

# Routine Screening for Fetal Limb Abnormalities in the First Trimester

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## ABSTRACT

**Objective:** To evaluate the routine screening for fetal limb abnormalities in the first trimester.

**Study Design:** Prospective study

**Place and Duration of Study:** Department of Radiology and Department of Obstetrics & Gynaecology, Khairpur Medical College, Khairpur Mir's from 1<sup>st</sup> October 2020 to 30<sup>th</sup> September 2022.

**Methodology:** The study enrolled all pregnant women in visiting the Gynaecology OPD for routine check-up. All the cases were referred to ultrasound diagnostic for screening of fetal limb abnormalities. The total sample size of the study was assigned as 1000 fetuses from 980 women as 10 women had twin pregnancies. Limb reduction shortcomings included transverse-limb reduction deficit as well as longitudinal-limb reduction deficit as seen through absent radius and also club hand, absent fibula phocomelia. The diagnosis of the limb shortening was based on the length of long bone if <5<sup>th</sup> percentile. Lethal skeletal dysplasia suspicion was based on narrow thorax and long bone abnormalities.

**Results:** The mean age of the pregnant women was 27.6±5.3 year. Around 26% of the women were nulliparous. The fetal limb anomaly was presented in 0.9% of the total cases. Within these, 0.7% was those women with single fetus while 0.2% had twin pregnancy. There were 60% of the women who were identified with fetal anomaly on their first screening visit within 1<sup>st</sup> trimester transverse limb anomaly was observed in 2 case within first trimester whereas 2 cases of club foot were identified in first trimester and one in third trimester.

**Conclusion:** The fetal limb anomaly can be feasibly screened within first trimester. It has a prevalence of 0.9% with transverse limb deficiencies and club foot to be presented in highest frequency.

**Keywords:** Fetal limb abnormalities, First trimester, Prevalence

## INTRODUCTION

Congenital abnormalities related with limb may be associated with structural anomalies or with the hereditary syndromes. The prognosis is dependent on the underlying causes. Early detection of fetal anomalies of limb allows ease in further case examination and testing as well as better identification of etiology. This facilitates in parent counselling and opting the right choice for the future of the newborn.<sup>1,2</sup>

The preliminary survey of fetal anatomy was conducted in the 1980s. In late 90s, Timor-Tritsch et al<sup>[2]</sup> performed transvaginal ultrasonography and identified fetal limb anomaly in 9-week pregnancy. The prevalence of limb anomalies is considered as six cases in 10,000 live births with a higher incidence of upper limb cases than lower limbs. Bilateral limb abnormalities are less common with higher tendency of right limb involvement than left.<sup>3</sup> The formation of limb occurs within 4-8 weeks of time. The primary-ossification center is formed in long bones by 12 weeks of gestational time.<sup>4,6</sup>

The limb positioning along embryos craniocaudal-axis is regulated through homeobox (HOX) gene-family whereas outgrowth of limbs is fibroblast growth factor gene dependent in addition to the bone morphogenetic-proteins (BMPs). The anteroposterior axis is patterned by Sonic hedgehog genes (SHH) regulation.<sup>5</sup> Within the various limb abnormalities which can now be identified club foot, Amelia, club hand, radial aplasia, phocomelia and sirenomyelia are most common [7-9]. The present study was designed for identifying the cases of fetal limb abnormalities through ultrasonographic screening. The results of this study provided substantial data on the incidence of fetal limb abnormalities and assisted in counselling parents for taking rightly decisions in context with their newborn health.

## MATERIALS AND METHODS

This prospective study was performed at Department of Radiology and Department of Obstetrics & Gynaecology, Khairpur Medical College, Khairpur Mir's from 1<sup>st</sup> October 2020 to 30<sup>th</sup> September 2022. The study enrolled all pregnant women in visiting the Gynaecology OPD for routine check-up. All the cases were referred to ultrasound diagnostic for screening of fetal limb abnormalities. The total sample size of the study was assigned as

1000 fetuses from 980 women as 10 women had twin pregnancies. The sample size was calculated through WHO sample size calculator using 95% CI and 80% power of test. Cases where examination remained suboptimal and the fetal limb evaluation was incomplete were also excluded from the study. A written informed consent was taken from all the women before referring them to ultrasonography. The ultrasound for fetal anomaly was done on Doppler used ultrasound by a specialized sonologist. The complete time of the diagnostic ultrasonography was 15-20 minutes. Structures like the skull, the choroid plexuses, cerebellum, the orbits, ears, lips, nose, thoracic diaphragm; four-chamber heart, three-vessel view, stomach, abdominal wall, the umbilical cord insertion, and bladder, in addition to all extremities and digits were visualized through ultrasound. A colored Doppler for the hearts 4 chambers, pulse-Doppler of ductus venosus was also performed. Fetus being in challenging position the ultrasound was halted and repeated within 15 min. All ultrasound images were kept in a protected database. Immediate genetic as well as perinatal counselling was delivered to all positive patients with fetal abnormality observed in ultrasonography. Limb reduction shortcomings included transverse-limb reduction deficit as well as longitudinal-limb reduction deficit as seen through absent radius and also club hand, absent fibula phocomelia. The diagnosis of the limb shortening was based on the length of long bone if <5<sup>th</sup> percentile. Lethal skeletal dysplasia suspicion was based on narrow thorax and long bone abnormalities. A well-defined questionnaire was designed for documenting the information regarding maternal demographics, clinical assessments, ultrasound reports, fetal abnormality status and type. Data was statistically analyzed through SPSS-25.

