

Syndactyly as Symptome or Part of Plurimalformative Syndrome in Pediatric Patology. Clinical and Therapeutical Considerations

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Background: Syndactyly is the most common congenital malformation of the limbs. Syndactyly can be classified as simple when it involves soft tissues only and classified as complex when it involves the bone or nail of adjacent fingers. Syndactyly can occur as isolated condition or in conjunction with other symptoms as one aspect of a multi-symptom disease.

Aim: The author's purpose is to present this condition in hospitalized patients in order to make some considerations about the frequency of association with other anomalies and the treatment of this condition.

Material and methods: Between 2000–2009, 83 cases of hand malformations were diagnosed and treated at Plastic Surgical Department of Children Hospital Brasov and Fogolyan Kristof Hospital Sfantu Gheorghe. Observational retrospective study on this group found that 39 of these were syndactyly and 44 polydactyly (control group).

Results: We have found 2 cases of sinpolydactyly and 12 cases of plurimalformation. The Apgar score as well the birth weight of children with plurimalformations were lower than of those with simple syndactyly ($p = 0.0153$). The average age of surgical intervention was 3.370 years ($SD = 4.267$, $p = 0.0001$). The hand malformation was bilateral in 26 cases. Out of the 39 cases of syndactyly, 17 needed full-thickness skin graft.

Conclusions: The goal of syndactyly release is to create a functional hand with the fewest surgical procedures while minimizing complication. Reconstruction of the web commissure is the most technically challenging part of the operation, followed by separation of the remaining digits.

Keywords: syndactyly, Poland syndrome, constriction band syndrome, epidermolysis bullosa

Introduction

Syndactyly is the most common congenital malformation of the limbs, with an incidence of 1 in 2000–3000 live births [1,2]. Syndactyly can be classified as simple when it involves soft tissues only and classified as complex when it involves the bone or nail of adjacent fingers. It is a shared feature of more than 28 syndromes, including Poland, Apert and Cohen syndromes. Syndactyly is a failure of the programmed cell death in which the fingers separate into individual appendages. This separation usually occurs during the sixth and eighth weeks of embryologic development. The root words of the term syndactyly are derived from the Greek words syn, meaning together, and dactyly, meaning fingers or digits.

Syndactyly can occur as isolated condition or in conjunction with other symptoms as one aspect of a multi-symptom disease. There are several forms of isolated syndactyly due to an autosomal dominant (not sex-linked) gene; males and females are equally likely to inherit the trait.

Syndactyly often can be associated with other syndromes [3], particularly the craniofacial syndromes, of which the Apert syndrome is the best known. Another syndrome is the Poland syndrome [4].

The Poland's syndrome is a rare congenital malformation characterized by three permanent unilateral deformities [5,6,7,8,9] simple syndactyly located on the 2-3rd

fingers on hypoplastic hand, brachydactyly and ipsilateral aplasia of the pectoralis major muscle.

The incidence of this anomaly is about 1 in 30,000 live births, more frequently in the case of males and the right side.

Constriction band syndrome can be associated with syndactyly, but the etiology of the syndactyly is different then in previously mentioned syndromes. The syndactyly found in the constriction band syndrome (also known as amniotic band syndrome [10]) is not the result of failure of differentiation during embryogenesis. In this condition, the fingers are already formed, and because of the injuries due to the constricting amniotic bands, the fingers heal together at the site of injury, causing postinjury syndactyly [4]. The extent of the involvement may be mild, with only a rudimentary small skin bridge connecting the 2 fingers.

Apert syndrome, or acrocephalosyndactyly, is a rare anomaly, occurring in 1 of 160,000–200,000 live births [11,12]. In nearly all patients with Apert syndrome, the cause is 1 of 2 FGFR2 mutations involving amino acids (Ser252Trp, Pro253Arg). The condition is inherited in an autosomal dominant mode. Craniosynostosis is present, characterized by brachycephaly and, frequently, turriccephaly; the anterior fontanelle is enlarged [13,14]. The maxilla is hypoplastic with a high-arched palate, class III malocclusion with an anteriorly open bite, and frequently,

