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A STUDY OF ROLE OF USG IN PRE-NATAL DIAGNOSIS

Radiodiagnosis

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ABSTRACT

Background: Ultrasound plays a central role in the provision of prenatal screening and diagnosis. Not only is ultrasound key to guiding prenatal diagnostic procedures, but integration of a genetics-based prenatal diagnosis program has been shown to increase the accuracy of diagnosis when compared to ultrasound alone. This study includes a discussion of prenatal diagnosis by sonography and its contribution to the provision of accurate and precise prenatal diagnosis.

KEYWORDS

Ultrasound, Non-Invasive, Pre-Natal, Diagnosis, Role.

Introduction:

According to most studies, 2% to 3% of living newborns have a congenital malformation.[1,2] When considering birth defects noted in the first years of life, this incidence is nearly doubled. With the decline in infant mortality in the United States from infection and malnutrition, congenital malformations are now a leading cause of infant mortality and responsible for greater intensive care nursery admissions.[3] Of those with a recognized cause, approximately 15 % to 20% are autosomal genetic diseases and 20% are cytogenetic in origin. Less than 1% of anomalies are thought to occur owing to teratogenic medications [4]. Some of the remaining defects are associated with other environmental exposures during pregnancy including infectious agents (3%), maternal disease states (4%), mechanical problems (1% to 2%), irradiation, and unknown environmental causes. The remainder are of unknown or complex etiology (multifactorial, polygenic, spontaneous errors of development and synergistic interactions of teratogens).[5] At present, the ideal time to screen for foetal aneuploidy is during the first trimester of pregnancy. This is a marked change in screening policy due to the significant advances which have been made in antenatal screening for fetal chromosomal abnormalities over the past 20 years [6]. In the past, invasive prenatal diagnosis for Down syndrome with amniocentesis or chorionic villus sampling (CVS) was offered only to women of advanced maternal age or those who previously had an affected child [7]. Subsequently, invasive diagnosis was offered to women younger than 35 years of age who had abnormal second trimester multiple-marker serum screening and also to those with abnormal second trimester sonographic signs so-called soft-markers-of Down syndrome. It should be noted that the most efficient multiple-marker screening test in the second trimester is the "quad" screen, comprising alphafetoprotein (AFP), human chorionic gonadotropin (hCG), unconjugated estriol (E3), and inhibin-A. This approach yields sensitivity for Down syndrome of up to 8100, when sonographic dating is used, for a 500 falsepositive rate. However, since the early 1990's, great interest has been directed toward first trimester screening with the use of sonographic and serum screening markers. In a recent survey of perinatologists in the United States, 4600 used nuchal translucency sonography and 27% used the serum markers PAPP-A and hCG during the first trimester to screen for Down syndrome[13]. With the advent of national training programs for nuchal translucency sonography and with the endorsement of many national professional bodies, it is likely that first trimester based screening programs for Down syndrome will become dominant, with only those patients presenting too late for this form of screening being offered second trimester tests [13-15].

In India also similar Standards are now being accepted and the present study puts in a sincere effort to find the most common USG markers that is helpful in the prenatal Diagnosis.

Aims and Objectives:

To find the Incidence of USG markers that is helpful in the prenatal Diagnosis.

Materials And Methods:

This study was done in the Department of Radiology at Travancore Medical College, Kollam

Total subjects which were included for the study was 60.
The patients were routinely scanned in the first trimester and then in the second trimester. In the first trimester the Fetal nuchal translucency, the Nasal Bone, Doppler sonographic evaluation of ductus venosus blood flow and abnormal tricuspid regurgitation were checked. In the Second trimester nuchal fold thickening, echogenic intracardiac focus, shortened long bones, hyperechoic bowel, renal pyelectasis, choroid plexus cysts (CPCS), clinodactyly, and hypoplastic or absent nasal bone were noted.

Patients who showed positivity for the different USG markers were noted and then were referred for triple marker test which is a Gold standard for detecting the different Aneuploidy. The significance of finding two or more markers in the same fetus is calculated.

