# Rare Case: Tetra-Amelia Syndrome

**Dhara Alifa, Muhammad Alamsyah Aziz, Mulyanusa Amarullah Ritonga, Adhi Pribadi**
Obstetrics and Gynecology Department, Faculty of Medicine, Universitas Padjadjaran – Dr. Hasan Sadikin General Hospital, Bandung, Indonesia
**Corresponding**: Dhara Alifa, Email address: dhara21001@mail.unpad.ac.id

**Abstract**
**Introduction:** Congenital abnormalities are anomalies that become a fear for a family, when a mother experiences pregnancy. Some abnormalities are temporary and can be corrected, while some are permanent and cannot be corrected, so screening at antenatal time is very important.
**Objective**: To explain and analyze a rare case of Tetra-amelia syndrome and how to diagnose it.
**Case:** A 32-year-old woman with a 32-week-old G3P0A2 pregnancy visited the maternal-fetal clinic. According to ultrasound data, a single fetus with a gestational age of 31-32 weeks and a fetal weight of 1837 grams is in breech presentation. Only the proximal components of the arm and leg are formed, leaving the radius bones, ulna, tibia, and fibula unformed. The femur has a length that corresponds to 16 weeks, while the humerus has a length that corresponds to 20 weeks. These findings also revealed a discrepancy in pregnancy age. A tetra-amelia abnormality was discovered at the end of the ultrasound scan. Caesarean section performed on August 6, 2021, at the age of 39 weeks, a baby girl has been born baby girl a baby girl weighing 2300 grams, a body length of 31 cm, with mild asphyxia.
**Conclusion:** During antenatal care, ultrasound on the unidentified distal part of the entire extremity can detect Tetra-amelia syndrome.

**Keyword:**Tetra-amelia syndrome, Ultrasound, Congenital Anomaly

# Kasus Langka: Sindrom Tetra-amelia

**Abstrak**
**Pendahuluan**: Kelainan bawaan adalah anomali yang menjadi trauma bagi keluarga, ketika seorang ibu mengalami kehamilan. Beberapa kelainan bersifat sementara dan dapat diobati, sementara beberapa bersifat permanen dan tidak dapat diperbaiki sehingga skrining pada waktu antenatal sangat penting.
**Tujuan**: Artikel ini untuk menjelaskan dan menganalisis kasus langka sindrom Tetra-amelia dan cara mendiagnosisnya.
**Kasus**: Seorang wanita berusia 32 tahun dengan kehamilan G3P0A2 berusia 32 minggu mengunjungi klinik fetomaternal. Hasil pemeriksaan ultrasonografi menunjukan janin tunggal dengan usia kehamilan 31 - 32 minggu dan berat janin 1837 gram dalam letak sungsang. Hanya komponen proksimal lengan dan kaki yang terbentuk, sedangkan tulang jari-jari, ulna, tibia, dan fibula tidak terbentuk. Femur memiliki panjang yang sesuai dengan 16 minggu, sedangkan humerus memiliki panjang yang sesuai dengan 20 minggu. Kelainan tetra-amelia dapat dideteksi dengan pemindaian ultrasonografi. Pada tanggal 6 Agustus 2021 presentasi sungsang, dilakukan operasi caesar, lahir bayi perempuan pada usia 39 minggu, berat 2300 gram, panjang 31 cm, disertai asfiksia ringan.
**Kesimpulan**: Pemeriksaan ultrasonografi pada perawatan antenatal dapat mendeteksi sindrom Tetra-amelia, bila bagian distal ekstremitas tidak teridentifikasi.

**Kata kunci**:Sindrom Tetra-amelia, Ultrasonografi, Kelainan Kongenital

## Introduction

Congenital abnormalities are also the causes of miscarriage, stillbirth, and death shortly after birth.[1] According to the World Health Organization, congenital abnormalities cause 295,000 infant deaths in the first 28 days of life each year.[2] Congenital disorders are disorders that have existed since birth and can be caused by genetic or non-genetic factors. In this case report, we obtained the results of a pre natal screening diagnosis of Tetra-amelia syndrome, which raises the question of whether the pregnancy should be continued or terminated due to the need for long-term care. It has a good prognosis until delivery based on ultrasound examination, which shows the general growth of the fetus. This case report will go over a caesarean section with signs of transverse lie and tetra-amelia.

## Case

A 32-year-old woman, G3P0A2 gravida 31-32 weeks, was referred to Hasan Sadikin General Hospital's Maternalfetal Division Clinic by a gynecologist with suspicions of limb anomalies. The woman had no history of taking medicines or herbs during pregnancy, nor did she have a family history of congenital abnormalities. There was no history of chronic conditions like high blood pressure, diabetes, or heart disease, all of which were rejected. Ultrasound tests at maternal-fetal clinics yield the following results: According to the gestational age of 31-32 weeks, a single fetus lives in breech presentation with an estimated fetal weight of 1837 grams. Only the proximal components of the arm and leg are produced, leaving the radius bones, ulna, tibia, and fibula unformed.