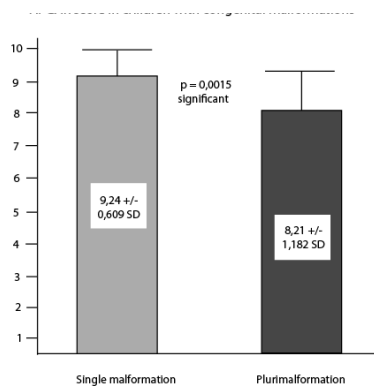


Fig. 1. The Apgar score in children with malformations

a cleft of the soft palate. The mid face is hypoplastic. This, together with the retrusion, causes exorbitism. Besides the characteristic facial anomalies of the Apert syndrome, it is also defined by the associated upper- and lower-limb anomalies. The anomalies in the hands [11] are mirror images of each other and are characterized by the following 4 common features: radial deviation of a short thumb as a result of an abnormally shaped proximal phalanx (ie, delta phalanx); complex syndactyly of the index, long, and ring fingers; symbrachyphalangism of the central segments of the index, long, ring, and small fingers; and simple syndactyly of the web space between the ring and small fingers.

Epidermolysis bullosa (EB) is a group of inherited bullous disorders characterized by blister formation in response to mechanical trauma.

Severe recessively inherited epidermolysis bullosa, as described by Hallopeau-Siemens [15], usually shows generalized blistering at birth and subsequent extensive dystrophic scarring that is most prominent on the acral surfaces. This can produce pseudosyndactyly (mitten-hand deformity) of the hands and feet. Flexion contractures of the extremities are increasingly common with age.

The physician who diagnoses the syndactyly should look for other associated not so evident anomalies. The patient's long term prognosis is determined by the presence and stage of other anomalies.

Material and methods

Between 2000–2009, 83 cases of hand malformations were diagnosed and treated at Plastic Surgical Department of Children Hospital Brasov and Fogolyan Kristof Hospital Sfantu Gheorghe. Observational retrospective study on this group found that 39 of these were syndactyly and 44 polydactyly (control group).

We analysed the following details: birth weight, Apgar scale at birth of the newborn, epidemiology (sex, age, urban or rural setting), uni- or bilateral hand malformation, hospitalisation period, other anomalies, if the syndactyly was part of a plurimalformative syndrome and the surgical techniques which were used for web reconstruction.

The data analysis was performed using the GraphPad InStat programme, the unpaired t test with Welch correc-

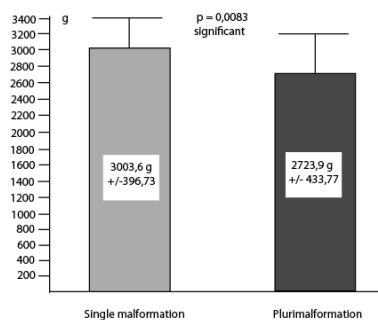


Fig. 2. The birth weight in children with malformation

tion where needed. The normal distribution of the data was analysed with the Kolmogorow Smirnov test.

Laboratory examination from blood and urine (hemogram, blood sugar level, hepatic test results, coagulation time and EKG) were all within normal range.

Radiologic comparative examinations were performed for all children.

Results

The surgical interventions were performed under general anesthesia using triangular and quadrangular flaps combination (butterfly flap). Any existing skin deficiency was closed with full-thickness skin graft.

There has been an increasing incidence over the years. As presented in a similar study, a predominance of male patients in syndactyly pathology can be observed (F:M = 25:14).

We have found 2 cases of sinpolidactyly and 12 cases of plurimalformation.

In our statistics there were 6 cases of constriction band syndrome and 5 cases of epidermolysis bullosa.

The Apgar score (Fig.1) as well the birth weight (Fig. 2) of children with plurimalformations were lower than of those with simple syndactyly (p = 0.0153).

The average age of surgical intervention (Fig. 3) was 3,370 years (SD = 4.267, p = 0.0001).

We used skin graft in 17 cases of syndactyly and in case of 22 patients Z-plastia was performed.

The hand malformation was bilateral in 26 cases.

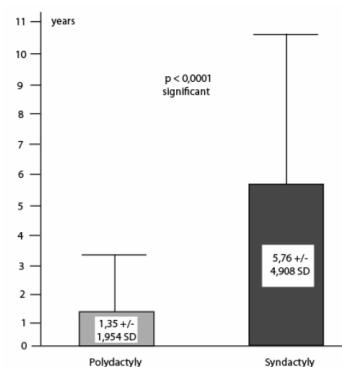


Fig. 3. The age of patients with syndactyly at the moment of surgery



Fig. 4. Complex syndactyly of 1 year old boy hands

Eighty-three cases were treated with hand malformation: 39 with syndactyly and 44 with polydactyly.

