Obstetrica şi Ginecologia LXIV(2016) 247-251 **Case Report**

REVERSIONAL UNILATERAL POSTAXIAL POLYDACTYLY TYPE I IN FETAL LIFE: A CASE REPORT

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Abstract

Introduction: In early second trimester a rudimentary and mobile unilateral extra digit, as an abnormal fork in the existing fifth finger, was diagnosed. Due to the association between polydactyly and several syndromes, efforts have been made to exclude associated anomalies. Spontaneous amputation was noted as pregnancy evolved towards the end mid-trimester. To our knowledge, this is the first report of a case of reversional unilateral postaxial hand polydactyly in fetal life.

Case presentation A 28 year-old Caucasian (16 weeks of amenorrhea) pregnant lady presented to our Prenatal Diagnostic Unit, for the second trimester scan. She was a low risk patient, with a history of non-eventful term pregnancy. First trimester screening scan for fetal anomalies was negative. A unilateral postaxial hand polydactyly on the left hand was diagnosed. A thorough search for additional anomalies was performed, and failed to find any associated ultrasound markers. Increased levels of anxiety were noted on both parents. This led to extensive multidisciplinary counselling. At the 25 weeks scan, the extra digit was absent.

At birth, a very small bud was present on the ulnar side of the fifth finger of the left hand.

Conclusion In our view, this case has an important teaching point, in terms of dynamic of intrauterine structures, as fetal extremities, in terms of psychological side-effects of detailed anomaly scan and in terms of counselling after the prenatal diagnostic of an anomaly.

Rezumat: Polidactilia postaxială unilaterală tip I, reversibilă prenatal: prezentare de caz

Introducere: În cazul de mai jos a fost diagnosticată prezenţa unui deget supranumerar, rudimentar, mobil (cu aspect de segment accesoriu al degetului V), la o singură mână, la debutul trimestrului II de sarcină. Au fost depuse eforturi pentru a se exclude alte anomalii, datorită asocierii cunoscute între polidactilie şi sindroamele fetale. Pe parcursul evoluţiei sarcinii, la sfârşitul trimestrului II, s-a constatat amputaţia spontană in utero. Acesta este primul caz care demonstrează cu imagini reversibilitatea polidactiliei unilaterale postaxiale la mână, în viaţa fetală.

Prezentare de caz: Pacienta, în vârstă de 28 de ani, s-a prezentat în Unitatea de Diagnostic Prenatal, la 16 săptămâni de amenoree, în vederea examinării ecografice. Pacienta era încadrată în grupa de risc scăzut, având în antecedente o naştere la termen, fără complicaţii. În primul trimestru fusese supusă screeningului pentru anomalii cromozomiale, utilizându-se testul combinat (încadrarea - în grupa de risc scăzut pentru anomalii cromozomiale). A fost diagnosticată polidactilia unilaterală postaxială la mâna stângă. S-a practicat o ecografie detaliată de anomalii, care nu a relevant prezenţa nici unui alt marker de anomalii congenitale structural sau cromozomiale. Părinţii au demonstrat nivele crescute de anxietate. Aceasta a dus la şedine repetate de consiliere în echipe multidisciplinare. La reexaminarea ecografică de la 25 de săptămâni de amenoree s-a constatat absenaţ degetului supranumerar. La naştere s-a constatat prezenţa unui rudiment de mici dimensiuni, pe partea ulnară a degetului V de la mâna stângă.

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KEY WORDS: polydactyly, prenatal diagnosis, ultrasound

Concluzii: În opinia autorilor, cazul are implicaţii educative, în sensul că subliniază dinamica intrauterină a structurilor fetale, la nivelul extremităţilor. De asemenea, evidenţiază efectul secundar nedorit al examinărilor ecografice, şi dificultăţile ridicate de acestea în consilierea cuplurilor, în cazurile în care se precizează antenatal diagnosticul unei anomalii minore.

