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A STUDY OF ROLE OF USG IN PRE-NATAL DIAGNOSIS IN A TERTIARY HOSPITAL

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] Congenital defects range from enzyme deficiencies caused by single gene defects to complex associations of structural defects. The continuum between purely biochemical abnormalities and structural birth defects includes disorders of structure, function, metabolism, and behavior. Birth defects result from the interaction between the genetic makeup of the embryo and the environment in which it develops. The basic developmental information is encoded in genes, but the genotype is subjected to environmental influences that can impact the observed phenotype. In some cases, the genetic information is expressed regardless of environment, whereas in others, environmental causes interfere with normal development despite a normal genotype. Although some processes are primarily environmental and others primarily genetic, the distinctions between the two are not perfect. Despite considerable advances and research over past several decades, the cause of more than half of human congenital abnormalities remains unknown. 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 scan for foetal malformation is during the first trimester. 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 [7-12]. In a recent survey of perinatologists in the United States, 4600 used nuchal translucency sonography and 27% used the serum markers PAPP-A and human Chorionic Gonadotropin during the first trimester to screen for Down syndrome[13]. With the starting of national training programs for nuchal translucency sonography it is likely that first trimester based screening programs for Down syndrome will become dominant.[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 Great Eastern Medical School and Hospital.

The study was conducted in seventy patients from August to November 2017 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. Enlarged nuchal translucency was noted. 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
32	1.29	0.55

Table 2: >2 mm Nuchal Translucency (NT)

Total	Mean	Standard Deviation
38	2.03	0.21

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
13	9	3	1

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
12	0.625	0.023	Significant

Figure 1:

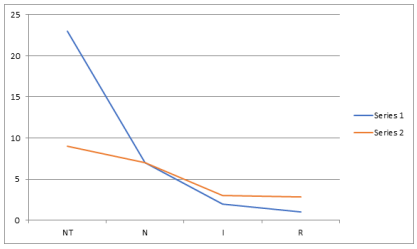

Image 1: USG showing Nuchal Translucency

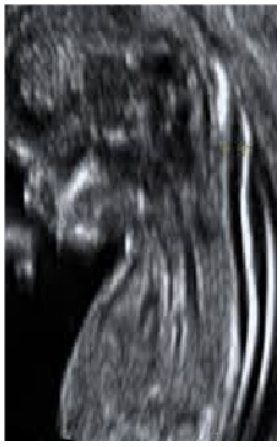

Table5: Echogenic Intracardiac Focus:

Total	Positive	Negative
70	27	42

Image 2: Echogenic Intracardiac Focus:

Table 6: Other Malformations Found:

Echogenic Intracardiac Focus	27
Shortened Long Bones,	03
Hyperechoic Bowel,	16
Renal Pyelectasis	03
Choroid Plexus Cysts (CPCS)	03
Clinodactyly,	02
Hypoplastic Or Absent Nasal Bone	04

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

DISCUSSION:

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. In a study conducted by Cicero et al,[15]. 701 fetuses with increased nuchal translucency were evaluated for the presence or absence of the nose bones during first trimester ultrasonography. The fetal nasal bones could not be visualized in 73% of Down syndrome fetuses (43 of 59) and in only 0.5% of unaffected fetuses (three of 603). The authors also felt that the absence of the fetal nose bone was not related to nuchal translucency thickness and therefore could be combined into a single ultrasound screening modality, with a predicted sensitivity of 85% for a 1% false-positive rate.[15] This study was subsequently expanded to a larger series of 3829 high-risk

First trimester Doppler sonographic evaluation of ductus venosus blood flow has been described as an adjunctive test for fetal aneuploidy screening. Forward triphasic pulsatile ductus venosus flow, Whereas reversed flow at the time of the atrial contraction has been associated with aneuploidy and fetal cardiac malformations.[8]