Results:

Table 1: First trimester Scan (<2mm Nuchal Translucency)

Total	Mean	Standard Deviation
41	1.19	0.51

Table 2: >2 mm Nuchal Translucency (NT)

Total	Mean	Standard Deviation
19	2.1	0.11

Table 3: The Nasal Bone(N), Doppler sonographic evaluation of ductus venosus blood flow (I) and abnormal tricuspid regurgitation®:

Total	Nasal Bone not developed	Ductus Venosus Inverse Flow	Abnormal tricuspid regurgitation
10	6	1	3

Table 4: Co-Relation between Nuchal Translucency with other abnormalities: (Test for Significance)

Nuchal Translucency with other abnormalities found on USG	X Value	P-Value	Significance
11	0.527	0.062	Not Significant

Image 1: USG showing Nuchal Translucency

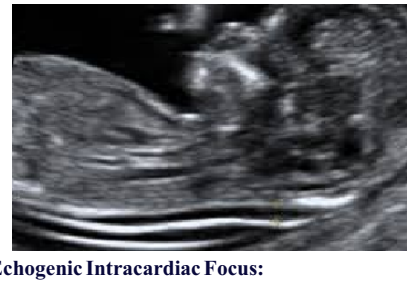

Table5: Echogenic Intracardiac Focus:

Total	Positive	Negative
60	19	41

Image 2: Echogenic Intracardiac Focus:

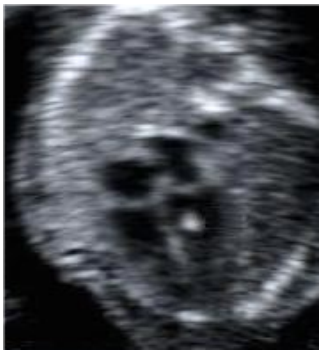

Table 6: Other Malformations Found:

Echogenic Intracardiac Focus	19
Shortened Long Bones,	08
Hyperechoic Bowel,	21
Renal Pyelectasis	03
Choroid Plexus Cysts (CPCS)	01
Clinodactyly,	02
Hypoplastic Or Absent Nasal Bone	06

None of the Malformations found were interrelated significantly with each other as the test for significance for inter – relation came to be insignificant.

Discussion:

The single most powerful marker available today for differentiating Down syndrome from euploid pregnancies is the first trimester sonographic measurement of the fetal nuchal translucency space. Nuchal translucency refers to the normal subcutaneous fluid-tilled space between the back of the fetal neck and the overlying skin. Normally, this space is small; however, in many fetuses with Down syndrome, this space can be significantly increased. By adhering to a standard ultrasonographic technique, it is possible to obtain accurate measurements of this area in the vast majority of fetuses between 10 and 14 weeks gestation (Crown-Rump Length of 36-84 mm). There is a direct correlation between increasing nuchal translucency measurement and risk for Down syndrome, other aneuploidies, major structural malformations, and adverse pregnancy outcome.[7] Septated cystic hygroma is seen in more than one in 300 first trimester pregnancies. In a recent prospective study of routine first trimester sonographic screening, septated cystic hygroma was shown to have a 50% chance of being associated with fetal aneuploidy, with most cases being Down syndrome, as well as cases of Turner syndrome and Trisomy 18.

Nasal bone sonography in the first trimester appears to be a clear association between the absence of the fetal nasal bones on first trimester ultrasound examination and Down syndrome. First trimester Doppler sonographic evaluation of ductus venosus blood flow has been described as an adjunctive test for fetal aneuploidy screening. An association has been suggested between fetal aneuploidy and abnormal tricuspid regurgitation noted during 1st trimester sonography.[7] Although these data are encouraging regarding an association between first trimester tricuspid regurgitation and chromosomal abnormalities, like ductus venosus assessment, it is unclear whether this form of screening will have any role in general population screening.

In the Second trimester ultrasound Nuchal translucency screening will not obviate the need for second trimester ultrasound for the detection of structural fetal abnormalities. The "genetic sonogram," which evaluates for structural malformations and a range of second trimester soft markers for aneuploidy, such as short femurs, echogenic bowel, echogenic intracardiac foci, and increased nuchal fold, has gained widespread acceptability.

Conclusion:

Discovering features of fetal aneuploidy on prenatal sonography and detection rates depend on various factors which includes indications for the scan, gestational age, the population examined and also the quality of the scan.

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