Cuvinte cheie: polidactilie, diagnostic antenatal, ecografie

Introduction

The second trimester detailed anomaly scan scan became, many years ago, the pillar of obstetric care. During it, each part of the fetal body should be examined. If any abnormalities are detected the significance of the findings must be discussed and the couple is given the opportunity to have further counselling. Current guidelines do not require counting fingers or toes as part of the routine mid-trimester scan (1,2). Still, in most cultures, physical perfection is the standard to which all couples aspire. Thus, polydactyly, especial if hands involved, is usually perceived by the parents as an important deformity, and women presenting with any fetal anomaly have heightened levels of anxiety during the pregnancy**.**

Polydactyly refers to the situation where there are more than the usual number of digits (five) in a hand or foot. It can be broadly classified as: pre-axial polydactyly: extra digit(s) towards thumb or toe and post-axial polydactyly: extra digit(s) towards 5th finger in limb (3).

Genetic syndromes have been associated with different kinds of polydactyly (4).

Ulnar polydactyly occurs as an isolated congenital condition, but can also be part of a syndrome (5). The syndromes which occur with ulnar polydactyly are: Greig cephalopolysyndactyly syndrome, Meckel syndrome, Ellis–van Creveld syndrome, McKusick–Kaufman syndrome, Down syndrome, Bardet–Biedl syndrome, Smith–Lemli–Opitz syndrome (6). Polydactyly is associated with different mutations, either mutations in a gene itself or in a cis-regulatory element responsible for the expression of a specific gene. The estimated incidence of postaxial polydactyly is 1 in 3000 live newborns (7,8,9).

Usually the extra digit is a small piece of soft tissue, but it can also contain bone without joints.

Case presentation

A 28year-old Caucasian pregnant lady presented to our Prenatal Diagnostic Unit, for the second trimester scan. She was a low risk patient, with a history of non-eventful term pregnancy. Previously first trimester genetic and anomaly scan, performed at 12 weeks and 2 days, was negative at she also was screened as a low risk pregnancy for chromosomal anomalies at the combined test (Figure 1).

The diagnostic scan was performed at 16 weeks of amenorrhea. The average ultrasound age was consistent with the menstrual age. A unilateral postaxial hand polydactyly on the left hand was diagnosed. The extra finger ultrasound suggested that it was the common form, as a rudimentary extremely mobile extra finger, connected to the small digit by a thin skin pedicle (”loose finger”). A thorough search for additional anomalies was performed, and we did not find any associated ultrasound markers. Increased levels of anxiety were noted on both parents. This led to repeated at request ultrasound exams and to extensive multidisciplinary counselling (Figure2).

At the 25 weeks scan, the extra digit was absent at ultrasound investigation (Figure 3).

At birth, a very small bud was present on the ulnar side of the fifth finger of the left hand. The newborn phenotype was normal, the clinical exam was negative for other structural anomalies, as was the postpartum evolution and the follow-up assessment.

