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TO EXCISE OR NOT TO EXCISE AN EXTRANUMERARY DIGIT IN SYNPOLYDACTYLY? A CASE REPORT WITH A 20 YEAR FOLLOW-UP

Małgorzata Miedzybłocka
Piotr Czarnecki
Leszek Romanowski

Department and Clinic of Traumatology, Orthopaedics and Hand Surgery, Poznan University of Medical Sciences, Poznan, Poland

SUMMARY

Introduction

Synpolydactyly is a congenital limb defect characterized by mesoaxial fusion of fingers and postaxial fusion of toes with accompanying digital duplication in the fused interdigital webbing. We present a case of a male patient with bilateral synpolydactyly who underwent surgical correction in infancy. Left hand was treated with syndactyly separation alone while on the right extra interdigital phalanges were removed in addition to syndactyly separation. At age 24 patient's hand functionality was reevaluated.

Aim

To compare patient's hand functionality bilaterally and over time.

Materials and methods

All prior existing patient records were reviewed for information regarding clinical evaluations, surgical interventions and rehabilitation. Current clinical assessments were carried out in alike manner, allowing for bilateral and then-and-now hand functionality comparison.

CZY RESEKOWAĆ NADLICZBOWY PALEC W SYNPOLIDAKTYLII? OPIS PRZYPADKU NA PODSTAWIE OBSERWACJI PO 20 LATACH

Małgorzata Miedzybłocka
Piotr Czarnecki
Leszek Romanowski

Katedra i Klinika Traumatologii, Ortopedii i Chirurgii Ręki, Uniwersytet Medyczny im. Karola Marcinkowskiego w Poznaniu

STRESZCZENIE

Wstęp

Synpolidaktylia jest wadą wrodzoną kończyn charakteryzującą się zrośnięciem palców środkowego i serdecznego ręki oraz czwartego i piątego stopy. Dodatkowo wewnątrz zrostu znajdują się elementy kostne nadliczbowego palca. Prezentujemy przypadek pacjenta z obustronną synpolidaktylią skorygowaną chirurgicznie w okresie wczesnodziecięcym. W obrębie ręki lewej wykonano jedynie rozdzielenie palcozrostu, natomiast w obrębie ręki prawej rozdzielono palcozrost oraz wyresekowano elementy kostne nadliczbowego palca. W wieku 24 lat pacjent zgłosił się ponownie celem kontroli funkcji operowanych palców.

Cel

Porównanie funkcjonalne operowanych palców obustronnie oraz w czasie.

Materiał i metody

Przeanalizowano historie choroby z wszystkich poprzednich hospitalizacji pacjenta pod kątem badań przedmiotowych, interwencji chirurgicznych i rehabilitacji. Oceny kliniczne i radiologiczne podczas aktualnej hospitalizacji przeprowadzono w sposób umożliwiający porównanie otrzymanych

Results
Age 3: LH – Middle finger ROM: PIP 5–35˚, DIP 0–20˚; Ring Finger ROM: PIP 0˚, DIP 0˚. RH – Middle finger ROM: PIP 5-7˚, DIP 0–10˚; Ring Finger ROM: PIP 5-30˚, DIP 5–25˚. Significant lateral axis deviation deformity of distal and middle phalanges of left and right ring digits respectively.

Age 24: LH – Middle finger ROM: PIP 5–7˚, DIP 7-9˚; Ring Finger ROM: PIP 0˚, DIP 0˚. RH – Middle finger ROM: PIP 5–7˚, DIP 5–7˚; Ring Finger ROM: PIP 15–16˚, DIP 25˚ flexion contracture. Significant lateral axis deviation deformity of distal and middle phalanges of left and right ring fingers and of distal phalanx of right middle finger.

Conclusion
Unexcised interdigital phalanges hindered ROM, but likely provided stability to IP joints preventing axial deformity and resultant functional loss.

