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A study of pre-natal diagnosis

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Abstract
Background: 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. 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. 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. 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. As the stakes are high and such cases should always be diagnosed as soon as possible a sincere attempt is being made in this study to understand the pre-natal diagnosis using the USG.
Methods: Nine hundred twenty one patients records of scanning were observed out of which thirty patients who were diagnosed to have some malformations in USG scanning are reported. This study is done in the Department of OBG, with the help of Department of Radiology, Kanachur Institute of Medical Sciences, Mangalore.
Results: Out of the observed 2140 patients thirty was observed to have congenital anomalies.
Conclusion: USG is able to detect the anomaly and is the gold standard for screening the patients.

Keywords: Ultrasound, non-invasive, pre-natal, diagnosis, role

Introduction

1% to 5% of living newborns have a congenital malformation based upon different geographical locations [1, 2]. It is now considered to be the most important cause of infant mortality [3]. 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]. The genetic diseases that are known to be accompanied with congenital malformations are often not well understood and has an element of surprise attached to it unless proved otherwise as seen in some familial cases. The raw emotions that run in the family of having a new guest, comes to a sudden halt. The ideal time to scan for foetal malformation is during the first trimester [6]. 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]. As the stakes are high and such cases should always be diagnosed as soon as possible a sincere attempt is being made in this study to understand the pre-natal diagnosis using the USG.

Aims and Objectives
To find the Incidence of USG markers in the first trimester scan.

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Materials and Methods

This study was done in the Department of Radiology at Kanachur Institute of Medical Sciences, Mangalore

Nine hundred twenty one patient's records of scanning were observed out of which thirty patients who were diagnosed to have some malformations in USG scanning are reported. This study is done in the Department of OBG, Srinivas Institute of Medical Sciences, Mangalore.

The study was conducted in seventy patients from November 2014 to April 2015.

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.

Results

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

Total	Mean	Standard Deviation
17	1.38	0.39

Table 2: >2 mm Nuchal Translucency (NT)

Total	Mean	Standard Deviation
13	2.35	0.12

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

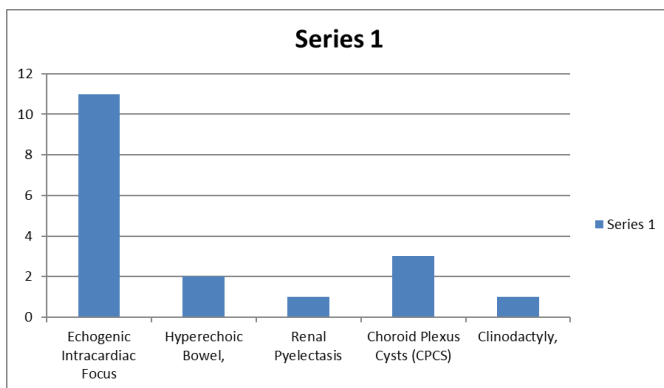| Total	Nasal Bone not developed	Ductus Venosus Inverse Flow	Abnormal tricuspid regurgitation
04	02	01	02

Fig 1: Other Malformations Found:

Table 4: Maternal age

Without congenital anomalies	With Congenital anomalies
26 ±1.38 Years	36±1.84 years

Discussion

In this study twenty one patients had nuchal translucency less than 2mm with a mean measurement of 1.09mm with standard deviation of 0.14mm. In nine patients the nuchal translucency was more than 2mm with a mean measurement of 2.14mm with standard deviation of 0.23mm. In six patients nasal bone was not developed in 2 patients, ductus venosus inverse flow was observed in 1 patient and abnormal tricuspid regurgitation was found in 1 patient. In eleven patients echogenic intracardiac focus was observed, hyperechoic

bowel was observed in 02 patients, renal pyelectasis was observed in one patient, CPCS was observed in three patients and clinodactyly was observed in one patient. The mean maternal age was found to be 34 with a standard deviation of 2.67 years further pointing out the fact towards the increased age can be a cause as suggested by other cases conducted by Malone FD *et al.* [16], and Snijders RJ *et al.* [17].

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

Conclusion

The quality of the scan and the ability of the OBG clinician play an important role in diagnosing the fetal malformations in the first trimester. This study may help in the diagnosis at the local level as it tries to give an image of the local incidence of the different malformations.

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