## RESULTS

The mean age of the pregnant women was 27.6±5.3 year. Around 26% of the women were nulliparous. Majority of the women (49%) has a singleton live birth before. The body mass index of 24% women was greater than 4000 grams (Table 1). The fetal limb anomaly was presented in 0.9% of the total cases. Within these 0.7% was those women with single fetus while 0.2% had twin pregnancy (Table 2).

There were 60% of the women who were identified with fetal anomaly on their first screening visit within 1<sup>st</sup> trimester where as 20% were identified in 2<sup>nd</sup> trimester and 10% in the 3<sup>rd</sup> trimester. Transverse limb anomaly was observed in 2 case within first trimester whereas 2 cases of club foot were identified in first trimester and one in third trimester, while sirenomelia and dysplasia was seen in 1 cases each. Malposition of fetal limb was not presented within enrolled fetuses (Table 3). Within the 2 twin pregnancy women, 1 woman presented with transverse limb deficiency in one of the fetuses while club hand was observed in another women having singletons (Fig. 1)

Table 1: Age, Parity and body mass index distribution within cases (n=1000)

Variable	No.	%
Age (Years)	27.6±5.3	
Parity		
0	260	26.0
1	490	49.0
2	220	22.0
≥3	30	3.0
Body mass index		
2500	270	27.0
2500-4000	490	49.0
>4000	240	24.0

Table 2: Fetal limb anomaly within fetuses

Fetal Limb anomaly	No.	%
Yes	9	0.9
Singleton pregnancy	7	0.7
Twin Pregnancy	2	0.2
No	992	99.2

Table 3: Distribution and frequency of fetal limb abnormality within trimesters

Limb defect	Detection rate (n=9)		
	First trimester	Second trimester	Postnatal
Absent radius and club hand	1	-	-
Absent fibula	-	-	-
Phocomelia	-	-	-
Transverse limb deficiency	2	-	-
Clubfoot	2	-	1
Lethal skeletal dysplasia	-	-	-
Sirenomelia	1	-	-
Limb dysplasia	1	1	-
Malposition	-	-	-
Split hand	-	1	-
Deformities of fingers	-	-	-
Total	6	2	1

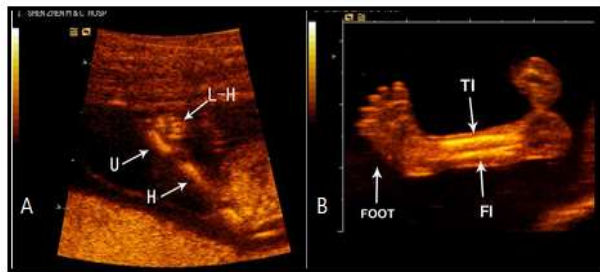


Fig. 1: Transverse limb deficiencies (A: Absence of left radii and club hand. H, humerus; U, ulna; L-H, left hand; L-Leg, B; left leg high-resolution ultrasound imaging presenting T1 as tibia and F1 fibula having short bone length)

## DISCUSSION

The feasibility of the first trimester in evaluation of the fetal limb anomaly has been widely investigated. The current research study results have elaborated the fact that it is the most suitable time for identifying the risk of fetal limb abnormality within pregnant women as majority of the cases were identified within the first trimester. Similar study conducted retrospectively had also reported that 63.9% of the anomalies are detected within the first trimester.

Therefore, suggesting it to be the most feasible time for screening and deciding the outcome of the pregnancy.<sup>10,11</sup>

Within the various types of the fetal abnormalities observed over the globe the most significant are transverse limb deficiencies, club hand and foot or sirenomelia which are often identified in the first trimester. In the present study it was observed that of the total 3 cases of club foot only two out of three were able to be detected in the 1<sup>st</sup> trimester however, the rest one fetus was detected in later trimester. This might be due to the reason of fetus positioning which made identification difficult within early trimester. However the other transverse limb deficiencies identification was in first trimester as similar to the available literature.<sup>12-16</sup>

The imaging of the long bones can be performed at 11<sup>th</sup> week of gestational period, whereas as that of fingers and feet it can be achieved in the late first trimester time. Most efficient method of identification of fetal limb anomalies includes transvaginal scanning through a Doppler supported three dimensional ultrasonography machines. It is recommended that transvaginal sonography be conducted at the time of NT measurement. In other available literature as from South Asian countries transabdominal visualization within first trimester is also acceptable.<sup>17,18</sup> In the current study transvaginal sonography was performed with the consent of the patients and recommendation of the sinologist.

The present study has elaborated 0.9% of the fetal limb abnormality. Studies elsewhere have noted a prevalence within 0.38-0.6% of fetal limb abnormality with result similar to the current study research.<sup>19,20</sup>

## CONCLUSION

The fetal limb anomaly can be feasibly screened within first trimester. It has a prevalence of 0.9% with transverse limb deficiencies and club foot to be presented in highest frequency.

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