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Case Report

Absence Fetal Hand: A Rare Congenital Anomaly

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Abstract

Congenital malformation of the upper limbs is not a very frequent, for they affect less than 0.2% of born babies. Many of these malformations are not severe, have a minor functional impact and may be surgically treated.

Methods: We are reporting one case of congenital absence of right fetal hand (Acheiria) diagnosed by ultrasonography at 20 weeks of gestation with completely normal right forearm, other 3 limbs and the other fetal anomalies by detailed anomaly scan. The role of

Conclusion: Ultrasound plays an important role in the diagnosis of this congenital anomaly. It provides findings that allow establishing the type and severity of abnormality.

Keywords: *Acheiria; Isolated Anomaly; Congenital Malformation; Hand Absence; Ultrasound*

Background

Acheiria means absence of one or both hands, it is a rare anomaly affects about 1 of 65,000 live births [1]. Congenital upper limb malformations most commonly are sporadic and usually diagnosed by ultrasound during prenatal examination as an isolated finding and less commonly to happen in association with other malformation or diagnosed as a part of syndrome [2]. The prenatal ultrasound plays an important role in diagnosis of upper limb malformations [3]. We report one case of absent right fetal hand diagnosed by ultrasonography at 20 weeks of gestation.

Case Report

A 26 years old pregnant female G4p3, previous 2 cesarean sections and one normal vaginal delivery, came to radiology department at 20 weeks of gestation for routine antenatal ultrasound examination (anomaly scan). She is the fourth child in the family, all of the 3 siblings are normal, detailed history was taken from the mother about her pregnancy, there was no chronic or febrile illness, no history of epilepsy, no history of x ray exposure, no history of drug use like anticoagulants or any other teratogenic drug. The father are 32 years old and there is no history of consanguinity between them.

There was no prior history of such or other anomaly in the family.

The abdominal ultrasound examination at 20 weeks of gestational age revealed: Right hand was absent (Figure 1-3), the right shoulder, elbow, arm and forearm were normal. The other 3 limbs were also completely normal, examination of heart, neck, chest including the heart, abdomen and pelvis also revealed no abnormality.

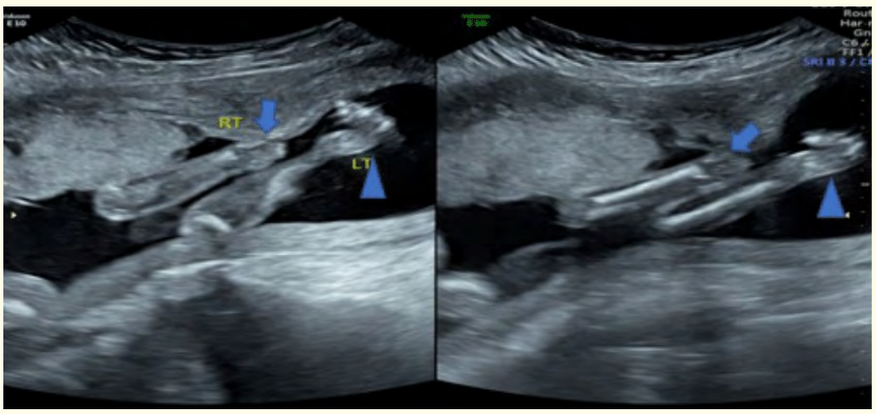

Figure 1: *Ultrasound: absent fetal right hand (arrow) and normal fetal right and left forearm and normal left fetal hand (arrow head).*

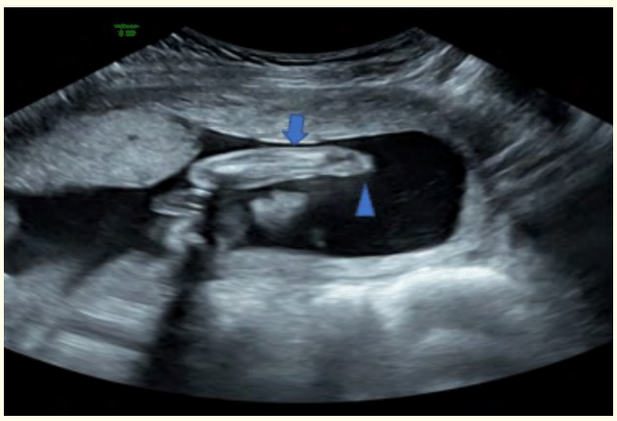

Figure 2: *Ultrasound: absent fetal right hand (arrow head) and normal fetal right forearm (arrow).*