Out of the 39 cases of syndactyly, 17 needed full-thickness skin graft.

The recovery process of 82 patients presented no difficulty, while only one case had a postoperative infection.

The 6 epidermolysis bullosa pseudosyndactyly cases were treated with grassolid application and full thickness graft for web reconstruction.

Discussions

The syndactyly is visible for everyone. The timing of surgery for syndactyly depends on the complexity of the case. Pattern of hand functions are usually established [18] between 6 and 24 months of age, and any changes in the function potential will require a retraining process after this age. Syndactyly of the border digit (thumb/index finger or ring/small finger) is treated at early age to prevent the larger digit from curving towards the smaller digit with growth.

Searching for eventually associated anomalies [3,12,16] is the duty of pediatric specialist, because they may need additional investigation or treatment.

Unreleased syndactyly can significantly impair finger and hand function [4]. The impairment is worse when the syndactyly is complete, is complex (Fig. 4), or involves the border digits with fingers of uneven lengths, such as the ring and small fingers or the thumb and index finger. Approximately 10–40% of cases are familial with variable penetrance. Syndactyly can occur as part of a syndrome or as sporadic events that are nonhereditary and nonsyndromic. One report indicates that there is an association of syndactyly with smoking during pregnancy [17]. Although many patients with syndactyly have been evaluated by multiple specialists and referred by their primary care pediatrician, the hand surgeon should also obtain a thorough prenatal, postnatal, and familial history. In addition to the hand being examined, the cranium, face, torso, and lower extremities should be examined for anomalies.



Fig. 5. Brachysyndactyly of the right hand in 6 years old girl.

Surgical restoration of the epidermolysis bullosa hand [15]: Mitten deformity of the hand occurs frequently in patients with the Hallopeau-Siemens dystrophic epidermolysis bullosa subtype. Repeated episodes of blistering and scarring eventually result in fusion of the web spaces. As a result, fine manipulative skills and digital prehension are lost. Surgical procedures can correct this deformity, but a high rate of recurrence is seen with mitten pseudosyndactyly. Typically, the dominant hand has earlier recurrence. Recurrence appears to be delayed by the prolonged use of splinting in the interphalangeal spaces at night.

The hand evaluation should proceed systematically.

1. Note and document the number of digits present, the level of web involvement, the length of the finger, and the appearance of the fingernails.

2. Photographing or drawing a picture of the hands during the initial visit is helpful.

3. Passively move the fingers to determine bony union; differential motion occurs only if no underlying bony union is present. Fusions of the fingernails often are associated with bony union, and a broad fingernail also may indicate a hidden polydactyly.

4. The extent of anomaly of tendons and neurovascular structures reflects the complexity of the syndactyly. In a simple complete or a complex syndactyly involving only the distal phalanx, the underlying tendon and neurovascular structures are usually normal. However, in an individual with brachysyndactyly (Fig. 5.) or complicated syndactyly, the bifurcation of the nerves and digital vessels may be located more distally, or only one side may be present.

5. Always obtain radiographs to help identify any other anomalies, such as bony synostosis, a delta phalanx, or symphalangism.

In itself, a minor incomplete syndactyly is not an indication for surgery if the only issue is its incongruous appearance. However, a syndactyly that prevents full range of

motion in the involved fingers warrants surgical release to increase functionality of the fingers.

As with any operation, exceptions to the rule exist.

Contraindication

In individuals with complex syndactyly in whom the conjoint fingers together are functional but individually hypoplastic, separation of the conjoint fingers may make the 2 individualized digits nonfunctional, because only 1 set of tendons and 1 neurovascular pedicle may be present. Carefully consider this possibility in those few individuals who have complex syndactyly. Otherwise, most patients with syndactyly benefit from surgical release.

Conclusions

The goal of syndactyly release is to create a functional hand with the fewest surgical procedures while minimizing complication. Reconstruction of the web commissure is the most technically challenging part of the operation, followed by separation of the remaining digits. The optimal age to perform syndactyly release, especially for single-web syndactyly of nearly equal-length finger is 18 months.

The prognosis in syndactyly release may be poorer when surgery is delayed beyond age 2 years because the cerebral cortex pattern of hand use must be retrained.

Complex syndactyly and syndactyly associated with other hand or organ anomalies warrant special consideration.

Careful planning and meticulous surgical techniques can minimize potential errors and allow satisfactory separation of syndactylized digits.

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