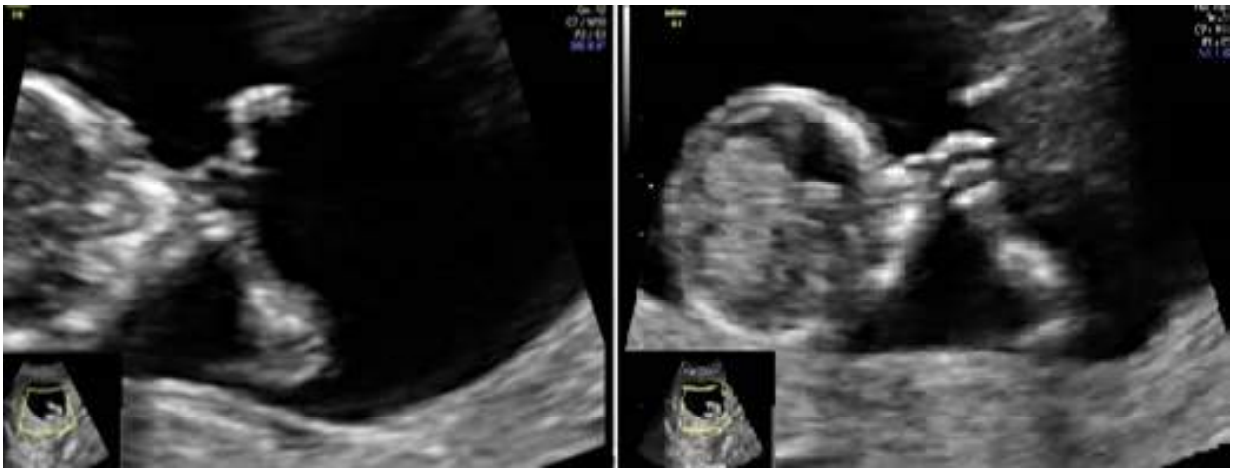

Figure 1. Aspects of the left fetal hand at the first trimester genetic and anomaly scan.

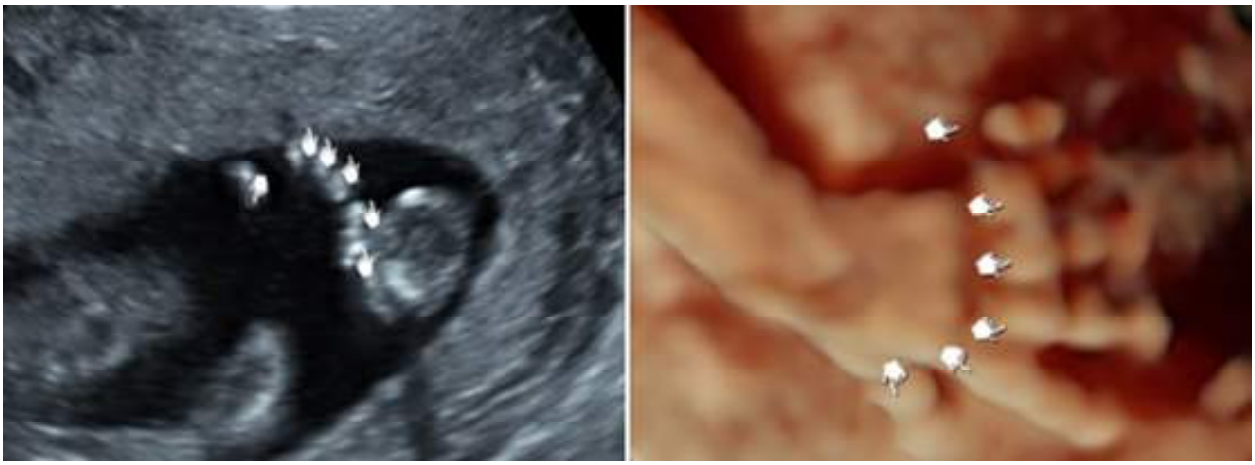

Figure 2. Early second trimester 2D and 3D aspects of the left fetal hand (16 weeks of amenorrhea).

