



## Original Article

# Role of Ultrasound as a Diagnostic Modality in Detecting Congenital Anomalies

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## Abstract

**Aim of the Study-** *The aim of this study is to assess the role of ultrasound as diagnostic modality in detecting congenital anomalies.*

**Materials and Methods-** *This observational study was carried out in the obstetric unit of a tertiary care centre in Kerala for a period of one year. All the pregnant women who delivered in the hospital after 14 weeks of gestation for period of one year were included in the study. All pregnancies terminated after detection of anomalies by ultrasound were included in the study. All fetuses and new born babies were examined by the pediatrician and sex, birth weight and type of anomaly was noted. Details of anomaly scan were noted.*

**Results-** *During the study period, 15227 babies were born, of which 379 had congenital malformations, making the prevalence 2.48 %. Out of the total malformed fetuses, anomaly was picked up by ultrasound in 161 cases (42.48%). Anomaly was picked up before 24 weeks in 21.7 % cases and after 24 weeks in 78.3 % of the cases. Maximum detection rates were for central nervous system and genitourinary system. Maximum cases detected before 24 weeks were CNS abnormalities and cardiovascular anomalies were totally missed before 24 weeks of gestation.*

**Conclusion-** *The ultrasound detection rates of congenital anomalies have to be improved to have safe termination of pregnancy and to avoid termination in substandard settings.*

**Keywords:** *Congenital anomaly, ultrasound, detection rates.*

## Introduction

The incidence of congenital anomalies is approximately 3-7 % of all pregnancies even though it may vary from place to place <sup>[1,2,3]</sup>. In India the incidence is around 2.5 % <sup>[4]</sup> and accounts for 8-15 % of perinatal deaths and 13-16 % of neonatal deaths in India <sup>[5,6]</sup>. Worldwide second trimester ultrasound scan done between 18- 21 weeks

remain the standard for detection of congenital anomalies. Population screening is offered for chromosomal anomalies and neural tube defects <sup>(7)</sup>. But this has been widely debated <sup>(8)</sup>.

In India, medical termination of pregnancy is allowed only up to 20 weeks of gestation irrespective of the anomaly being lethal <sup>(9)</sup>. We have a large number of cases where congenital

anomalies are detected after 20 weeks of gestation which makes it difficult to terminate the pregnancy. This causes substantial emotional and economic burden on the families and societies and leaves the families and health providers with unanswered questions regarding the causes, recurrence risks and preventive measures.

Hence the study was planned to assess the prevalence of congenital anomalies in a tertiary care centre in Kerala, India and to assess the detection rate of congenital anomalies by ultrasound.

### Materials and Methods

The study was a observational study conducted in the obstetric unit of Sree Avittom Thirunal hospital, Trivandrum, Kerala, India. All the pregnant women who delivered in the hospital after 14 weeks of gestation for period of one year were included in the study. All pregnancies terminated after detection of anomalies by ultrasound were included in the study. All fetuses and new born babies were examined by the pediatrician and sex, birth weight and type of anomaly was noted. Details of anomaly scan were noted.

### Results

Total babies born in the study period were 15,227 [including the second trimester abortions from 14

weeks of gestation]. Total babies with congenital abnormality were 379 (2.48 %).

**Table 1:** Pattern of anomalies observed

SYSTEM	Number	%
Musculoskeletal system	91	24
Central nervous system	81	21.4
Genitourinary system	62	16.4
Gastrointestinal tract	54	14.2
Cardiovascular system	35	9.2
Syndromes	12	3.2
Miscellaneous	44	11.6
Total	379	100

Out of the total malformed fetuses, anomaly was picked up by ultrasound in 161 cases (42.48%), as in table 2..Anomaly was picked up before 24 weeks in 21.7 % cases and after 24 weeks in 78.3 % of the cases.(table 3)

**Table 2:** Ultrasound pickup rate

Ultrasound scan pick up	Number	%
Count detected	161	42.5
Count missed	218	57.5
Total	379	100

**Table 3:** classification according to gestational age at which diagnosis was made

Gestational age	Number (%)
≤24weeks	35 {21.7}
> 24 weeks	126 {78.3}

Out of the 35 patients who had fetus with anomalies detected before 24 weeks, 25 underwent termination of pregnancy.

