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THE ROLE OF 2D ULTRASOUND IN DETECTION OF CNS ANOMALIES

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INTRODUCTION

The main purpose of prenatal diagnosis is to gather genetic, anatomical, biochemical and physiological information about the fetus to detect potential abnormalities that may have impacts both during the fetal period and after birth. Thus, we can provide families with information, genetic counseling, and/or therapeutic alternatives for any anomalies detected. [(1)]

Detection rate of congenital anomalies is about 28% in private practice and hospitals, 60 to 80% in Ob/Gyn's ultrasound labs.[(2-4)] While congenital defects constitute 3% of all births, monogenic disorders and chromosomal syndromes constitute 1.4% and 0.6% of all births respectively.[(5)]

During the last 25 years, the development of increasingly sophisticated equipments (digital techniques, grey scales, color Doppler and 3D and 4D sonography) enabled the diagnosis of a growing number of malformations so that it is now possible to diagnose about 80% of congenital abnormalities with reliable structural images.[(6-9)]

No doubt that ultrasound provides many clinical advantages. The Cochrane database confirms that ultrasound enables the earlier detection of fetal malformations.[(7,10)] Parents seek reassurance about the absence of fetal congenital anomalies and overall fetal health. Therefore, people view routine ultrasound as a part of obstetrical care, capable of filling important gaps by delivering much key information for improving obstetrical practice. [(11)]Fetal anomalies screening (FAS) requires higher education and qualifications than obstetrical ultrasound.[(12)]In most European countries approximately 98% of pregnant women are examined by ultrasound, frequently two to three times (usually once per trimester).

Progressive improvements in ultrasound equipment within the field of prenatal diagnosis of structural fetal anomalies have permitted to obtain a sensitivity range of 90 to 95 percent and specificity range of 95 to 100 percent, with utilization of high definition equipment and an expert sonographer in fetal dysmorphology.[(13-15)]Although some brain anomalies are only visible late in gestation, there is a strong tendency towards a more detailed neurosonogram in the second or even first trimester of pregnancy. 3D ultrasound is a valuable tool in detailed structural analysis of the brain.

Three-dimensional ultrasound has been proposed as a potentially valuable tool for the examination of the fetal brain and for the prenatal diagnosis of intracranial anomalies. Benefits would include: (1) the ability to define the severity, location, and extent of central nervous system anomalies [(16-18)] ;(2) the possibility of reconstructing and visualizing the corpus callosum in the sagittal plane from volume data sets acquired with transverse sweeps through the fetal head[(19)];(3) the use of rendering and rotation techniques in volume data sets acquired with color or power Doppler imaging to improve visualization of cerebral blood flow [(18,20–23)]; (4)the possibility of increasing the speed of fetal neurosonography performed by 2D transvaginal ultrasonography and, at the same time, obtaining tomographic planes of section comparable with those that can be obtained by CT or MRI[(18)]; and (5) the possibility of visualizing the 3 horns of the ventricular system in a single plane (3-horn view).[(24)]

Also 3DUS is a valuable tool for examining the fetal spine, for it uses multiplanar display, volume rendering with the maximum-intensity projection mode (also known as skeletal mode), or a combination of both methods.[(25,26,27-29)].Volume rendering with maximum-intensity projection allows clear depiction of bony structures and, depending on the gestational age of the fetus, visualization of the entire spine in a single image[(25,28)] .Additional features that improve the characterization of spinal anomalies include the possibility of rotating the volume data set and visualizing the spine from multiple perspectives.[(28)] Several investigators have reported on the prenatal diagnosis of anomalies affecting the fetal spine by 3DUS, including scoliosis, hemivertebrae, and neural tube defects.[(3,25,28)] Other applications have included the measurement of the size and volume of the vertebral bodies, spinal canal, and spinal length.[(30-34)] .Three-dimensional ultrasound has also been shown to be useful as an adjunctive modality to determine the level of the defect in cases of spina

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bifida.
[25,26,29,35,36].

MRI is a complementary method to ultrasound (US), useful for fetal assessment, which is helpful in formulating prognosis and perinatal management and can detect occult abnormalities in up to 50% of cases for certain indications.[37]Fetal MRI offers several advantages over prenatal US. It has higher contrast resolution, is not affected by the shadowing from the calvarium or by low amniotic fluid volume, and can be easily performed using commercially available ultrafast T2-W sequences. In addition, fetal MRI is particularly helpful in the detection of gyration and neurulation anomalies and disorders of the gray and white matter. However, fetal MRI is limited by fetal motion, the small size of the structure being imaged, and the marked distance between the receiver coil and the structure being imaged. Therefore, fetal MRI is typically not performed before 22 gestational weeks. Because the fetal brain is a dynamic structure, it is important for radiologists to familiarize themselves with the normal appearance of the fetal brain at different gestational ages in order to be better able to identify and characterize abnormalities with fetal MRI. [38-41]

Cerebral malformations are encountered in about 1% of all births [42]. about 0.61% of children admitted to a pediatric clinic present with solitary or multiple central nervous system (CNS) malformations [43]. Nearly 10% of all congenital malformations in perinatal autopsy series are CNS anomalies, among which neural tube defects (45.5%), hydrocephaly (12.4%) and neuronal proliferation disorders (8.8%) are among the most frequently encountered [42,44]. Frequently additional cerebral, extra-cerebral, syndromic and chromosomal malformations are associated [45]. Still in about 60% of cases the etiology of cerebral malformation remains unknown.

Accurate prenatal diagnosis of central nervous system (CNS) abnormalities is essential in counselling parents, as they are the most common developmental abnormalities causing considerable mortality[46]. Advanced sonography combined with methodology of approaching the fetal brain and fetal MRI has improved the assessment of fetal intracranial structure and diagnosis of the prenatal brain abnormalities.[47,48]Prenatal assessment of the fetal central nervous system is very important as anomalies in this region often determine survival, physical appearance and function in society[7,48].

Sonographic guidelines for screening the fetal brain in a systematic way may increase the detection rate [49].Many malformations of the CNS and fetal neural axis can be detected easily and reliably:[48,50-53] agenesis of corpus callosum, anencephaly, arachnoid cyst, cranial tumors, craniosynostosis, Dandy-Walker malformations, ventriculomegaly, hydrocephalus, diastematomyelia, encephalocele, vein of Galen malformation, holoprosencephaly, hydrancephaly, iniencephaly,intracranial hemorrhage, microcephaly, spina bifida, meningomyelocele, Arnold-Chiari malformation and teratomas.[46,48,50–55]

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