Keywords: polydactyly; syndactyly; limb malformation; HOXD13

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Wyniki
Wiek 3 lata: KGL – palec środkowy ROM: PIP 5–35˚, DIP 0–20˚; palec serdeczny ROM: PIP 0˚, DIP 0˚. KGP – palec środkowy ROM: PIP 5–7˚, DIP 0–10˚; palec serdeczny ROM: PIP 5–30˚, DIP 5–25˚. Znaczne odchylenie osiowe paliczków dalszych lewego palca serdecznego oraz paliczków środkowych prawego palca serdecznego.
Wiek 24 lata: KGL – palec środkowy ROM: PIP 5–7˚, DIP 7–9˚; palec serdeczny ROM: PIP 0˚, DIP 0˚. KGP – palec środkowy ROM: PIP 5–7˚, DIP 5–7˚; palec serdeczny ROM: PIP 15–16˚, DIP 25˚ przykurczu zgięciowego. Znaczne odchylenie osiowe paliczków dalszych i środkowych obu palców serdecznych oraz paliczka dalszego prawego palca środkowego.

Wnioski
Niezresekowane dodatkowe paliczki w ręce lewej utrudniają zakres ruchu palców środkowego i serdecznego, ale najprawdopodobniej zapewniają stabilność stawom IP uniemożliwiając ich osiową deformację skutkującą utratą funkcji.

Słowa kluczowe: polidaktylia; syndaktylia; deformacje kończyn; HOXD13

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Introduction

Synpolydactyly (SPD), or syndactyly type II is a rare, dominantly inherited congenital limb malformation characterized by a joint presentation of syndactyly (fusion of digits) and polydactyly (duplication of digits). The phenotypic expression of SPD is highly variable with a broad spectrum of clinical features (Wall *et al.* 2016). One to all four limbs can be involved, however the manifestation is never present in the feet, unless the hands are also affected (Malik and Grzeschik 2008). The severity of involvement ranges from partial skin syndactyly to complete digital duplication (Goodman and Scambler 2001). Typically, SPD is manifested by bilateral webbing between the middle and ring finger and between the fourth and fifth toe (Brison *et al.* 2012), with partial or complete digit duplication within the syndactylous web (Malik and Grzeschik 2008). Additionally, there may also be little finger camptodactyly, clinodactyly or brachydactyly as

well as variable syndactyly of the second to fifth toes with middle phalangeal hypoplasia (Goodman and Scambler 2001). Other minor local anomalies and various metacarpal or metatarsal abnormalities may also be present (Merlob and Grunebaum 1986).

Although the incidence of syndactyly and polydactyly has been well researched and documented, no reliable epidemiological estimates of incidence of syndactyly combined with polydactyly have ever been published (Tian *et al.* 2011). The most extensive epidemiological report to date, in which the authors attempted to calculate synpolydactyly frequency in the general population based on a cohort of over 4.000.000 polydactyly cases has been published in 1996 (Castilla *et al.* 1996). In this study Castilla and colleagues proposed an overall synpolydactyly birth prevalence rate of 0.4 based on their cohort finding of 15 synpolydactyly cases in total, all non-syndromatic and with no associated anomalies. Still, most researchers do not accept this prevalence as being truly representative of a world-wide synpolydactyly occurrence due to the fact that the study was limited to data collection in just two geographical regions. Moreover the birth prevalence rate differed significantly between those two areas (0.5 vs. 0.1) which may suggest an ethnic factor.

Despite lack of precise estimates, synpolydactyly is known to be less frequent than either syndactyly or polydactyly (Tian *et al.* 2011). It is officially listed as a „rare disease" by Orphanet, a European rare disease database. In the European Union the European Medicines Agency currently defines a condition rare when it affects no more than 5 people in 10.000 (Richter *et al.* 2015).