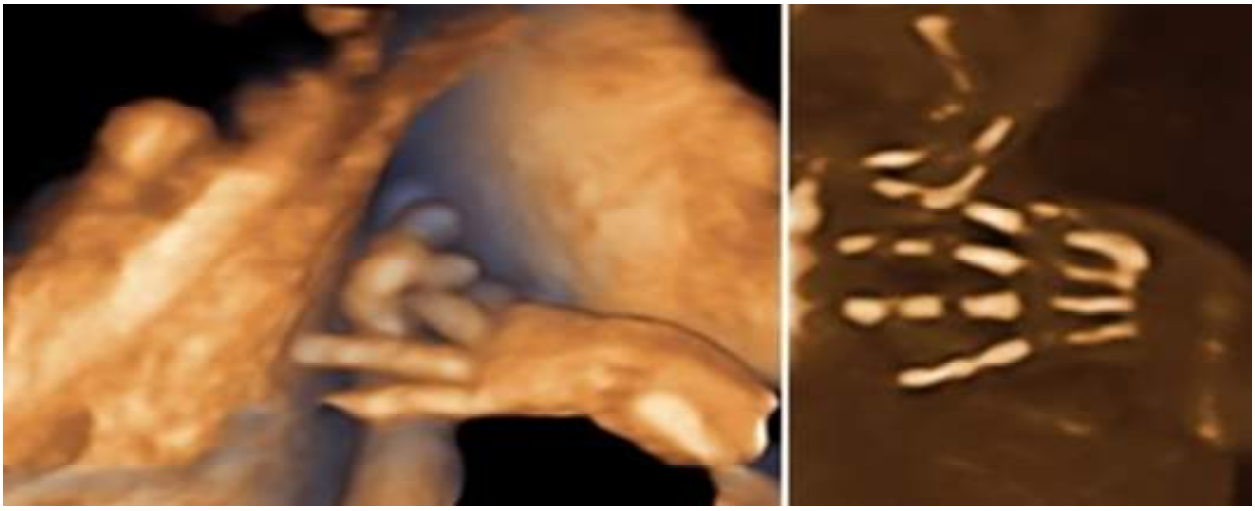

Figure 3. Late second trimester 3D reconstruction of the left fetal hand. Absence of the extra digit (25 weeks of amenorrhea).

Discussion

Fetuses with a congenital upper extremity deformity should be examined by specialists in prenatal diagnosis and geneticists for other congenital anomalies.

In the case reported, the parents are Caucasians, thus an a priori lower risk couple (10). Ulnar polydactyly occurs ten times more often in negroid ethnicities and is most common in African populations.

The incidence in Caucasians is reported as 1 in 1,339 live births, compared with 1 in 143 live births in Africans.

