

A fetus with pre-and post-axial polydactyly

Fetal preaksiyal ve postaksiyal polidaktili

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Summary

Polydactyly, one of the congenital deformities of the hands and/or feet occurs as an isolated disorder or as part of a syndrome. Isolated polydactyly is often autosomal dominant, while syndromic polydactyly is commonly autosomal recessive. Polydactyly commonly involves only the hand or the foot, however involving both hands and feet is rare. We report a case of a fetus with pre-axial and post-axial polydactyly involving both hands and pre-axial polydactyly involving both feet, diagnosed at 20th-week-ultrasound scan.

Key Words: Pre-axial polydactyly, post-axial polydactyly, syndactyly, fetus, pregnancy.

Özet

El ve/veya ayakların konjenital deformitelerinden biri olan polidaktili, izole olabileceği gibi, bir sendromun parçası da olabilir. İzole polidaktili sıklıkla otozomal dominant iken, sendromik polidaktili sıklıkla otozomal resesiftir. Polidaktili sıklıkla sadece el veya ayakta bulunurken, her ikisinde bulunması oldukça enderdir. Olgumuz, 20. gebelik haftası ultrasonunda tanı alan her iki elde hem preaksiyal ve postaksiyal; ayrıca her iki ayakta preaksiyal polidaktili olgusudur.

Anahtar Sözcükler: Preaksiyal polidaktili, postaksiyal polidaktili, sindaktili, fetus, gebelik.

Introduction

Polydactyly is a congenital anomaly with a wide range of manifestations, ranging from varying degrees of mere splitting to completely duplicated fingers and toes. The incidence in the general population has a wide variation among various racial groups, ranging from 0.37-2 per 1000 live births (1). Polydactyly can generally be classified into pre-axial, central, and post-axial types according to the anatomic location. Pre-axial polydactyly involves the radial/medial aspect of the limb, while the post-axial type involves the ulnar/lateral aspect of the limb. The central type involves the duplication of the index, middle, or ring fingers. Isolated polydactyly is often autosomal dominant, while syndromic polydactyly is commonly autosomal recessive (2). We present a rare situation of an affected fetus with pre- and post-axial polydactyly involving both hands and pre-axial polydactyly involving both feet.

Case Report

A 40-year-old gravida 3 para 1 with an obstetric history of an aspiration curettage and a caesarean section was admitted to our hospital for continued prenatal care. The woman related that she underwent a surgical procedure in early childhood due to post-axial polydactyly in both hands. Routine first trimester tests, including a blood count, urine analysis, urine culture, TORCH screening, and first trimester trisomy screening, were all within normal limits. She was also informed about familial polydactyly. At 20 weeks gestation, an ultrasound examination revealed pre- and post-axial polydactyly in both fetal hands. Also, pre-axial polydactyly and syndactyly were noted involving both feet. The couple were counseled and offered karyotype analysis. The amniocentesis revealed a normal karyotype and there was no other ultrasonographic abnormalities. The prenatal care continued as scheduled, and at 38 weeks gestation, the patient delivered a 3600 gr baby by caesarean section (Figure-1,2,3,4). After delivery the newborn underwent a complete physical examination and no other abnormalities were found.

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Makalenin Geliş Tarihi : 19.04.2010 Kabul Tarihi : 04.08.2010



Figure-1. Right hand with pre- and post-axial polydactyly.



Figure-2. Left hand with pre- and post-axial polydactyly.



Figure-3. Left foot with pre-axial polydactyly and syndactyly.

Discussion

The etiology of polydactyly can be classified as environmental and genetic. The genetic abnormalities are mostly single gene diseases (3). Mutations in the *GLI3* gene are associated with post-axial polydactyly in a

single family (4) and other loci for this disease have been identified on chromosomes 7q21-q34 and 19p13.1-13.2 (5, 6).



Figure-4: Right foot with pre-axial polydactyly and syndactyly.

Most inherited cases of polydactyly involving the hand and foot are of autosomal dominant inheritance; however, a few syndromic cases, such as Meckel-Gruber syndrome, short-rib-polydactyly syndrome, Smith-Lemli-Opitz syndrome, Joubert syndrome, and Mohr syndrome, are autosomal recessive. Environmental factors include maternal diabetes and drug use, such as azathioprine and valproic acid.

Castilla et al. (2) stated that congenital anomalies, such as cardiac and brain malformations are accompanied by polydactyly in 14.6% of cases. These concomitant structural malformations may help the practitioner to make the diagnosis earlier, however, isolated polydactyly is often missed prenatally. In grayscale, polydactyly has to be confirmed in both axial and coronal views as oblique views may give erroneous appearance. Currently, the best imaging tool is 3D ultrasound with surface mode reconstruction, which is a valuable tool for evaluation of the hands and feet.

Surgical correction of bifid digits (especially the thumbs) in pre-axial polydactyly is almost always indicated, not only for cosmetic improvement, but also for enhanced function. Surgical intervention is generally performed when the child is about 18 months of age, but no later than 5 years of age. Later revisions needed for angular deformities and instability may be performed at 8-10 years of age. In post-axial polydactyly, in the case of duplication of soft parts only (type 1), the use of ligatures (commonly used) around the base of the duplication is not recommended due to reports of fatal haemorrhage. In cases of partial duplication of digits, including the osseous structures (type 2) and complete duplication, in addition to the metacarpals (type 3), the extra digits should be excised through an elliptical incision. The procedure should be performed when the child is about 1 year of age (7).

In conclusion, as polydactyly may be a part of a syndrome and 14.6% concomitant congenital abnormalities occur, the fetus should be examined ultrasonographically for other malformations, genetic counseling should be conducted and karyotype analysis should be offered, even if it is an isolated anomaly.

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