Inherited in an autosomal dominant fashion synpolydactyly is clinically and genetically one of the most heterogeneous malformations (Brison *et al.* 2012), showing incomplete penetrance and variable, asymmetrical expressivity. There are three known, genetically distinct types of SPD with identified gene loci (Malik and Grzeschik 2008). Synpolydactyly type 1 (SPD1) is associated with a 2q31.1 locus mutation, synpolydactyly type 2 (SPD2) is linked with a defect on the 22q13.31 locus, and synpolydactyly type 3 (SPD3) is localized the 14q11.2-q12 locus abnormality (Wall *et al.* 2016). SPD1 is caused by mutations in the HOXD13 gene, a key regulator of limb development. These mutations fall into two groups: (1) polyalanine expansions which cause typical SPD, and (2) frameshifting deletions which are responsible for atypical forms of SPD (Zhou *et al.* 2013). In „classical" SPD, polyalanine expansions vary in number, in most cases, from 7 to 10 (the largest found to date being 14) (Goodman and Scambler 2001). The larger the expansion, the more complete the penetrance, and the more severe the phenotype. Typical SPD patient is heterozygous. Homozygous subjects have uniquely different phenotypic expression (Yucel *et al.* 2005) and for that reason this condition is normally excluded from typical SPD classification and is considered as a separate entity.

SPD exhibits incomplete penetrance estimated by various authors (Akarsu *et al.* 1996; Malik and Grzeschik 2008; Quinonez and Innis 2014) to reach 97% with the remaining individuals expected to be phenotypically normal gene carriers (Akarsu *et al.* 1996). The condition is also characterized by variable expressivity, including both inter-and intra-familial variability (Quinonez and Innis 2014). In addition, clinical expression can skip a generation. Therefore SPD-affected parent can produce phenotypically normal offspring, as in a documented case of a woman who inherited the entire affected chromosome from her affected father and subsequently passed it on to her affected son, but herself was phenotypically normal (Akarsu *et al.* 1996).

Despite past attempts (Malik and Grzeschik 2008) to develop a uniform classification scheme for synpolydactyly, no widely accepted classification system exists, rendering characterization of its varied

phenotypic manifestations to remain largely descriptive. Aiming to facilitate reporting and treatment outcomes of SPD cases, Wall and colleagues developed a new radiographic classification of typical SPD phenotypes based on the most proximal level of skeletal involvement, characteristic features and associated anomalies (Wall *et al.* 2016). According to the authors Type 1 begins at the metacarpal level, Type 2 at the level of the proximal phalanx and Type 3 at the level of the middle or distal phalanx. Both Types 1 and 2 consists of A and B subtypes. Type 1A is characterized by a distal bifurcation of the 3rd metacarpal with synpolydactyly affecting the middle and ring fingers. Key feature of Type 1B is an extranumerary 3-or 4-boned digit syndactylized to the ring finger or both the ring and middle fingers. This digit begins proximal to the metacarpophalangeal joint level, but the metacarpal is not bifurcated. Distinguishing feature of Type 2A is delta proximal phalanx of the ring finger. Moreover the ring finger is duplicated and syndactylized to the middle finger. Type 2B consists of parallel or divergent duplication of ring or middle fingers is combined with syndactyly between these fingers. A delta phalanx is not present. In Type 3 duplication occurs between syndactylized ring and middle fingers.

A case of a male patient with tetrasynpolydactyly who underwent surgical correction of upper extremity synpolydactyly in infancy is presented.

Aim

To compare patient's hand functionality bilaterally and over time in view of different surgical approach to each hand.

Material and methods

The patient first presented to our department at the age of 8 months with congenital finger and toe malformations on all extremities. All other body parts were developed normally. Clinical findings included complete bilateral syndactyly between the long and ring fingers and no sign of fingernail dysmorphia of the syndactylized digits. Radiographic imaging (X-ray) revealed complete digital duplication within the syndactylous web of both hands. The extranumerary interdigital phalanges were thinner (on the right) and shorter (on the left) than their counterparts in either long or ring fingers. Bilateral apical synostosis involving the distal phalanges of the extranumerary and ring fingers was also present. There was no polydactylous skeletal involvement past the level of the proximal phalanx (Figure 1). The remaining fingers of each hand were unaffected by the condition.

