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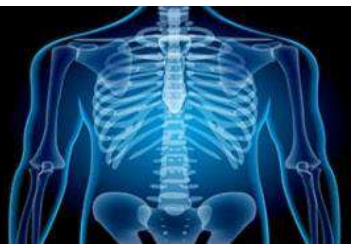

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Fetal anomalies on USG

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Abstract
USG has been the key invention of the last century and has helped tremendously in the science of pre-natal diagnosis of fetal anomalies. According to leading studies across the globe around 1% to 3% of living newborns have a congenital malformation. Although getting exact statistics in our country is a herculean task, it may be more than the developed nations as access to tertiary care is not easy in all parts of the country. This study includes a discussion of prenatal diagnosis by sonography and its contribution to the provision of accurate and precise prenatal diagnosis.

Keywords: Fetal, anomaly, USG, cross sectional study

Introduction
According to leading studies across the globe around 1% to 3% of living newborns have a congenital malformation [1, 2]. The incidence of diagnosing these defects after birth is quite high even in today's world. Congenital malformations are now a leading cause of infant mortality and responsible for greater intensive care nursery admissions [3]. 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. 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 [13-15] dominant. 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. This study puts in an effort to find the role of USG in diagnosis of fetal anomalies.

Aims and Objectives
To study the fetal anomalies that are encountered in USG.

Materials and Methods
This study was done in the Department of Radiology, Kanachur Institute of Medical Sciences, Mangalore. The study was done from Feb 2019 to July 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. 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.

Inclusion criteria

All patients who were diagnosed were included in the study for statistical purposes.

Exclusion criteria

Patients proved otherwise later by other tests were excluded.

Results

Table 1: USG showing nuchal translucency

>2mm nuchal translucency	Mean	Std Deviation
11	2.08	0.19

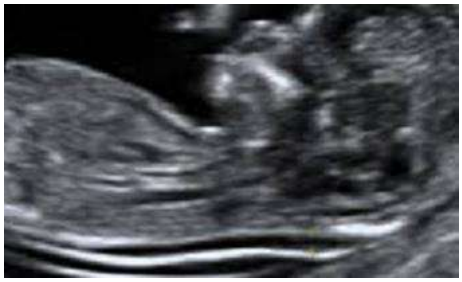

Fig 1: USG showing nuchal translucency

Table 2: Other abnormalities

Conditions	Frequency
Ductus Venosus Inverse Flow	1
Abnormal tricuspid regurgitation	2
Nasal bone under development	7
Hyperechoic Bowel	2
Shortened Long Bones,	1
Clinodactyly,	1
Renal Pyelectasis	1
Echogenic Intracardiac Focus	11

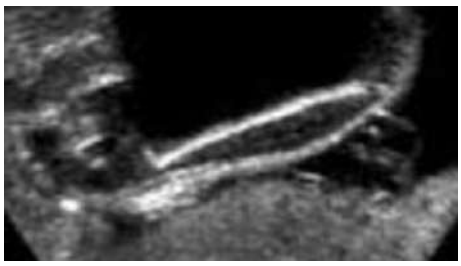

Fig 2: Short femur

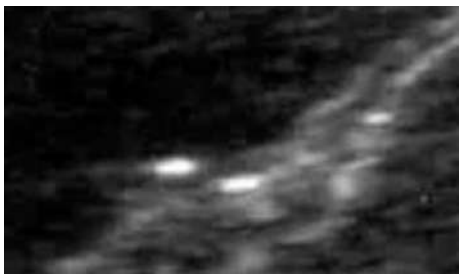

Fig 3: Absent nasal bone (Enlarged image)

Discussion

Congenital malformations were not the leading cause of morbidity and mortality not a long time ago but with advent of USG and its implementation in diagnosing the congenital anomalies, we are able to diagnose more. 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.

All pregnancies should be considered theoretically at risk unless proved otherwise 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 quite 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.

Thus, there are a plethora of fetal defects that can be diagnosed using a USG. This is rather a wonderful opportunity to learn for the budding sonologists and this paper is intended to be helpful for the same.

Conclusion

The experience of the sonologist is very much needed in diagnosing the congenital anomalies. Multiple scans also lead to accurate diagnosis. This is a boon to the society as sensitivity and the specificity of the USG in diagnosing a prenatal deformity seems to be high.

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