

# CASE REPORT

## TRANSVERSE LIMB DEFECT: A CASE REPORT

Chitra Andrew<sup>1</sup>, Natasha Nawani<sup>2</sup>

### HOW TO CITE THIS ARTICLE:

Chitra Andrew, Natasha Nawani. "Transverse Limb Defect: A Case Report". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 64, November 24; Page: 14082-14086, DOI: 10.14260/jemds/2014/3883

**ABSTRACT: CONTEXT:** Transverse limb defects are a relatively rare problem (3.5–6.9/10000 births) with a fairly low prenatal detection rate (55%). The possible etiology for this condition could be as part of a genetic condition or amniotic band syndrome. This rate can be improved with the use of careful anatomical survey in the first trimester. **AIM:** In this case report, transverse limb defect was missed in the first trimester although detailed examination of the limbs was attempted. The defect was subsequently detected in the second trimester. The ways by which guidelines could be better implemented to ensure early detection of anomalies are discussed. **CONCLUSION:** Early detection is desirable as it reduces physical and emotional morbidity if termination is opted for in major anomalies and thus benefits the patient greatly.

**KEYWORDS:** Limb reduction defect, Transverse limb defect, amniotic band syndrome, first trimester screening.

**INTRODUCTION:** The incidence of limb reduction defects varies from 3.5 – 6.9 per 10,000 births. The prenatal detection rate is approximately 55%. However, this varies considerably depending on the application of imaging guidelines, and on the population studied. The defects range from complete absence of limbs (Amelia) to partial absence (meromelia). The commonest limb reduction defect is a terminal transverse defect, which is usually unilateral, isolated, and sporadic in occurrence. This is defined as the partial or complete absence of one or more fetal limbs beyond a certain point, leaving a stump, and is usually categorized according to the last remaining bone segment. This case report describes an isolated transverse limb defect wherein the first trimester scan was thought to show both limbs appearing normal but in the second trimester at the targeted scan, the transverse limb defect was detected. This emphasizes the need for more careful evaluation in the first trimester to pick up these rare anomalies.

**CASE REPORT:** A 29 years, primigravida with complex congenital heart disease (Corrected transposition of great arteries with pulmonary stenosis) and congenital deafness was referred to the fetal medicine unit for a target scan. The patient had undergone LV-PA conduit using 18mm Gore-Tex tube and monocusp pericardial valve at the age of 21 years. On confirmation of pregnancy at 6 weeks a cardiologist opinion was obtained. The first trimester scan done at 11 w+3 showed a single live intrauterine gestation with a CRL of 48mm corresponding to the period of amenorrhea.

The NT was 1.8mm and the combined serum biochemistry was done. The post-test risk was 1 in 895 (screen negative). Detailed examination of the fetus at this scan and was reported as normal.

The image was available with the patient and showed upper limbs of both sides. (Fig. 1) The maternal Echo done at the same time showed no abnormalities in the repairs performed and she was started on Tablet Digoxin 0.25mg at 7 weeks of gestation. Other than this medication, only folic acid was given. Later at 16 weeks, iron and Calcium were added.

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A targeted ultrasound was performed and was consistent with biometry of 19 weeks. The placenta, liquor and fetal activity were normal. On detailed targeted imaging, the right humerus was normal in length and the elbow was imaged. Distal to the elbow, the radius and ulna were seen for a length of 1 to 1.5mm and were absent beyond that length. (Fig. 2, 3) The hand was not imaged. This was also confirmed by 3 d imaging. (Fig. 4) No attachment of amniotic membrane to the distal segment of the upper limb was noted on careful observation. Movements and muscle mass was normal in the proximal segment. The left upper limb was imaged and confirmed to be normal in the proximal, mid and distal segments. Movements of the left upper limb was also normal in shoulder and elbow joints. Hand opening and closure was also seen. The lower limbs were structurally normal in the proximal, mid and distal segments and movements were seen. The other fetal structures and Fetal ECHO were normal.

The patient and her family were counseled in detail on two separate occasions to ensure that all information was understood. The patient could understand the discussions aided by labeled drawings and written words as she was trained in lip reading in English and educated to postgraduate level. The possibilities of prosthesis for the right limb and that the other limb was normal in function thus could be used for writing etc. was emphasized and discussion with plastic surgeons was offered. The family was very positive about the fetus and its potential outcome. However the patient decided that as she had undergone severe emotional difficulties in childhood due to hearing impairment she could not raise a child with physical disability. After the second counseling, termination of pregnancy was opted for.

The patient was induced with two doses of PG e2 gel and expelled the fetus after 8 hours of induction. The fetus weighed 357 grams, and showed transverse defect of the right forearm. (Fig. 5) The left upper limb was normal (Fig. 6) and there were no other external anomalies. The placenta was retained and manual removal was performed. The patient made a full recovery and was discharged from hospital after two days. Due to the manual removal the placenta was piecemeal and could not be further evaluated to confirm amniotic bands or to confirm the portion of limb in the placenta. The family declined perinatal examination which was offered. The couple was counseled that the recurrence rates of transverse limb defects is very low due to its sporadic occurrence. Pre-pregnancy folic acid and first trimester scan in a referral center was advised.

Most cases of limb defects are believed to be secondary to vascular insult occurring very early in embryonic life or amnion rupture sequence. Amnion rupture sequence causes formation of amniotic bands which can cause many different scenarios. A study of 1,010 pre-viable fetuses (9 – 20 weeks developmental age) was performed to determine the result of amnion rupture sequence. 18 fetuses were affected with the incidence of 1:56. Eleven fetuses had limb constrictions and amputations only; 7 fetuses also had non-limb involvement, such as encephalocele, unusual facial clefts, and abdominal defects. In 6 pregnancies, constrictions of the umbilical cord by amniotic bands were the cause of fetal intrauterine death.