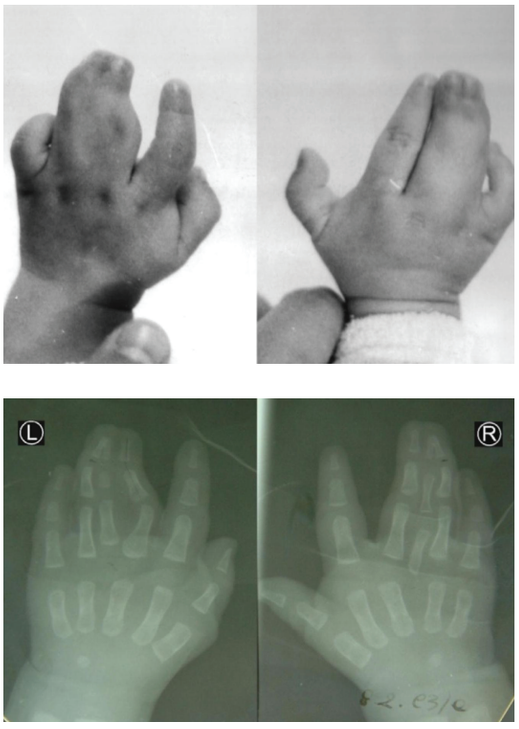

Figure 1. Macroscopic and radiographic images of patient's hands at 8 months old.

Clinical examination of the lower limbs revealed six toes on each foot with syndactyly of toes four, five and six. Toenails of all syndactylized digits were shaped normally. Radiographic imaging showed proximal phalangeal synostosis of fifth and sixth toes with shortened and triangularly shaped fifth metatarsal and a hypoplastic sixth

metatarsal in the left foot and fifth toe phalangeal hypoplasia with a distally bifid (y-shaped) fourth metatarsal in the right foot. Tarsal bones were normal bilaterally. Toes one, two and three on each foot were unaffected by the condition.

According to the patient's caregivers, this congenital abnormality has been present in the family for five generations – patient's mother had a similar deformity, while his grandmother was autosomally dominant for the SPD causing gene. The patient history was otherwise uneventful – he was delivered at term, with no prenatal complications and at the time of admission and was reaching all developmental milestones on schedule.

with syndactyly separation in 1994 in 1995. At the age 24 he was readmitted to our department, seeking to improve functionality and the overall cosmetic appearance of his hands. The patient reported no medical concerns regarding his feet.

In this twenty year gap between admissions, the patient denied having undergone any additional treatment for his condition, surgical or otherwise. Physical examination revealed significant functional impairment, especially flexion at both interphalangeal joints of middle and ring digits bilaterally. On the left the ring finger ROM has been compromised by the remaining interdigital phalanges, while middle finger ROM

