

İzole Postaksiyal Polidaktili Tip A/B'li Bir Çocuk: Olgu Sunumu

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ÖZET:

Postaksiyal polidaktili (PAP) ellerin ve/veya ayakların beşinci parmağının duplikasyonları ile karakterizedir. İki fenotipik çeşit tanımlanmıştır. Tip A, ekstra parmak tam gelişmiştir. Tip B, beşinci parmağın rudimenter bir eki ile karakterizedir. İzole PAP genellikle otozomal dominant kalıtmıdır. Biz burada her iki elinde ve ayağında total 24 parmağa sahip izole PAP'lı nadir bir olguyu rapor ettik.

Anahtar Kelimeler: Postaksiyal polidaktili, doğumsal defektler

SUMMARY:

Isolated Postaxial Polydactyly Type A/B in A Child: Case Report

Postaxial polydactyly (PAP) is characterized by fifth digit duplications in hands and/ or feet. Two phenotypic varieties have been described. In type A, the extra digit is fully developed. A rudimentary extra fifth digit characterises type B. Isolated PAP is usually autosomal dominant inheritance. Herein we reported a rare case with isolated PAP who had both hands and feet with 24 digits in total.

Key words: Postaxial polydactyly, birth defects

INTRODUCTION

Polydactyly, a very frequent malformation, is clinically characterized by the appearance of an extra digit in the hands and/or feet, when there is excessive radial division, or large or bifid digit when division is incomplete (1). These anomalies are classified as postaxial or preaxial polydactylies according to their respective manifestation on the ulnar/fibular or radial/tibial face of the hands or feet; and may concomitantly occur on both limbs and in both sides of the body (2). Polydactyly can rare occur sporadically but it can also be inherited with in many cases an autosomal dominant pattern of inheritance. Many genes have been associated with limb malformations in human (3). In rare cases, polydactyly may be caused by external factors like exposure to toxins (4).

CASE REPORT

A 1-month-old baby girl was the first child of an unrelated couple. She was born following a normal gestation and delivery. The baby was 52 cm tall and weighed 4100 gr (Fig.1). She had the extra fifth digit (PAP-A) in left hand and feet (Fig 2a, 3a-b) and a rudimentary

extra fifth digit (PAP-B) in right hand (Fig. 2b). The baby was examined carefully to exclude other congenital deformities. Fig. 1 Baby with postaxial polydactyly



Fig. 2a Postaxial polydactyly-A in left hand.



Fig. 3 a,b Bilateral postaxial polydactyly-A in feet.**Fig. 2b** Postaxial polydactyly-B in right hand.

Ophthalmic examination revealed no pathology. Ultrasonography depicted no abnormalities of internal organs. The proband showed a normal karyotype. Because of the departmental resources was inadequate, we could not perform genetic loci analysis in the case. The family history was unremarkable.

DISCUSSION

Polydactyly can be generally subclassified into preaxial and postaxial forms according to its anatomical location (5). Most cases of polydactyly of the hand and foot were postaxial, and preaxial involvement was uncommon (6). Postaxial polydactyly (PAP) is characterized by duplications of the fifth digit in hands and/or feet. Two phenotypic varieties have been described, in type A (PAP-A); the extra digit is well formed and articulates with the fifth or an extra metacarpal. Type B (PAP-B) is characterised by rudimentary extra fifth digit and is usually represented by an extra skin tag (pedunculated postminimus) (2). The incidence varies from 1/3300 to 1/630 and from 1/300 to 1/100 livebirths in Caucasian- and African-Americans, respectively (7). As to the PAP, Castilla *et al.* (8) studied 1,733 cases in Latin America during 26 years, and reported an association of black African ancestry, males, twin pregnancy, low education level of

mothers, and parental consanguinity, with frequent familial recurrence. The present study did not show any significant association between these characteristics and polydactyly. PAP is either seen as an isolated malformation or associated with other defects. Associated defects can be restricted to the limbs. If not, they can be part of a syndrome, or of a multiple congenital anomaly case (9). The syndromic cases have a heterogeneous aetiology, for instance trisomy 13. Partial cutaneous syndactyly between toes 2–3, 4–5, and other, is a frequent finding in individuals with PAP (9). Isolated PAP usually segregates as an autosomal dominant trait, with variable penetrance and expression. Penetrance rates of 0.68 and 0.43 have been estimated for types A and B, respectively (10), although higher estimates have been published (11).

The aetiology of polydactyly can be classified into environmental and genetic. Congenital hand malformations including polydactyly are mostly single gene diseases (12). Currently, there are four loci for isolated PAP in humans. PAP-A1 has been described in three families with PAP-A/B with a disease-causing mutation in the human transcription regulator *GLI3* gene on 7p13.6 (13-15). PAP-A2 on 13q21–q32 has been reported in a Turkish kindred with PAP-A only (16). PAP-A3 has been published in a Chinese family with PAP-A/B on 19p13.1–13.2 (17). In another study, Galjaard *et al.* (18) found a fourth locus in a family with PAP-A/B and partial cutaneous syndactyly on 7q21–q34. In this family there are patients with three types of PAP (type A, B or A/B) with or without syndactyly. Most inherited cases of polydactyly involving the hand and foot are of autosomal dominant inheritance but still a few syndromic cases (Smith-lemlie-opitz syndrome, Meckel-gruber syndrome, Short rib-polydactyly syndrome type I and II, Ellis-van Creveld syndrome, Carpenter syndrome and Bardet-Biedl syndrome), are of autosomal recessive inheritance (19). The hereditary aspect of this malformation behaves differently according to geographical and consequently racial variations; as a result, the occurrence of extra digits manifests itself in varied ways (1).

The extra digit is removed for the treatment of postaxial polydactyly. Surgical removal of the supernumerary digit is provided to improve cosmetic appearance. In this study, we present a child with 24 digits in both hands and feet.

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