Transverse limb defects can also be caused by ingestion of teratogens such as misoprostol which may be used for first trimester termination of pregnancy.<sup>(1)</sup> They can also be caused by maternal ingestion of vasospastic drugs and maternal diabetes, smoking and ingestion of alcohol.<sup>(2)</sup> Amniotic band syndrome in the second trimester has been reported following diagnostic amniocentesis but its spontaneous occurrence is rare.

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This case is reported to emphasize the importance of first trimester scan which should be carefully performed according to standard guidelines.<sup>(3)</sup> The guidelines suggest demonstrating four limbs with three segments and as an option recommend demonstration of normal orientation of hands and feet. The image showed both upper limbs looking apparently normal but the three segments are not individually detailed.

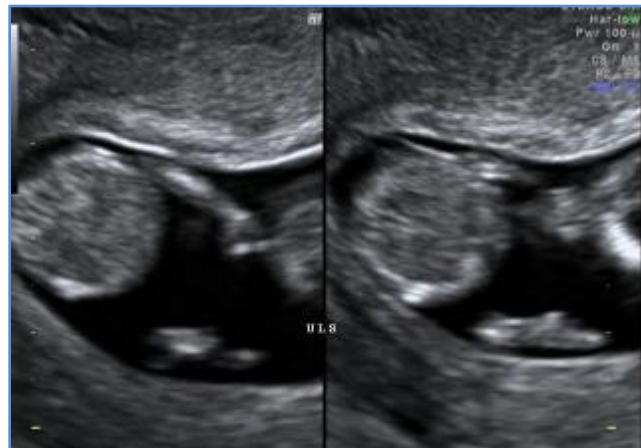
The practice of demonstrating the both the humerii and then both bones of the forearms, followed by the coronal section of the fetal hand demonstrating the open fingers, helps greatly in improving detection rates not only of larger transverse limb reduction defects but also of reduction of fingers, ectrodactyly and polydactyly. First trimester screening for anomalies is now standard of care and performed with trans-abdominal or trans-vaginal route to obtain the necessary images. (Fig. 7, 8) As an additional learning point, the CRL of the fetus at the time of the first trimester scan was 48mm.

This small size of fetus would have benefitted from trans-vaginal evaluation.<sup>(4, 5)</sup> Early detection provides the patient with a choice of termination of pregnancy at an earlier gestational age, reducing the physical morbidity and to some degree the emotional morbidity associated with the procedure. In a patient with co-morbidities, the advantage of a timely diagnosis is substantial. Although first trimester scanning cannot be relied on exclusively as a targeted scan, it does have the potential to detect many anomalies early thus benefiting the patient.

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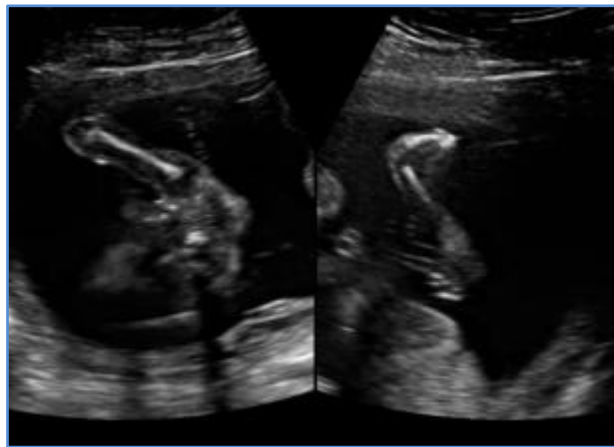
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**Fig. 1: First trimester upper limbs**



**Fig. 2: B mode Right arm defect**



**Fig. 3: Fetal right arm transverse defect**



**Fig. 4: Fetal right arm 3D**



**Fig. 5: Post expulsion Right upper limb**

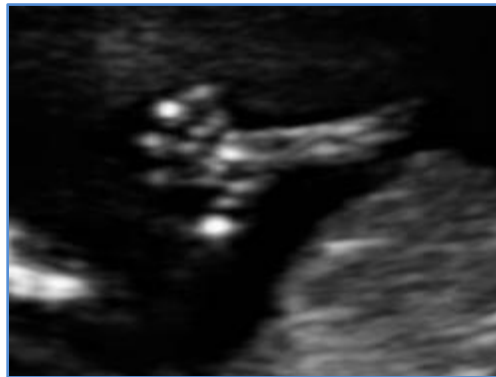
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**Fig. 6: Post expulsion Left**



**Fig. 7: Normal upper limb super**



**Fig. 8: Normal hand first trimester**

### **AUTHORS:**

1. Chitra Andrew
2. Natasha Nawani

### **PARTICULARS OF CONTRIBUTORS:**

1. Associate Professor, Department of OBGYN, Sri Ramachandra Medical College, Chennai.
2. Assistant Professor, Department of OBGYN, Sri Ramachandra Medical College, Chennai.

### **NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:**

Dr. Chitra Andrew,  
Department of Obstetrics and Gynecology,  
Sri Ramachandra Medical College,  
# 1, Ramachandra Nagar,  
Porur, Chennai-600116.  
E-mail: chitraandrew@gmail.com

Date of Submission: 29/10/2014.  
Date of Peer Review: 30/10/2014.  
Date of Acceptance: 18/11/2014.  
Date of Publishing: 24/11/2014.