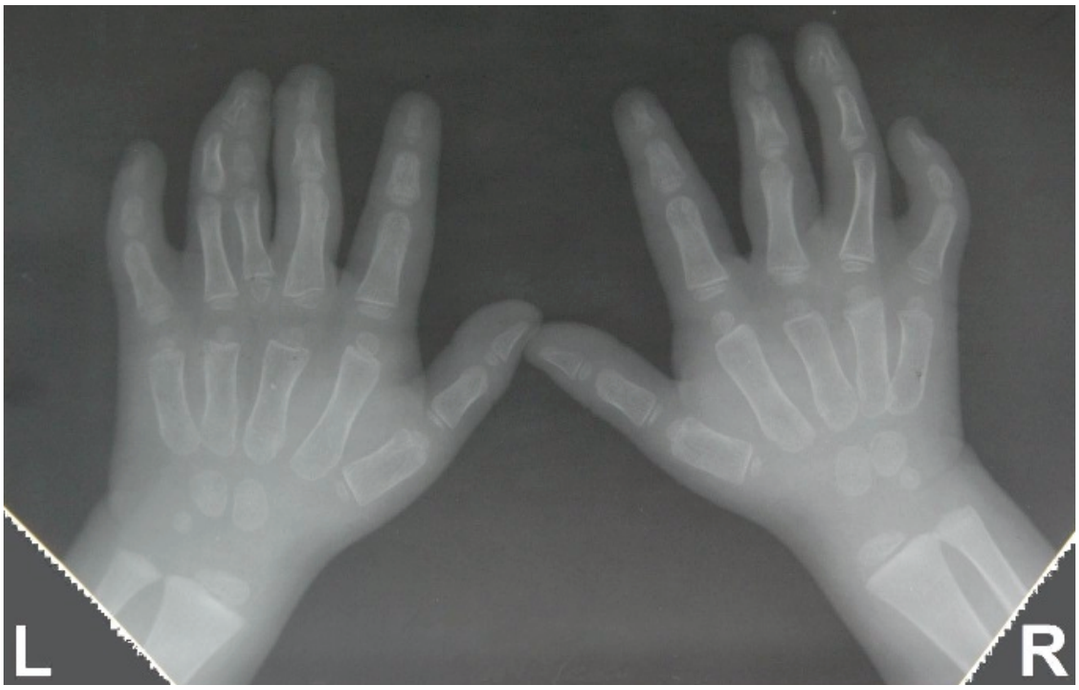

Figure 2. Radiographic images of patient's hands at 3 years old following syndactyly separation with and without the removal of interdigital phalanges in the right and the left hand respectively.

In the first 3 years of his life the patient underwent several surgical procedures in our department: (1) left hand syndactyly separation without removal of extra interdigital phalanges in 1993 (2) right hand syndactyly separation with interdigital phalanges removal in 1994 (Figure 2) and (3) bilateral amputation of extranumerary toe

has been hindered by arthrosis, namely at the DIP joint. On the right the ring finger ROM of the PIP and the DIP joint has been compromised due to joint instability (subluxation) and arthrosis respectively, while middle finger ROM has been hindered by arthrosis, namely at the PIP joint. There are also constricted bands of post-operative

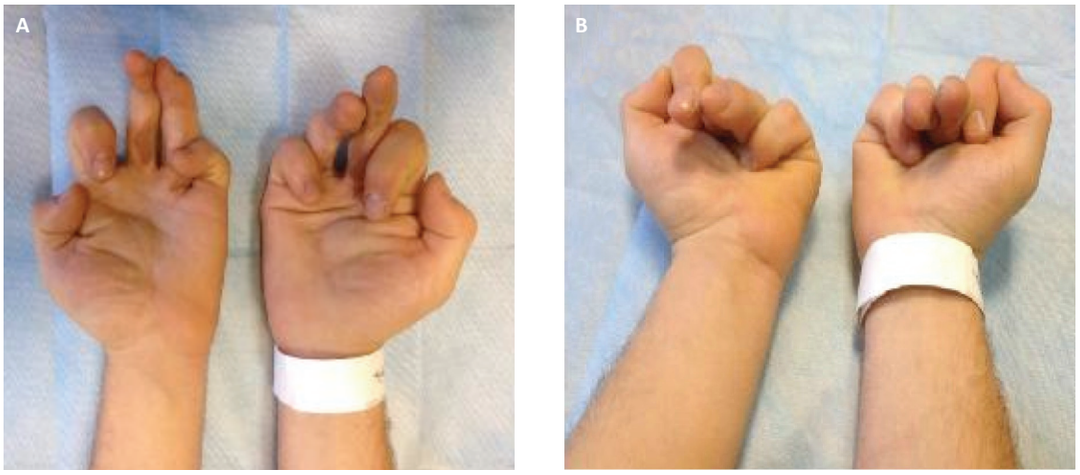

Figure 3. Compromised ROM, especially flexion of affected digits. (A) flexion at the PIP and DIP joints, (B) flexion at the MP, PIP and DIP joints.

