

Symbrachydactyly Of Hand: Report Of A Case With Review Of Literature

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Citation

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Abstract

Symbrachydactyly is a uncommon congenital hand anomaly characterized by short and fused fingers. We report here an interesting case of symbrachydactyly along with the comprehensive review of the historical aspect, etio-pathology, classification, clinical features and management.

INTRODUCTION

The symbrachydactyly is one of the relatively rare congenital malformations of the hand and is literally defined as a combination of brachydactyly (short fingers) with syndactyly. The brachydactyly usually involves middle phalanges of the digits

(brachymesodactylous) while the syndactyly is predominantly cutaneous. They often manifest sporadically though familial occurrence is not uncommon. The exact cause of Symbrachydactyly is unknown but some insult to the mesenchyme during early gestational period is considered to be the causative factor. The condition usually manifests unilaterally and based on morphological features, classified ranging from simple shortness of middle phalanges to complete absence of the digital rays. The management depends upon the severity of the manifestation and the options range from reassurance to complex surgical reconstructions to improve the digital length for enhanced mechanical advantage and prehension.

CASE REPORT

A 6 year old male child presented to us with congenitally shortened left hand with fused index, middle and ring fingers. (Figures 1, 2)

Figure 1

Figure 1.



Figure 2

Figure 2



The other hand and both the feet were normal. There were no associated cardiovascular, gastrointestinal or genitourinary system anomalies. The antenatal history was not significant in relation to exposure to viral infection, drugs, alcohol, or radiation in first trimester. There was no sibling or family history of any hand/feet or other congenital anomalies. On examination, the child was having a hypoplasia of the left hand and was relatively small by 30% of the uninvolved hand. The index, middle and ring fingers of the involved hand were short with complete, simple syndactyly. (Figures 3, 4)

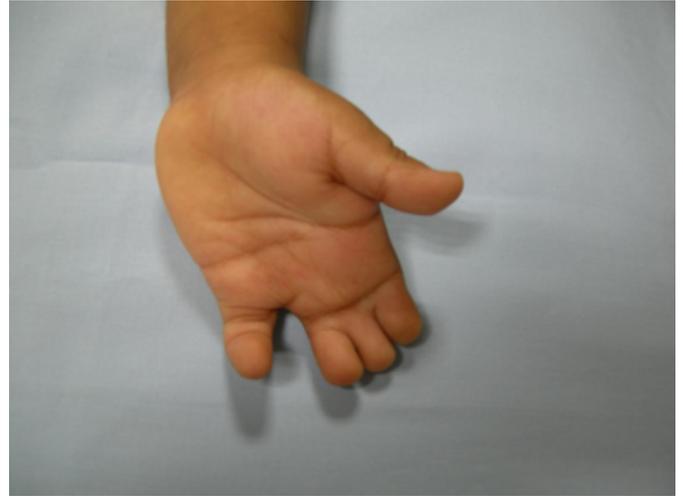
Figure 3

Figure 3



Figure 4

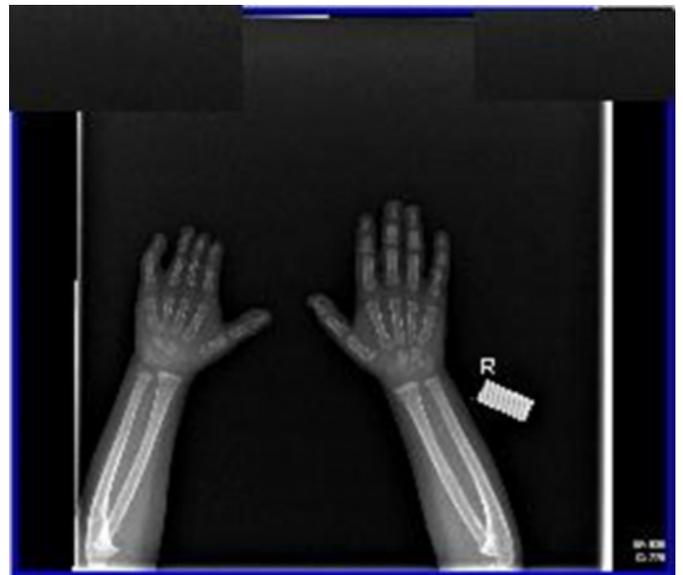
Figure 4



No constriction ring was seen in any of the fingers and the thumb, though shortened, was found to be relatively normal. The routine hematological investigations didn't reveal any abnormality. The X-ray of the affected hand revealed generalized hypoplasia of the metacarpals and phalanges with predominant involvement of the middle phalanges. (Figure 5)

Figure 5

Figure 5



DISCUSSION

Congenital hand malformations are not uncommon and have an estimated incidence of 2.3 per 1,000 of total births.^[1] They rank second after congenital heart disease as the most common birth defects observed in the infants.^[2] These anomalies can occur as isolated malformations, in

combination with another hand and/or foot, or as part of a syndrome.