All pregnancies are theoretically at risk for fetal malformations. Other risk factors include increasing maternal age particularly after 35 years due to higher risk of non-disjunction, abnormal biochemical screening results are also quiet common, history of previous fetal aneuploidy, known balanced translocation which are run in family, or other structural rearrangements in one or in isolated cases where both parents are involved and abnormalities visualized on prenatal ultrasound. In aneuploid fetuses, sonography may reveal gross structural abnormalities, other findings like growth retardation, and also aneuploidy markers. "Soft" USG markers are variations in normal anatomy that, except for their relationship to aneuploidy (especially trisomy 21), are unlikely to be clinically significant. Some of the most common sonographic markers seen in the second trimester include, echogenic intracardiac focus, shortened limb bones, hyperechoic bowel which may be isolated or multi-focal, renal pyelectasis, choroid plexus cysts, clinodactyly, and absent or deformed nasal bone. Structural or major anomalies which include central nervous system anomalies, facial abnormalities, cystic hygroma, diaphragmatic hernia, cardiac defects, gastrointestinal abnormalities, genitourinary anomalies, nonimmune hydrops, and extremity abnormalities. Many fetuses with trisomies 18 and 13 have multiple major structural anomalies which include CVS and CNS anomalies; however, this may not necessarily apply to Down syndrome cases. Only 25% of second trimester fetuses with Down syndrome have ultrasonographically detectable major congenital anomalies.[6] before 20 weeks, structural anomalies were detected by sonography in only 16% to 17% of trisomy 21 fetuses.[17]

CONCLUSION:

Discovering features of fetal malformations on prenatal sonography and detection rates depend on various factors. These factors include indications for the scan, gestational age, the population examined , quality of imaging, criteria for an abnormal scan, the type of chromosomal abnormality, and experience of the examiner.

REFERENCES:

1. Evans MI, Hume RF jr,johnson MP, et al: Integration of genetics and ultrasonography in prenatal diagnosiszjust looking is not enough. AmJ Obstet Gynecol 17421925, 1996.
2. Heinonen OP, Sloane D, Shapiro S: Birth defects and drugs in pregnancy. Littleton, MA, Publishing Sciences Group, 1977.
3. Carlson BM (ed): Human Embryology and Developmental Biology, 3rd ed. St. Louis, Mosby, 2004.
4. Abuhamad A: Technical aspects of nuchal translucency measurement. Semin Perinatol 292376, 2005.
5. Malone FD, Berkowitz RL, Canick JA, D'Alton ME: first-trimester screening for aneuploidy: Research or standard of care? AmJ Obstet Gynecol 182:490, 2000.
6. Nicolaides KH, Heath V, Cicero S: Increased fetal nuchal translucency at 11-14 weeks. Prenat Diagn 222308, 2002.
7. Moscoso G: Fetal nuchal translucency: A need to understand the physiological basis. Ultrasound Obstet Gynecol 5:6, 1995.
8. Malone FD, D'Alton ME, for the Society for Maternal Fetal Medicine: first-trimester sonographic screening for Down syndrome. Obstet Gynecol 10221066, 2003.
9. Snijders RJ, Noble P, Sebire N, et a1: UK multicenter project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10-14 weeks of gestation. Lancet 3512343, 1998.
10. Wapner R, Thom E, Simpson JL, et a1: first-trimester screening for trisomies 21 and 18. N Engl] Med 34921405, 2003.
11. CanickJA, Kellner LH: first-trimester serum screening for aneuploidy: Serum biochemical markers. Semin Perinatol 232359, 1999.
12. Malone FD, Ball RH, Nyberg DA, et a1: first-trimester septated cystic hygroma: Prevalence, Natural history, and pediatric outcome. Obstet Gynecol 1062288, 2005.
13. Molina FS, Avgidou K, Kagan KO, et a1: Cystic hygromas, nuchal edema, and nuchal translucency at 11-14 weeks of gestation. Obstet Gynecol 1072678, 2006.
14. Comstock CH, Malone FD, Ball RH, et al: Is there a nuchal translucency millimeter measurement above which there is no added benefit from first-trimester serum screening? Am J Obstet Gynecol 1952843, 2006.
15. Cicero S, Curcio P, Papageorghiou A, et a1: Absence of nasal bone in fetuses with trisomy 21 at 11-14 weeks of gestation: An observational study. Lancet 35821665, 2001.
16. Nicolaides KH, Spencer K, Avgidou K, et a1: Multicenter study of first-trimester screening for trisomy 21 in 75,821 pregnancies: results and estimation of the potential impact of individual risk-oriented twostage first-trimester screening. Ultrasound Obstet Gynecol 25:221, 2005.
17. Falcon 0, Faiola S, Huggon I, et a1: Fetal tricuspid regurgitation at the 11 + O to 13 + 6-week scan: association with chromosomal defects and reproducibility of the method. Ultrasound Obstet Gynecol 27:609, 2006.