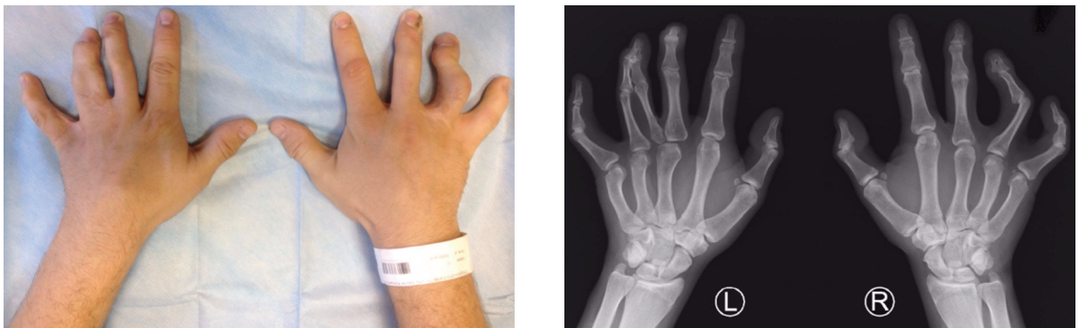

Figure 4. Macroscopic and radiographic images of patient's hands at 24 years old.

scar tissue in web spaces between middle and ring fingers, which may further limit movement (Figure 3). Both middle fingers appeared grossly normal in shape, but were shorter and thinner than the unaffected index counterparts, with the size discrepancy decidedly more conspicuous in the left hand (Figure 4). There are no sensory deficits in any of the affected fingers. The other fingers reveal no pathology. The patient is in good general health with no comorbidities. He works as a mechanic and reports that poor finger function interferes with his job performance and overall quality of life.

Materials and methods

All prior existing patient records were reviewed for information regarding clinical evaluations, surgical interventions and rehabilitation. Current clinical assessments were carried out in alike manner, allowing for bilateral and then-and-now hand functionality comparison.

Results

Physical and radiological findings gathered during patient's last two consecutive admissions to our department are presented in tables below.

Table 1. Digital ROM comparison bilaterally and over time.

		Left Hand			Right Hand		
	Digit	MCP	PIP	DIP	MCP	PIP	DIP
Age 3	middle	N	5–35˚	0–20˚	N	5–7˚	0–10˚
	ring	N	0˚	0˚	N	5–30˚	5–25˚
Age 24	middle	N	5–7˚	7–9˚	N	5–7˚	5–7˚
	ring	N	0˚	0˚	N	15–16˚	25˚ FC

N = normal, FC = flexion contracture

Table 2. Joint deformity comparison bilaterally and over time.

		Left Hand			Right Hand		
	Digit	MCP	PIP	DIP	MCP	PIP	DIP
Age 3	middle	N	6˚ VL	10˚ VR	N	10˚ VR	11˚ VL
	ring	N	10˚ VR	40˚ VR	N	20˚ VR	N
Age 24	middle	I	3˚ VL	12˚ VR Severe Arthrosis	N	10˚ VR Severe Arthrosis	19˚ VL Arthrosis
	ring	N	20˚ VR Arthrosis	32˚ VR Arthrosis	N	65˚ VR	30˚ VL Arthrosis

N = none, I = incomplete joint surface overlap, VR = varus (inward), VL = valgus (outward)

Discussion

Synpolydactyly is a rare congenital limb anomaly with a broad spectrum of phenotypic variability (Wall *et al.* 2016). Although the condition is usually evident upon visual examination, its variable expressivity presents considerable challenge in determining the type of SPD during the diagnostic process.

Based on our subject's clinical presentation we classify his condition as typical heterozygous synpolydactyly with a large polyalanine expansion. The following features justify this diagnosis:

1. Presence of cardinal phenotypic features of „classical" SPD:
   a. malformations are located mesoaxially in the hands (middle and ring finger) and postaxially in feet (fourth and fifth toe)
   b. thumb, index and little fingers as well as toes one, two and three are unaffected
   c. webbing is complete, reaching the tips of involved digits (Malik and Grzeschik 2008)
2. Condition severity is less debilitating than that of homozygous counterparts which present with short and severely malformed hands and feet as well as with polydactyly of the preaxial, mesoaxial and postaxial digits of the hands (Akarsu *et al.* 1995).
3. Bilateral hand and foot involvement correlating with a large poly (A) expansion

In addition, using radiographic classification developed by Wall *et al.* we determined that our patient most closely fits into Type 2B.