The symbrachydactyly is an infrequently seen congenital anomaly where syndactyly is accompanied by brachydactyly. The incidence of symbrachydactyly is reported to be around around 1 in 10, 000.^[3] The condition is typically unilateral and sporadic in occurrence. If they present bilaterally; they are usually accompanied by peromelia and micrognathia (Hanhart syndrome). The condition of symbrachydactyly was first described by Poland in 1841 in a Guy's hospital report.^[4] Since then the combination of brachymesophalangy, dermal syndactyly and an associated pectoral muscle defect has been reported under the name of Poland's syndrome by various authors.^{[5],[6],[7]} Pol in 1921 mentioned the term 'Symbrachydaktylie' in his extensively written paper on 'Brachydaktylie-Klinodaktylie-Hyperphalangie und ihre Grundlagen' for the combination of phalangeal reduction and syndactyly and classified the patients into one with pectoral muscle defect and one without.^[8] Muller in 1937 suggested a spectrum in this condition ranging from a hand with brachymesophalangy to the rudimentary nubbins.^[9]

Congenital anomalies of the hand and upper extremity has been traditionally classified based on morphology and osseous anatomy with innumerable classifications been drafted over the past 100 years. Ideally, the classification should reflect the full spectrum of morphologic abnormalities, should include etiology, should be uncomplicated, easy to use, should enhance communication among the clinicians and provide not only a descriptive framework but also guide the treatment.^[10]

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(IFSSH) proposed seven categories classification based on the etio-pathogenic classification of Swanson et al. in 1976.^[11] The seven groups are: I. Failure of formation: transverse (A) or longitudinal (B); II. Failure of differentiation; III. Polydactyly; IV. Overgrowth; V. Undergrowth; VI. Amniotic band syndrome; VII. Generalized skeletal syndromes. Under this broad classification, the symbrachydactyly falls under the failure of development, failure of separation of parts and the undergrowth (Group I, II and V respectively). Blauth and Gekeler^[12] in 1971 enhanced the concept of symbrachydactyly and further classified it into four types based on clinical findings; short finger type, cleft hand type, monodactyly type and peromelia

type. The short finger type (group I) consists of shortening or aplasia of some or all middle phalanges and is combined with more or less extensive interdigital skin bridges (syndactyly). The cleft hand type (group II) includes more severe hypoplasias of one or more center rays, increasingly affecting the ulnar part of the hand and there always remain bud like excrescences of all the affected fingers. The monodactyly type (group III) comprises of hands in which every three phalangeal finger has been reduced to mere bud like rudiments with nails. In peromelia type (group IV), all that left of the fingers are hump like protuberances. This classification was also supported by Buck-Gramcko and many other authors. Yamauchi and Tanabu further classified it based on radiographic pattern of bone reduction (teratologic sequence) into: triphalangia type, diphalangia type, monophalangia type, aphalangia type, ametacarpia type, acarpia type and forearm amputation type.^[3] Recently, the Japanese Society for Surgery of the Hand (JSSH) proposed a modification of the IFSSH classification based on newer knowledge on teratology. The symbrachydactyly in all stages were transferred to group I. Two new groups were introduced. A group "failure of finger ray induction" including typical cleft hand (IC), central polydactyly (III) and (bony) syndactyly (II) along with a group of "unclassifiable" cases was added.^[13]

The exact etiology of this condition is still unknown. No extrinsic or environmental factors have also been identified as the causative factors. The presentation is usually sporadic but occasionally familial presentation is seen. In pathological consideration, the transverse deficiency is believed to occur following some kind of insult to the mesenchyme in the early embryonic period (around Carnegie stage 17-20) with resultant cell necrosis in the undifferentiated mesenchyme of the hand plate.^[3] If the damage is mild and formation has continued the intercalated transverse deficiency may occur. The vascular disruption in the early gestation is considered an important cause for this.^[14] Webbing in the symbrachydactyly results from failure of the apical ectodermal ridge under the influence of damaged mesenchyme.^[15]

The associated anomalies in symbrachydactyly apart from syndactyly and pectoral muscle related defects (Poland's) includes similar bone reductions in the foot, club foot, deformity of auricle, cleft lip and palate.^{[16],[17]} Few cases of associated ipsilateral renal hypoplasia / aplasia and mobius syndrome had also been reported.^{[6],[18]}

Surgical correction of sybrachydactyly is a demanding challenge but it offers a chance to improve the hand function and its aesthesis. Almost all type of sybrachydactyly causes some malfunction of the hand which can be remedied successfully by surgery. The most effective time considered for surgery in group I-II is the preschool age. Surgeries for other groups should be done in 2 - 3 years age group in order to attain natural grasp and pincer action as early as possible.

[19]

For group I (short finger type), as function is quite normal, the syndactyly release is usually the only procedure needed. The procedure is similar to those for conventional cutaneous syndactyly release. When multiple digits are involved it is advisable to do syndactyly release in stages so as to avoid interference in circulation of the central digits. If all the four web spaces are to be separated, the first and third space should be separated first followed by second and fourth after 6 months. For group II-IV, as the function of hand is jeopardized, the main consideration is to improve grasp and pinch. For Group II (cleft hand type), presence of a thumb and at least one ulnar finger allows pinch function. The surgical treatment, when needed, consists in separation of webbed fingers, resection of nonfunctional digital stumps, or finger translocation. For group III (monodactyly type) as all long fingers are absent, the pinch function can be created by a micro-vascular second toe transfer in ulnar location. This can be performed with or without the meta-tarso-phalangeal joint to achieve the ultimate functional goal of a "tip-to-tip" two fingers pinch. The bone lengthening by intercalary /only bone grafts and distraction is an alternative procedure to create a pincer grip. The iliac bone is the preferred site for the bone graft. At the most severe end of the spectrum i.e. group IV (peromelia type), surgery often may not be possible or worthwhile, however complex surgical reconstructions like nonvascularized multiple toe phalangeal transfers, web space deepening, distraction lengthening and microsurgical multiple toe transfers have been attempted with varying success rate.

CONCLUSION

Sybrachydactyly of the hand is a relatively rare congenital hand anomaly. The malformed hand should be properly assessed for the complexity of the problem and should be carefully evaluated for hand functions. As the deformity varies considerably in severity treatment needs to be individualized for the condition. In cases where hand functions are expected to be reasonably improved by surgery an appropriate surgical option should be considered keeping

in mind that reconstruction must do no harm.

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