

A Study of Pre-Natal Diagnosis

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Abstract

Background: 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. 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, Srinivas Institute of Medical Sciences, Mangalore. **Results:** Out of the observed 921 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.

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INTRODUCTION

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 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 ideal time to scan for foetal malformation is during the first trimester⁶. 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.

MATERIALS AND METHODS

This study was done in the Department of Radiology at Srinivas 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 January to June 2019.

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
21	1.09	0.14

Table-2: >2 mm Nuchal Translucency (NT)

Total	Mean	Standard Deviation
09	2.14	0.23

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
06	02	01	01

Table-4: Other Malformations Found

Echogenic Intracardiac Focus	11
Hyperechoic Bowel,	02
Renal Pyelectasis	01
Choroid Plexus Cysts (CPCS)	03
Clinodactyly,	01

Table-5: Maternal age

Without congenital anomalies	With Congenital anomalies
24 ±2.12 Years	34±2.67 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.

REFERENCES

1. Wapner, R., Thom, E., Simpson, J. L., Pergament, E., Silver, R., Filkins, K., ... & Wilson, R. D. (2003). First-trimester screening for trisomies 21 and 18. *New England Journal of Medicine*, 349(15), 1405-1413.
2. Canick, J. A., & Kellner, L. H. (1999). First trimester screening for aneuploidy: serum biochemical markers. In *Seminars in perinatology* (Vol. 23, No. 5, pp. 359-368). WB Saunders.
3. Malone, F. D., Ball, R. H., Nyberg, D. A., Comstock, C. H., Saade, G. R., Berkowitz, R. L., ... & Carr, S. R. (2005). First-trimester septated cystic hygroma: prevalence, natural history, and pediatric outcome. *Obstetrics & Gynecology*, 106(2), 288-294.
4. Molina, F. S., Avgidou, K., Kagan, K. O., Poggi, S., & Nicolaides, K. H. (2006). Cystic hygromas, nuchal edema, and nuchal translucency at 11–14 weeks of gestation. *Obstetrics & Gynecology*, 107(3), 678-683.
5. Comstock, C. H., Malone, F. D., Ball, R. H., Nyberg, D. A., Saade, G. R., Berkowitz, R. L., ... & Carr, S. R. (2006). Is there a nuchal translucency millimeter measurement above which there is no added benefit from first trimester serum screening?. *American journal of obstetrics and gynecology*, 195(3), 843-847.
6. Cicero, S., Curcio, P., Papageorghiou, A., Sonek, J., & Nicolaides, K. (2001). Absence of nasal bone in fetuses with trisomy 21 at 11–14 weeks of gestation: an observational study. *The lancet*, 358(9294), 1665-1667.
7. Nicolaides, K. H., Spencer, K., Avgidou, K., Faiola, S., & Falcon, O. (2005). Multicenter study of first- trimester screening for trisomy 21 in 75 821 pregnancies: results and estimation of the potential impact of individual risk- orientated two- stage first- trimester screening. *Ultrasound in Obstetrics and Gynecology: The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology*, 25(3), 221-226.
8. Falcon O, Faiola S, Huggon I, et al: Fetal tricuspid regurgitation at the 11 + 0 to 13 + 6-week scan:

- association with chromosomal defects and reproducibility of the method. *Ultrasound Obstet Gynecol* 27:609, 2006.
9. Evans, M. I., Hume Jr, R. F., Johnson, M. P., Treadwell, M. C., Krivchenia, E. L., Zador, I. E., & Sokol, R. J. (1996). Integration of genetics and ultrasonography in prenatal diagnosis: just looking is not enough. *American journal of obstetrics and gynecology*, 174(6), 1925-1933.
 10. Heinonen, O. P., Slone, D., & Shapiro, S. (1977). *Birth defects and drugs in pregnancy*. Publishing Sciences Group Inc., Littleton, Massachusetts, USA.
 11. Carlson, B. M. (ed). (2004). *Human Embryology and Developmental Biology*, 3rd ed. St. Louis, Mosby.
 12. Abuhamad, A. (2005). Technical aspects of nuchal translucency measurement. In *Seminars in perinatology* (Vol. 29, No. 6, pp. 376-379). WB Saunders.
 13. Malone, F. D., Berkowitz, R. L., Canick, J. A., & D'Alton, M. E. (2000). First-trimester screening for aneuploidy: research or standard of care?. *American journal of obstetrics and gynecology*, 182(3), 490-496.
 14. Nicolaides, K. H., Heath, V., & Cicero, S. (2002). Increased fetal nuchal translucency at 11–14 weeks. *Prenatal Diagnosis: Published in Affiliation With the International Society for Prenatal Diagnosis*, 22(4), 308-315.
 15. Moscoso, G. (1995). Fetal nuchal translucency: a need to understand the physiological basis. *Ultrasound in Obstetrics and Gynecology: The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology*, 5(1), 6-8.
 16. Malone, F. D., & D'Alton, M. E. (2003). First-trimester sonographic screening for Down syndrome. *Obstetrics & Gynecology*, 102(5), 1066-1079.
 17. Snijders, R. J. M., Noble, P., Sebire, N., Souka, A., & Nicolaides, K. H. (1998). UK multicentre project on assessment of risk of trisomy 21 by maternal age and fetal nuchal-translucency thickness at 10–14 weeks of gestation. *The Lancet*, 352(9125), 343-346.