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A Study of Pre-Natal Diagnosis Using USG and Other Mode of Interventional Investigations

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Abstract: ***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. Genetic diseases are often perceived as so rare that the average practitioner will seldom encounter them. However, our increasing knowledge and technologic advances in prenatal diagnosis have demonstrated that this is far from the case. These improvements in prenatal screening and diagnosis mean that many more at-risk couples are able to have unaffected children. Thirty patients who came in for the routine Prenatal Check-up scan were made to undergo USG scanning and the results are reported. This study is done in the Department of OBG, in Yenepoya Medical College, Mangalore. Indications for the scan, gestational age, the population examined (high risk versus low risk), quality of imaging, criteria for an abnormal scan, the type of chromosomal abnormality, and experience of the examiner are all the factors that has to be looked before coming to a final diagnosis.***

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

1. Introduction

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. 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. 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]. 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[7].

This study is intended to find the most common pre natal Diagnosis encountered in USG in the first and second trimester scanning and also to compare the non - interventional technique fare in front of the gold standard interventional techniques.

2. Aims and Objectives

Compare the non - interventional technique fare in front of the gold standard interventional techniques.

3. Materials and Methods

This study was done in the Department of OBG, in Yenepoya Medical College, Mangalore.

The study was conducted with Thirty patients who came to attend the routine first and second trimester check – up.

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.

In the Second trimester nuchal fold thickening, echogenic intracardiac focus, shortened long bones, hyperechoic bowel.

Then the routine interventional study was carried out like

- Chromosomal essay
- Biochemical Essays
- Other marker studies

The USG and the Interventional studies were compared so as to find out the effectiveness of using USG in diagnosing a pre-natal Diagnosis.

4. Results

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

Total	Mean	Standard Deviation
6	1.31	0.57

Table 2: >2 mm Nuchal Translucency (NT)

Total	Mean	Standard Deviation
09	2.07	0.52

Table 3: The Nasal Bone (N)

Total	Nasal Bone not developed
15	2

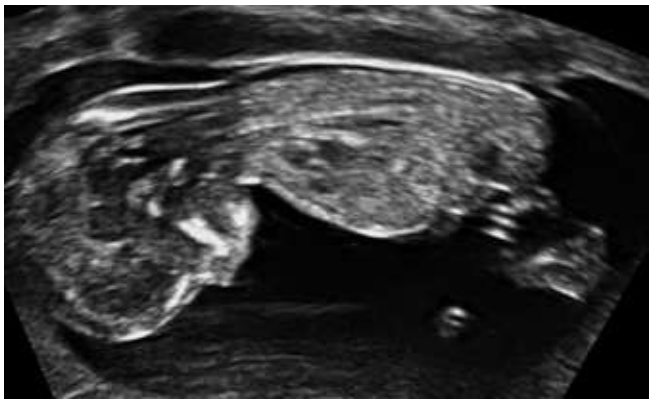

Image 1: USG showing Nuchal Translucency

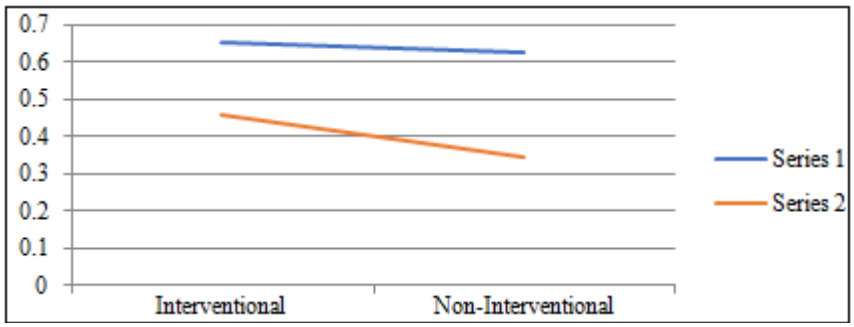

Image 3: Figure to show the co-relation between the interventional and non-interventional Techniques

The above diagram closely shows that the interventional and non interventional (USG) are on the verge of co-existence. Even though the chromosomal studies and other biochemical markers are gold standard tests, USGscreening for the mass has to be employed as in the study it clearly shows that the linear graph is possible if the graph is extended towards the left.

5. Discussion

It is now clear that a subset of fetuses with very large nuchal translucency measurements that have an extremely high risk of fetal aneuploidy or other adverse pregnancy outcomes can be effectively identified in the first trimester. To differentiate 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).

Table4: Echogenic Intracardiac Focus

Total	Positive	Negative
15	02	13

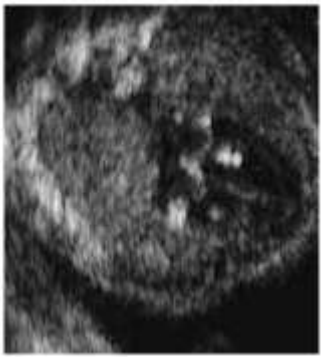

Image 2: Echogenic Intracardiac Focus:

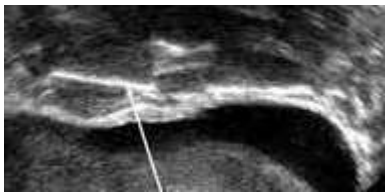

Image 3: Shortened Long Bones

Table 5: Other Malformations Found:

Echogenic Intracardiac Focus	02
Hyperechoic Bowel	07
Shortened Long Bones	06

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. 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.

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.

6. Conclusion

Indications for the scan, gestational age, the population examined (high risk versus low risk), quality of imaging, criteria for an abnormal scan, the type of chromosomal abnormality, and experience of the examiner are all the factors that has to be looked befor coming to a final diagnosis.

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