Case Report

A case of acheiria

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Abstract

Congenital limb anomalies are rare. Acheiria is a congenital limb abnormality that presents as an absence of the hand and it is often diagnosed by prenatal ultrasonography. Herein we present a case of an 11-year-old female patient with acheiria. This case stresses on further studying the relationship between advanced pregnancy age, drugs or herb use during pregnancy and acheiria.

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Introduction

Acheiria is a rare congenital limb abnormality that manifests as an absence of the hand [1]. The prevalence of absent hands or fingers is 0.93 per 10,000 people, while the prevalence of upper extremity anomalies is around 27 per 10,000 people [2]. Acheiria is often identified by prenatal ultrasonography as an isolated finding, although it can be seen with other abnormalities [1,3]. Herein, we describe a case of a left congenital hand deformity in a female born to a mother who was 45-year-old. Their attempts to have a baby failed for 27 years even with the use of assisted reproductive technology, until she used an unidentifiable herb.

Case presentation

An 11-year-old right-handed female patient presented to our clinic by her parents complaining of a left-hand deformity since birth. She was a result of natural conception without assisted reproductive techniques, with eventless pregnancy which ended with an elective cesarean section. Her birth weight was around 3.5 kilograms with no need for a neonatal intensive care unit admission. Her neonatal physical examination was otherwise normal; the deformity was on her left hand, with normal right upper (including the hand) and bilateral lower limbs. No signs of hypotonia or spasticity throughout, and normal deep tendon reflexes. She was also free of
cardiopulmonary, gastrointestinal, urogenital, and other deformities. Her mother was 45 years of age, had a monthly visit during pregnancy, and denied consumption of any drugs during pregnancy apart from vitamins. In addition, she denied any use of illicit drugs, smoking, radiation exposure, infections, travels, or any abnormal finding during the routine obstetric clinical, laboratory, and ultrasonography follow-up. The patient’s immunization history was up to date, she was reaching appropriate milestones throughout her childhood, and there is no family of limb deformities. At the time present, no changes were observed on the physical examination (Fig. 1). She has normal fine and gross motor activity in the right hand. Moreover, the patient is doing well and reaching her milestones appropriately. An X-ray was ordered; it showed a complete absence of the left hand’s bones with the only rudimentary soft tissue of fingers (Fig. 2). Depending on the aforementioned findings the patient was diagnosed with acheiria. It is worth mentioning that they had unexplained infertility for 27 years before conceiving their daughter. They had multiple trials of ART over the years with no success conceiving. The mother reports ingesting a herbal material of unknown name or origin. However, the usage of this herb in the subsequent siblings didn’t cause a deformity.

Acheiria continues to be stressful for the patient, the family, and the treating physician due to its negative psychological and functional impact on the patient’s life. The purpose of this report is to provide information on a single instance of acheiria and perhaps to describe the events that lead to it.

Acheiria cases have been reported with a focus on diagnosis, pathogenesis, and related abnormalities. Even with routine sonography follow-ups, isolated cases of acheiria are more likely to go unnoticed, as in our case, despite the fact that prenatal ultrasound is still the gold standard for acheiria detection [5]. Some diseases, such as fetal hydantoin syndrome, Cornelia de Lange syndrome, and amniotic band syndrome, can be linked to acheiria [6,7]; however, the majority of cases are sporadic [4].

Since the etiology and risk factors for the majority of cases of acheiria are still unclear, a detailed investigation of the mother’s history during and before pregnancy was conducted to identify any potential risk factors that may have contributed to the development of this anomaly. The isolated nature of the anomaly, the mother’s negative family, medical, and surgical history and the lack of drug usage throughout pregnancy point us in the direction of a sporadic cause. Even though numerous unsuccessful attempts to conceive naturally and with assisted reproductive technology necessitated the usage of an unidentified herb, the fact that the same herb had no effect on the subsequent sibling makes it less plausible that it was the source of this oddity. The mother’s advanced age (45 years old) and the fact that this was her first pregnancy are 2 additional factors worth considering when investigating cases of acheiria.

We suggest future research focus on mother’s ingestion of any unusual medicines or herbs during pregnancy with the fo-

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**Discussion**

Congenital limb abnormalities are rare. Upper limb abnormalities are more common than lower limb malformations [4]. Anomalies that are unilateral or right-sided are more frequent than those that are bilateral or left sides [1].
cus on her age throughout her pregnancy, even though there isn’t any conclusive proof tying an unidentified herb or advanced maternal age to the appearance of this aberration.

Conclusion

Congenital hand absence (acheiria) is a rare abnormality, with the right side being more frequent. It can be detected via prenatal ultrasound. The causality and risk factors are not yet clear due to the scarcity of literature. It may be associated with fetal hydantoin syndrome, Cornelia de Lange syndrome, and amniotic band syndrome. However, most cases are sporadic.

Patient consent

Written informed consent for the publication of this case report was obtained from the patient.

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