**Table 4:-** Antenatally reported cases with ultrasound

System	Total no: of cases	Count detected	%	Count missed	%
Central nervous system	81	60	74.1	21	25.9
Gastrointestinal system	54	26	48.1	28	51.9
Cardiovascular system	35	8	22.9	27	77.1
Genitourinary	62	43	69.4	19	30.6
Musculoskeletal	91	9	9.9	82	90.1
syndromes	12	1	8.3	11	91.7
Miscellaneous	44	14	31.9	30	68.1
Total	379	161		218	

Maximum detection rates were for central nervous system and genitourinary system.

**Table 5:-** Classification according to period of gestation at which anomalies were detected (system wise)

System	Up to 24 weeks		Above 24 weeks	
	Number	%	Number	%
Central nervous system	16	26.7	44	73.3
Gastrointestinal tract	2	7.7	24	92.3
Cardiovascular system	0	0	8	100
Genitourinary	8	18.6	35	81.4
Musculoskeletal	2	22.2	7	77.8
Syndromes	0	0	1	100
Miscellaneous	7	50	7	50
Total	35		126	

Maximum cases detected before 24 weeks were CNS abnormalities and cardiovascular anomalies were totally missed before 24 weeks of gestation.

### Discussion

The incidence of congenital anomalies in the study was 2.48 %. 78 newborns (20.58 %) had multiple anomalies. Despite being advised to do an anomaly scan before 20 weeks, many patients did their anomaly scan only by 24 weeks of gestation. It reflects the lack of knowledge of the patients and also the deficit in the health care system.

In this study, out of the 379 cases, anomaly was picked up by ultrasound only in 42.5 % of the cases. Detection rates were highest for central nervous system anomalies (74.1%) and genitourinary system (69.4 %). It was lowest for musculoskeletal (9.9%) and cardiovascular system anomalies (22.9%). According to Crane JP et al, the screening ultrasound has tripled the rate of anomaly detection but reached only 35 %<sup>(15)</sup>. The pickup rate at routine ultrasonographic screening in the Eurofetus Study, in which 2262 malformed fetuses are registered, was 56.2%. Within the subset of major anomalies, detection was high for central nervous system anomalies (88.3%) and urinary tract anomalies (88.5%), and lower for heart and great vessel anomalies (38.8%)<sup>(10)</sup>

Even though the patients who had malformations had their ultrasound done before 24 weeks of gestation, only 21.7 % of the anomalies detected by ultrasound could be identified before 24 weeks. This was similar to another study conducted in India where the ultrasound detection rate of anomalies before 20 weeks of gestation was 33 %

<sup>(11)</sup>. In Eurofetus study, 55 % of the major anomalies could be detected before 24 weeks of gestation. This difference may be due scarcity of good resolution machines, lack of expertise and over work.

The detection of anomalies can definitely affect the rate of termination of pregnancy and hence decreases the number of live births for mothers with congenital anomalies<sup>(10,13)</sup>. The low detection rates can lead to increased perinatal mortality and morbidity. It also causes a lot of anxiety to the family<sup>(12)</sup>. On the other hand RADIUS trial found no statistically significant effect on the rate of induced abortion<sup>(14)</sup>.

Hence it is important that more focus is given in identifying structural malformations before 20 weeks except those conditions which are said to appear further late or reported with confirmation at a later gestational age like few posterior fossa abnormalities, duodenal atresia, and few renal abnormalities.

### Conclusion

In a country like India, where medical termination is possible only till 20 weeks of gestation, anomaly scan between 18- 20 weeks of gestation is essential. The detection rates have to be improved to have safe termination of pregnancy and to avoid termination in substandard settings.

**Sources of support** \_ nil

**Conflicting interest**- nil

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