The treatment objective is to provide the SPD affected patient with a limb that is both functional and cosmetically acceptable. However, phenotypic variability and complexity of skeletal and soft-tissue anomalies in synpolydactyly cases pose a great challenge in surgical decision-making (Wall *et al.* 2016). Furthermore, due to its rarity published information regarding its treatment is scarce. Therefore most authors „lump" synpolydactylous hand malformations with the other types of syndactyly and recommend surgical approach identical to that for central syndactyly. Recommendations regarding treatment of supernumerary digits may be found in publications regarding polydactyly treatment. Authors are in agreement that in cases of central syndactyly and/or polydactyly early surgical intervention is crucial. Osteotomy and ligament

reconstruction should be performed to prevent angular growth deformities. It is equally important to surgically release any syndactyly which may hinder motion of the affected fingers (Wolfe *et al.* 2017). Most surgeons recommend surgical correction of aforementioned malformations between 6 and 12 months of age and certainly no later than at 18 months. In complicated cases multiple surgeries may be required.

In the first three years of his life our patient underwent several corrective surgical procedures of his hands and feet. While the syndactylous fusion and duplication of toes received definite treatment (bilateral amputation of extranumerary toe and syndactyly separation), the management of hand malformations differed between left and right limb. On the right, staff surgeons performed syndactyly separation with interdigital phalanges removal, but on the left syndactyly separation was the only surgical correction performed. This unusual approach gave the authors a rare opportunity to examine and compare patient's hand function, two decades after the surgical intervention. According to patient records from past admissions, at three years of age the patient had a relatively good range of motion in treated fingers of the right hand, where supernumerary digit was removed, but no movement in the PIP and DIP joints of the duplicated left ring finger. Now 24 and seeking to improve functionality of his left hand the patient was readmitted to our department. Although surgical excision of the extranumerary digit my improve cosmetic appearance it is unlikely to significantly improve finger function as affected PIP and DIP joints have undergone deformity unamendable to surgical correction. Authors agree that early intervention is of great importance to maintain proper joint movement.

Additionally, regular postoperative follow-ups are crucial for SPD patients, especially for those who experience functional deterioration or secondary deformities in their operated limbs. In the twenty years since his last surgery our patient has not been in for follow-up nor has he been consulted by any of our staff, thus allowing the lateral axis deviation deformity in the operated digits, namely the right ring finger to progress unhindered. Delay in treatment has caused irreversible bony and soft tissue deformities which are no longer fully amenable to surgery or rehabilitation and will continue to impair his hand function and cause life quality decrease.

Conclusion

At age 3 unexcised interdigital phalanges hindered ROM, while the corresponding right digit had a 30 degree ROM in both IP joints. Over time ROM decreased in IP joints of all affected digits, most profoundly in the PIP joint of left middle finger (arthrosis) and PIP and DIP joints of right ring finger (axial deformity). Currently there is no functional movement in either ring finger, but the right digit has a decidedly worse cosmetic appearance as compared to its left counterpart.

Unexcised interdigital phalanges hindered ROM, but likely provided stability to IP joints preventing axial deformity and resultant functional loss and poor cosmetic outcome.

Still excision of extradigit in these cases is debatable and it is not possible to clearly state which technique is more appropriate. As we present a case report this issue should be further investigated and conclusions drawn based on case series.

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Author responsible for correspondence:
Małgorzata Miedzybłocka
Department and Clinic of Traumatology, Orthopaedics and Hand Surgery, Poznan University of Medical Sciences
miedzyblocka.m@gmail.com

Autor odpowiedzialny za korespondencję:
Małgorzata Miedzybłocka
Katedra i Klinika Traumatologii, Ortopedii i Chirurgii Ręki, Uniwersytetu Medycznego im. Karola Marcinkowskiego w Poznaniu
miedzyblocka.m@gmail.com