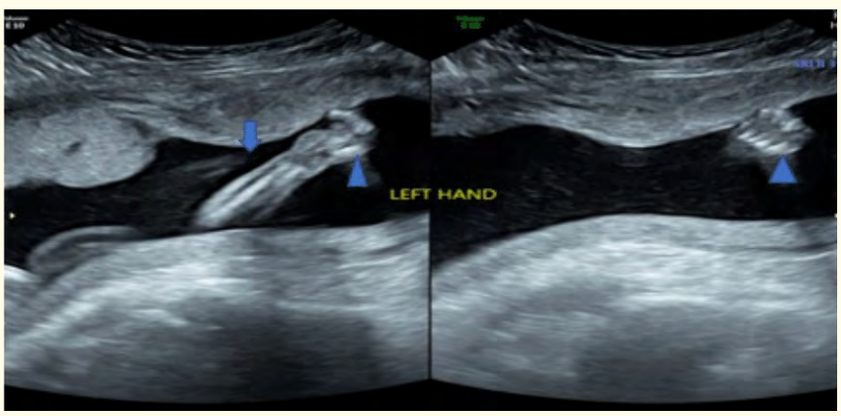

Figure 3: *Ultrasound: normal left fetal hand (arrow head) and forearm (arrow).*

The patient is referred to fetal maternal medicine unit for confirmation, for genetic counselling and for karyotype analysis and it is proved that the right fetal hand is absent with no detectable cause.

Discussion

Congenital limb abnormalities are rare, account about 6: in 10,000 live births, more common in the upper limbs than the lower limbs. Congenital limb abnormalities are more common unilateral than bilateral, and more in the right side than the left side [2].

Congenital limb abnormalities are mostly sporadic trait, to lesser extent occur in association with other malformations either skeletal or non-skeletal or in association with systemic disorders [2]. The causes of limb abnormalities are complex, such as vascular compromise, intrauterine events, chromosomal abnormalities, single gene disorders, and exposure of the mother to teratogenic drugs or maternal chronic disease, however in many patients the cause is still not known [3].

Embryonic development of the limb: By ultrasound, the limb buds are first seen at about the 8^{th} week of gestation, by 9^{th} week of gestation the femur and humerus are seen, by 10^{th} week of gestation tibia/fibula and radius/ulna are seen and digits of hands and feet are seen from 11^{th} week and all long bones are seen from 11^{th} weeks. Congenital upper limb malformations happen usually at 3^{rd} to 8^{th} weeks of embryogenesis [3].

Congenital upper limb malformations are divided commonly according to the clinical findings and radiological findings into four types: 1) Failure of formation. 2) Failure of differentiation. 3) Duplication. 4) Brachydactyly. The definition of failure of formation is arrest of the development of upper limb and called congenital amputation also [2]. The Congenital amputations may occur at any level: Amelia is limb absence, hemimelia is forearm absence, acheiria is hand absence, adactyly is fingers absence and aphalangia is phalanges absence [4].

The main differential diagnosis of congenital hand absence is Amniotic band syndrome, which occurs as a result of partial rupture of the amniotic membrane intrauterine, this leads to formation of fibrous bands that entangle the fetal limbs, fingers, or any other fetal parts resulting in limb or digital amputations or limb deformities which characteristically asymmetrical [2].

Prenatal ultrasound examination is the first and the most important tool in prenatal diagnosis of fetal anomalies. If limb abnormalities are detected, the discerption of the anomaly should be done carefully as well as careful examination to look for or rule out associated abnormalities and to differentiate either if it is single gene disorder or chromosomal abnormalities. The possible causes, prognosis, treatment options and risk of recurrence in future pregnancy should be explained to the parent and this may require a multidisciplinary meeting of radiologist, obstetrician, neonatologist, clinical geneticist, and orthopedic surgeons [3].

If prenatal fetal hand anomaly is detected, the patient should be referred to a specializes clinic in the diagnosis and management of fetal malformations and genetic syndromes (Fetal Maternal Medicine Unit). A complete work-up of the fetus has to be done, and complete ultrasound examination of the fetus and the fetal heart to confirm the presence or absence of other associated abnormalities. The patient may need focused repeated ultrasound examinations to detect the underlying anomalies if present. If prenatal fetal hand anomaly is detected it is essential to do genetic counseling and karyotype analysis [5].

Conclusion

Ultrasound play an important role in the diagnosis of this congenital anomaly. Absence of family history, x ray, drug history, in addition to non-specific linkage to any environmental factor lead us to suggest that this abnormality is of rare occurrence. There is very little information on these anomalies in the literature, their management remains a challenge, however carful investigations will allow the identification of families at high risk and will facilitate therapeutic measures to repair the damages.

Disclosure

The authors declared no conflict of interest.

Bibliography

1. Sabry MA and TI Farag. "Hand Anomalies in Fetal-Hydantoin Syndrome: From Nail/phalangeal Hypoplasia to Unilateral Acheiria". *American Journal of Medical Genetics* 62.4 (1996): 410-412.

2. Ermito Santina., *et al*. "Prenatal Diagnosis of Limb Abnormalities: Role of Fetal Ultrasonography". *Journal of Prenatal Medicine* 3.2 (2009): 18-22.

3. Aucourt Julie., *et al*. "Congenital Malformations of the Hand and Forearm in Children: What Radiologists Should Know". *Seminars in Musculoskeletal Radiology* 16.2 (2012): 146-158.

4. Bisneto Edgard Novaes França. "Congenital deformities of the upper limbs.: part i: failure of formation". *Revista Brasileira de Ortopedia* 47.5 (2012): 545-552.

5. Rypens Françoise., *et al*. "Obstetric US: Watch the Fetal Hands". *RadioGraphics* 26.3 (2006): 811-829.

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