



AN EPIDEMIOLOGICAL STUDY OF CONGENITAL DEFORMITIES OF HAND AT A TERTIARY CARE HOSPITAL

Orthopaedics

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ABSTRACT

Background: Congenital deformities of hand encompass a myriad of deformities, all of which carry different functional & cosmetic implications. The most commonly encountered malformations of the hand are syndactyly, polydactyly, congenital absence, camptodactyly, clinodactyly & radial club hand.

Aim: Our aim was to obtain a cross section of the epidemiology of congenital hand deformities and to be well adapted to help the parents & the child, whenever possible, regarding rehabilitation & surgical options for optimal functional outcome.

Settings & Design: This was a cross-sectional study of 50 patient attending at a tertiary care hospital for the duration of one and half year.

Methods & Materials: All the patients were examined clinically with detailed history including family history of similar anomalies. Clinical photographs were taken with due permission. Radiographs were obtained to confirm diagnoses. Functional assessment with the disabilities of arm, shoulder & hand (DASH) outcome questionnaire was done whenever possible.

Statistical Analysis Used: This was an observation study. The results obtained were analyzed using charts & diagrams.

Results: Total 50 cases recorded, 34 male & 16 female. Polydactyly (8) & symbrachydactyly (8) most commonly observed anomalies. Syndactyly (6) second most common anomaly. Anomalies were more commonly found on right side (23). Bilateral in 13 cases. Symbrachydactyly was more common in left in male patients. One male patient had bilateral trigger fingers & trigger thumb. Most of the patients were below 15 years (45).

Conclusion: We may conclude that although rare, congenital deformities of hand include a wide variety of disorders. Actual incidence may be higher than observed. 10% patients have significant cosmetic or functional deficit. Most common disorder found in our institute is polydactyly & symbrachydactyly. It is more common in boys.

KEYWORDS

congenital hand deformity, syndactyly, polydactyly, camptodactyly, clinodactyly & radial club hand.

INTRODUCTION

Congenital anomalies affect 1 to 3% of newborns and approximately 10% of these children have upper extremity abnormalities.^{1,2} Congenital limb anomalies are second only to congenital heart disease in the incidence of birth malformations.³ Most anomalies occur spontaneously or are inherited.⁴ Few malformations are attributed to teratogens^{5,6,7}.

Congenital malformations of hand encompass a myriad of deformities, all of which carry different functional & cosmetic implications for the patients & parents. The most commonly encountered anomalies of the hand are syndactyly, polydactyly, congenital absence, camptodactyly, clinodactyly & radial club hand. Isolated nongenetic defects include unilateral transverse failure of formation, defects due to constriction bands, longitudinal radial & ulnar dysplasias, macrodactyly & preaxial polydactyly. Isolated autosomal dominant deformities include lobster claw deformity, symphalangism, brachydactyly, triphalangeal thumb, camptodactyly & postaxial polydactyly. Syndactyly may occur sporadically or as a dominant trait^{8,9}. Approximately 10% of the patients with congenital hand deformities have significant cosmetic & functional deficits. Up to two thirds of the patients of congenital hand deformities have additional birth defects^{10,11,12}. The American Society for Surgery of the Hand & the International Federation of Societies for Surgery of the Hand accept the classification system devised by Swanson, Barsky & Entin which divides congenital deformities of hand into seven categories viz.

1. Failure of formation
 - Transverse arrest
 - Longitudinal arrest
2. Failure of differentiation (separation) of parts
 - Soft tissue involvement
 - Skeletal involvement
3. Duplication
4. Overgrowth
5. Undergrowth
6. Constriction ring syndrome
7. Generalized abnormalities & syndromes

This classification does not delineate etiological factors, treatment, prognosis, & there is ongoing work for further revision of the system²⁰.

MATERIALS AND METHOD

An institution based, prospective, non-interventional, observation study was conducted of 50 patients attending the Orthopaedics OPD, Paediatrics OPD, Disability boards at a tertiary care hospital over a period of one and half years from January 2017 to September 2018.

Inclusion Criteria: Patients with congenital deformities of hand.

Exclusion Criteria: Patients with acquired hand deformities.

Patients with neurological abnormalities.

Patients with congenital hand anomalies were added to the study after obtaining written informed consent. Data was collected in a simple format with two parts viz. patient particulars & disease details.

