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CASE REPORT

Ectrodactyly: A rare malformation of the limbs

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Abstract

Objective: To report a case of a patient born with ectrodactyly without prenatal diagnosis, despite the possibility of obstetric ultrasound guided diagnosis. **Methods:** Case report based on the newborn's physical examination in the delivery room and it medical records. Research on PubMed and SciELO databases guided the discussion and description of this rare congenital malformation. **Results:** The patient had been diagnosed with ectrodactyly during the newborn physical examination, without any other external associated malformation. Due to the innapropiate prenatal care, the malformation wasn't diagnosed before the patient was born. **Conclusion:** Obstetric ultrasound allows the diagnosis of congenital malformations, and, therefore, to prepare the parents so that they will be able to cope and accept their child's physical condition, as well as the prompt involvement of a multi professional team in the child's care. Ectrodacyly's treatment should be individualized.

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INTRODUCTION

Ectrodactyly is a congenital abnormality in fetal embryological development of autosomal dominant inheritance with variable penetrance that affects about one in approximately every 90,000 to 150,000 live births[1,2].

Its anatomical feature is the congenital absence of the central digits, formed by the second, third, and fourth digits, thus generating a large median cleft in the hand that is associated with aplasia/hypoplasia of metacarpals and/or phalanges[2]. The presence of the first and fifth digits is largely due to embryogenesis occurring in different periods, which preserves them[2]. In addition to the changes in the hands, the feet can also be affected, and together they make up ectrodactyly as a syndrome[3].

Ectrodactyly is classified into two basic subtypes: the typical form (1/90,000 live births) and the atypical form (1/150,000 live births). In the typical form, the absence of the metacarpal bones and phalanges results in a V-shaped defect that divides the hand into an ulnar portion and radial portion. In the atypical form, defects in the metacarpal bones of the medial fingers generate a wide U-shaped cleft[4].

The case report was based on care in the delivery room, which is described in the patient record. A study of databases such as PubMed and Scielo was performed in order to obtain a better description to report this rare malformation of the limbs.

CASE REPORT

A female newborn patient referred to as R.N.S. was born via cesarean after 39 weeks and 3 days of gestation, weighing 3795 grams and measuring 51 cm (AGA). Apgar 8/9. The mother had a history of a previous miscarriage, prenatal care consisting of three consultations, and serological testing only in the first trimester, the results of which were all negative. The mother also had a history of smoking and alcohol consumption during pregnancy. The father had a history of drug use.

The infant presented no complications at birth and was found to have active, regular breathing movements, and heart rate greater than 100 beats per minute. During the physical examination shortly after birth, the malformation of the hands was noticed, with the absence of the second, third and fourth digits of the left hand and the absence of the fourth digit of the right hand, preserved mobility, and adequate grip in both hands. There were no abnormalities in the lower limbs and the other aspects of the physical examination have been normal thus far (Figures 1 and 2).

After the initial assessment and a review of the literature, the patient was diagnosed with ectrodactyly, as confirmed by a hand surgeon with whom the patient is likely to have follow-up consultations for future conduct to be determined.

Figure 1. Absence of second, third, and fourth fingers.

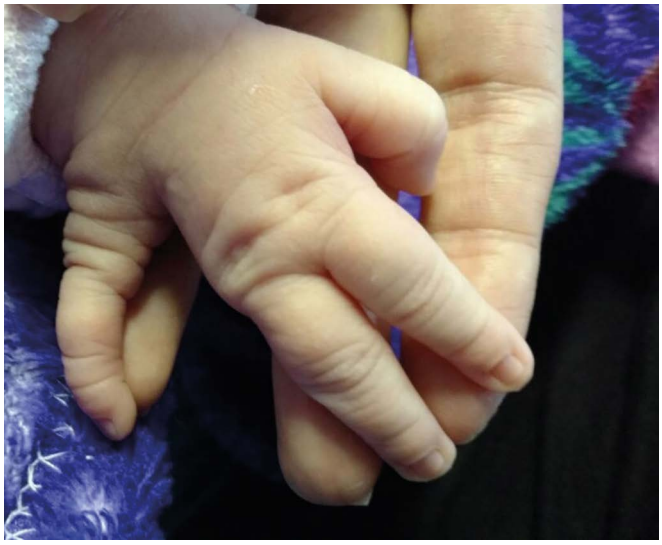

Figura 2. Absence of fourth digit.

DISCUSSION

Ectrodactyly may occur alone (the non-syndromic form) or may be associated with other malformations (the syndromic form) such as cleft palate, ectodermal dysplasia, the involvement of the toes, craniofacial abnormalities, tibial aplasia, and sensorineural hearing loss, as well as 40 other associations previously described in the literature[3,5]. The most commonly described version of the syndrome is characterized by ectrodactyly, ectodermal dysplasia, and cleft palate/lip (EEC syndrome)[5].

In addition to cleft shape (V or U), other changes described in the literature seek to distinguish between typical

and atypical forms[4,5]. Overall, syndactyly, associated cleft lip, foot involvement, genetic inheritance, the lack of digits, and bilateralism are more characteristic of the typical subtype.

In the case reported herein, the patient was diagnosed in the first physical examination, during which no other external malformations were observed (she was therefore diagnosed with the non-syndromic form). In addition, the patient exhibited both typical and atypical features in terms of bilateral malformation, the lack of a family history of ectrodactyly or other malformations of the limbs, with lip and palate intact and an absence of syndactyly.

Due to inadequate prenatal care, diagnosis in this period was not possible. However, obstetric ultrasounds are an important tool for diagnosing malformations in the first trimester of pregnancy[2,5]. In addition to serving as a diagnostic method, ultrasounds allow for the search of associated congenital abnormalities. An early diagnosis of ectrodactyly allows the parents to prepare psychologically and makes them more likely to accept the child's physical condition. It also allows for the early involvement of a multidisciplinary team in the child's care[2,5].

Based on the wide range of associated malformations, treatment for ectrodactyly should be individualized[6]. The goal of surgical management of hand malformations is to achieve grip capacity through the reconstruction of the first commissure and to close the cleft generated by the phalangeal and metacarpal abnormalities[4]. The use of prostheses may also be indicated[6,7]. In addition to improving the patient's psychosocial well-being, the goal of the surgery is to achieve as much limb function as possible, and patients often adapt well to residual handicaps[4,6]. Conservative management is reserved for children with severe cases and major deficits in neurodevelopment[4].

Due to the involvement of autosomal inheritance, genetic counseling should be the cornerstone of malformation prevention. This genetic counseling should be accompanied by early prenatal diagnosis via ultrasound[5].

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