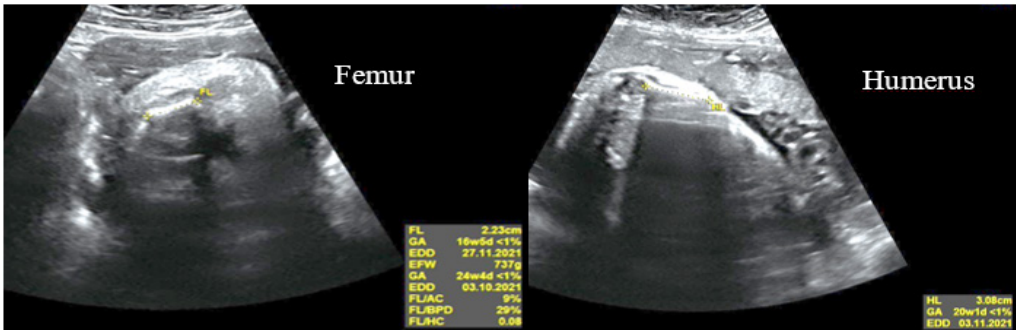


**Figure 1 Ultrasound Findings. In the ultrasound findings, it could be seen that the proximal parts of the arm and leg are only formed by the femur and humerus bones, without the distal part.**

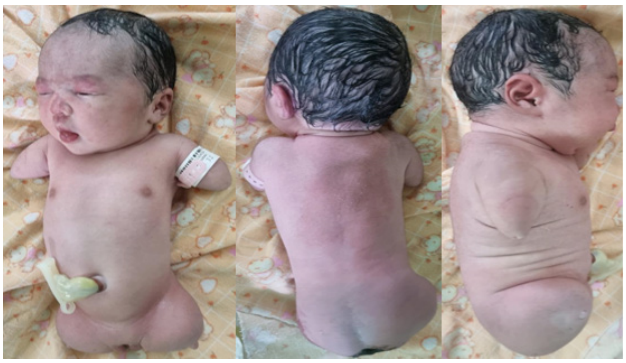

**Figure 2 Baby after birth. Bilaterally, arms and legs were visible only with incomplete proximal parts.**

The femur has shown a length according to the age of 16 weeks, while the humerus indicates the age of 20 weeks (Figure 1). These results also showed a mismatch with the age of pregnancy. At the conclusion of the ultrasound examination, there was a tetra-amelia disorder. On August 6, 2021, at the age of 39 weeks, breech presentation, performed caesarean section, and the birth of a baby girl weighing 2300 grams, with a body length of 31 cm, with mild asphyxia (Figure 2).

## Discussion

Tetra-amelia syndrome is a very rare limb disorder characterized by a complete or nearly complete absence of the extremities, either unilaterally or bilaterally. This can happen if the embryo is disrupted during the early stages of intrauterine development. It can occur as a single anomaly, but it is more commonly seen in conjunction with other congenital malformations.[1] Tetra-amelia is an extremely rare human genetic disorder that affects 1.5 to 4 out of every 100,000 live births.[3] The anomaly is distinguished by the absence or incomplete of all four limbs and may be accompanied by other anomalies. It was previously thought to follow the recessive autosomal inheritance mode.[4] The disease's locus has been assigned to chromosome 17q21 via homozygosity mapping, with a mutation in the WNT3 gene in the affected family fetus.[5]

Neonates born with tetra-amelia syndrome may experience health issues that result in postnatal death.[6] Other organs, such as the face, head, nervous system,heart, and diaphragm hernia can also be malformed as a result of this syndrome.[7] In some cases, babies with tetra amelia syndrome's lungs do not fully develop, resulting in severe postpartum asphyxia and severe breathing difficulties.[8] The development of the upper and lower limbs is similar; the lower limbs will be one to two days slower.[9] Tetra-amelia can be diagnosed early on through genetic testing or ultrasound.

Early detection of tetra-amelia can be accomplished through genetic testing or ultrasound during antenatal care. A possible sign of tetra-amelia syndrome when there is no identification of the distal part of the superior and inferior extremities on ultrasound. As a result, ultrasound examination plays a role in the verification of congenital abnormalities during pregnancy. Many factors can contribute to congenital anomalies, including single gene defects, chromosomal abnormalities, multifactorial offspring, environmental teratogens, and micronutrient deficiencies.[10] Tetra-amelia syndrome is associated with a type of multiple congenital anomaly.

## Conclusion

Tetra-amelia syndrome is a rare case and can be detected during antenatal care with ultrasound on the unidentified distal part of the entire extremity.

## Recommendation

Ultrasound screening should be done in every pregnancy.

## Conflict of interest

None to Declare

## References

1. https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=294967
2. https://www.who.int/health-topics/congenital-anomalies#tab=tab_1
3. Lenz W. Genetics and limb deficiencies. Clin Orthop Relat Res 1980;148:9-17.
4. Magdariaga DLM, Leon MA.Tetra-amelia syndrome: a case report. MEDISAN.2020; 24(2):263-267, 2020
5. Likith RP, Manoj. Characterisation of

tetra amelia syndrome by snp based on computational genotyping analysis.asian j pharm clin res. 2014;7(5):65-66

6. Al Riyami N, Ahmed A, Tanzeem S, Abdul-Latif M. Fetal Amelia: A Case Report Oman Medical Journal.2012; 27 (1): 54-55 DOI 10. 5001/omj.2012.11
7. Diarra NM, Mamour G, Birame NS, Abdoulaye D, Gothe FLLN, et al. (2020) A Report of 2 Cases of Tetra-Amelia Syndrome. Gynecol Obstet Case Rep.2020;6(1):4 DOI: 10.36648/2471-8165.6.1.858.
8. Mina SS, Arya S, Siddiqui K, Salhan M, Mina D. Tetra-Amelia syndrome with congenital diaphragmatic hernia - A rare entity. *The Indian journal of child health.*2017;4:457-458.
9. Tickle C. How the embryo makes a limb: determination, polarity and identity. *J Anat*. 2015;227(4):418-430. doi:10.1111/joa.12361.
10. Dutta H, Baruah M, Borbora D.Maternal nutrition and the risk of congenital malformations in the tea garden community of Assam, Northeast India. clinical epidemiology and global health.2016. (4).p.63–68. http://dx.doi.org/10.1016/j.cegh.2015.07.006