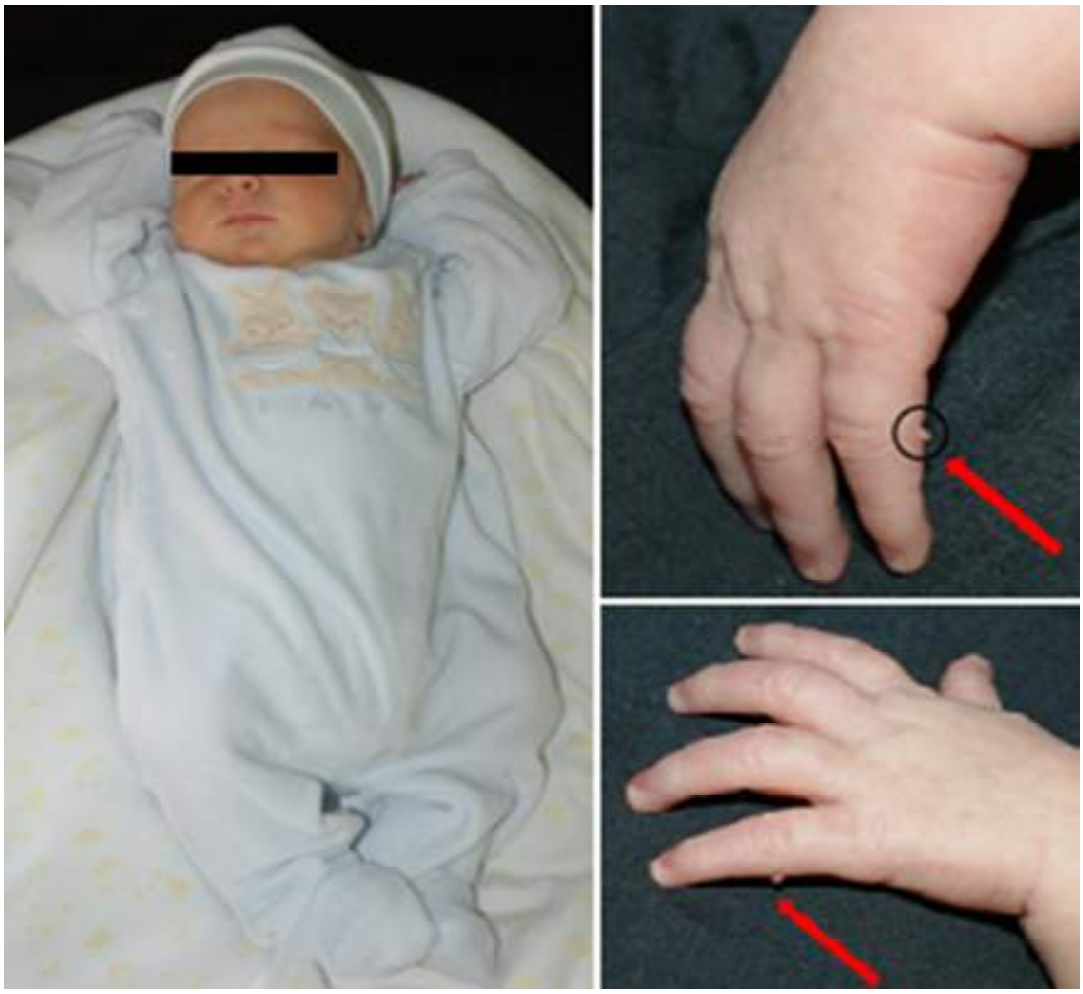

Figure 4. Postnatal aspect of the newborn.

Although in patients with African ancestry ulnar polydactyly mostly occurs isolated, the presentation in Caucasians is often associated with a syndrome (11). In our case, the newborn karyotype was normal.

In almost 14% of all patients, this type of polydactyly is hereditary (12). It usually passes on in an autosomal dominant manner with variable expression and incomplete penetrance. It is genetically heterogenic, mutations in different genes can be the cause (12, 13). We found a negative family history.

Although nowadays guidelines do not require counting fingers or toes as part of the routine mid-trimester scan, in many setting, like ours, it is attempted on daily basis in every patient. There is proof that maternal anxiety is almost as profound before invasive maneuvers as before non-invasive ones, like a detailed ultrasound exam. And there is evidence that the prenatal diagnosis has long-term emotional impact (14, 15). The psychological side effects of prenatal diagnostic procedures are a reality of nowadays medicine. In our case, the parents were highly educated, hence more vulnerable at a suspicion of an obvious hand anomaly in their to be borne baby.

An abnormal ultrasound finding, although minor, always leads to parental anxiety and emotional confusion. After identifying an abnormality, we should be able to clarify whether there is an important feature or not. This might prove to be difficult in some cases. On the other hand, definition of an abnormality must be correlated with diagnostic accuracy, and there are not many reports on the diagnosis accuracy in polydactyly yet.

Ulnar polydactyly usually does not interfere with hand function, but in our case, in the prenatal counselling sessions, the parents made it clear that they will opt for operatively treatment after birth (16, 17), for social reasons.

Integration of high resolution ultrasound technology into standard clinical care (18, 19) will require thoughtful changes in patient counselling.

Although the evolution in our case is distinct, it underline the principles of uncertainty, and lack of correlation between the the sequential scan findings. A correct diagnosis is essential for adequate counselling in pregnancy, and sometimes diseases are evolving (worsening or spontaneously resolving) throughout the pregnancy. Clinicians involved in antenatal diagnostic should be competent, tactful and comprehensive when counselling about any suspected anomaly.

Conclusion

Recent technological advances in ultrasound imaging offer the opportunity to detect much earlier an increasing number of fetal malformations.

A report on a unilateral postaxial polydactyly type I that spontaneously regress in fetal life may be more important to specialists involved in prenatal diagnostic and in fetal medicine. It demonstrates that duplications disorders may evolve continuously before birth.

Competing interests

The authors declare that they have no competing interests.

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