lacking proper health awareness of monitoring at early pregnancy. All the women diagnosed with fetal CNS anomalies were undergoing 3D detailed ultrasound after initial 2D detailed evaluation.

As regarding our study Holoprosencephaly was the most prevalent malformation (4/30 with percentage 13.33 %), followed by anencephaly (3 /30 with percentage 10 %) Dandy-Walker malformation (3/30 with percentage 10 %) and hydrocephalus (2/30 with percentage 6.66 %), other study ***(Hadžagić-Ćatibušić et al., 2008)*****,** had total number of 127 different CNS malformations. The spectrum of CNS malformations was: neural tube defects 49/127 (38,6% of all CNS malformations), hydrocephalus 34/127 (26,8%), microcephaly 24/127 (18,9%), agenesis of corpus callosum 10/127 (7,9%), Dandy Walker malformation 6/127 (4,7%) and other CNS malformations 4/127 (3,1%). The most frequent CNS malformations was neural tube defect and hydrocephalus that was present in 83/127 of patients (65%). Hydrocephalus is one of the most frequent CNS malformation in both studies.

CNS anomalies occur in significant frequency and some of them are incompatible with life whereas others are life limiting. Ultrasound is an effective investigation for in-utero screening for anomalies including CNS. Ultrasound imaging in antenatal period practically gives an anatomical record of the developing fetus. Confirmation of the anomalies by 3D / 4D ultrasonography definitely helps in increasing accuracy of diagnosis and better counseling of the patient. Additionally, it provides very useful educational information. This also provides psychological benefits for some patients by confirming the reality of fetal anomalies.

Despite extensive epidemiological studies, the etiology of CNS malformations remains obscure in most cases. Women, who previously have had a child with a neural tube defect, have an increased risk of recurrence as reported in our study an early diagnosis of CNS malformations allows a precise prognosis to be made. The efficiency of ultrasound screening in the prenatal diagnosis of NTDs has been demonstrated. The important component of better detection rate of CNS malformations is the improvement in ultrasound technology. The development of techniques for prenatal diagnosis of fetal malformations has raised considerable ethical and practical problems, because of elective terminations. 3D detailed Ultrasonography is used nowadays as a routine procedure for the detection of fetal malformations. It is used for pregnant women due to its efficiency, availability, low cost and real time capability. In our study CNS malformations have been detected prenatally in 90 % of patients. Similar results, from the study of NTD have been published, with detection rate of 80 %. Prenatal diagnosis of brain malformations has improved with the advances of 3D ultrasonography imaging techniques The information obtained has significant implications for parental counselling regarding both the type of malformation and neurological and developmental prognosis. Hydrocephalus can be effectively managed by ventriculo-peritoneal shunt or other shunting operations. But, even in such cases, neurodevelopmental disorders may persist after successful operation as diffuse brain abnormalities may be present in addition to hydrocephalus. Up to now, few studies for CNS Congenital Malformations has been established in our country, although the first steps were made. In our study There were additional minor associated anomalies in two cases (ventriculomegaly associated with Dandy walker malformation and corpus callosum agenesis) ***(Rumack et al., 2011).***

Discovering minor abnormalities is important as they can be associated with a syndrome. With the present level of ultrasound the false positive diagnosis is extremely rare.

Conclusion

Three dimensional ultrasonography play an important role in detection and diagnosis of some fetal central nervous system anomalies which may be difficult to be detected by two dimensional ultrasonography due to position of the fetus. For example, the acquisition of the median plane may be impossible with 2D transabdominal ultrasound and require special expertise in transvaginal-transfontanelle scanning, as well as prolonged examination time. Obtaining the median plane, which is important in evaluating the integrity of the corpus callosum and in diagnosing complete or partial agenesis and evaluating the entire lateral ventricles, can easily be done by manipulation of 3D-ultrasound volume by aligning all three planes according to the previously described protocol and placing the marker dot in the center of the coronal plane.

Two dimensional ultrsonography remains the gold standard in assessment of fetal anomalies, and the three dimensional ultrasonography therefore, is not a screening technique but an adjunct to two dimensional ultrasonography for those fetuses in whom malformations are already determined or suspected on the basis of standard sonography, newer ultrasound techniques like 3D and 4D ultrasound are used in diagnosis of birth defects.

9/3/2019

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