After getting approval from the Institutional Ethical Committee patients fulfilling the inclusion criteria were added to the study group on getting written informed consent. All the patients were examined clinically with detailed history including family history of similar anomalies. Clinical photographs were taken with due permission. Radiographs were obtained to confirm diagnoses particularly in radial & ulnar ray defects. Any suspected syndromic patient was referred to paediatric OPD for opinion & further work up as necessary. Functional assessment with DASH¹³ scoring system was done when possible.

The data collected was analyzed to draw an inference regarding cross section of the epidemiology of congenital deformities of hand at our institute during the study period.

RESULTS & ANALYSIS

Total 50 cases recorded, 34 male & 16 females. Polydactyly (eight) & symbrachydactyly (eight) most commonly observed anomalies. Syndactyly (six) second most common anomaly. Anomalies more commonly found on right side (23). Bilateral in 13 cases. Symbrachydactyly was more common in left in male patients. One

male patient had bilateral trigger fingers & trigger thumb. Most of the patients were below 15 years (45). DASH scoring was done in 8 cases. Polydactyly is the most common deformity observed along with symbrachydactyly. Equally distributed in male & females. No specific side preponderance. Included deformities like bifid thumb & mirror hand. There was family history of polydactyly in three cases. DASH functional scoring was done in two cases & results were 11.7 & 10.8.

Total eight cases of Symbrachydactyly were observed. Among them seven are male, one female. Left side predominance observed in male patients. DASH scoring done in one case (39.2). In one patient there was brachydactyly in opposite hand.

Six cases of Syndactyly were observed, three male & three females. Bilateral involvement seen in two cases. In a female patient there was syndactyly of toes, hypercalcaemia & history of seizure disorder (syndromic). Family history of syndactyly in a bilateral case.

Five cases of Clinodactyly were observed. Four male & one female. Right side predominance seen. DASH scoring done in one case (9.2).

Four cases of Camptodactyly were observed, Three male & one female. Bilateral involvement seen in two cases. In one baby with bilateral involvement, bilateral congenital talipes equinovarus (CTEV) was seen. There was also family history of CTEV. DASH functional scoring was done in one case (19.2).

Two cases of Trigger Finger were observed. No female patients in our study. Bilateral involvement seen in one case. In one patient there was bilateral trigger fingers & thumb. DASH score was (71.7).

Three cases of Trigger Thumb were observed. Two male & one female. Bilateral involvement seen in one male with bilateral trigger fingers. DASH score (71.7).

Two cases of Radial Club Hand were observed. One male & one female. Bilateral involvement in male baby. The baby had congenital heart disease (syndromic). In the female baby there was bilateral CTEV.

Total Four cases of Cleft Hand were observed. Three male, one female. Bilateral involvement seen in one case along with bilateral cleft feet. DASH scoring was done in two cases (65.8 & 67.5).

Four cases of Ulnar Dysplasia were observed, Three male & one female. Bilateral involvement in one male baby. In all cases there was elbow involvement (fused or unstable) on the same side with absent digits & thumb. In a bilateral involvement there was right tibial hemimelia also.

Arthrogryposis Multiplex Congenita Four cases observed. Two male & two female. Bilateral involvement in all four cases. In all cases there was involvement of two or more joints in the body particularly elbow, shoulder & hip.

DISCUSSION

Congenital anomaly of hand includes a wide spectrum of disorders. It is relatively uncommon but the actual incidence may be higher in general population as minor deformities do not seek treatment. Some of the patients included in this study sought treatment for other conditions & were incidentally discovered to having hand anomalies from birth. Population based studies on congenital hand deformities in literature found higher incidence than that estimated by studies on patients seeking treatment.^{1,16,8}

Congenital anomaly of hand is more common in boys. In our study anomalies were found mostly on the right side except in symbrachydactyly where left side predominance was seen in male patients. In literature no evidence of side preponderance is found.^{8,14,15,}

^{18, 19} Bilateral anomalies are found in about 25% cases. Non hand anomalies are found in about 22% cases, mostly in the lower limbs (18%). In 10% cases there is known occurrence in relatives. This data corroborates with the population studies in Stockholm¹⁵, Edinburgh¹⁴ & Western Australia⁸ Syndromic associations included Holt Oram syndrome (radial club hand with congenital heart disease), Barakat syndrome (hypoparathyroidism, sensorineural deafness, renal disease) with syndactyly of fingers & toes.

The most commonly observed congenital anomaly in our study was symbrachydactyly & polydactyly. Occurrence was sporadic or as a dominant trait. Polydactyly included deformities like bifid thumb & a very rare anomaly, mirror hand. Other common anomalies included syndactyly, clinodactyly, camptodactyly, ulnar dysplasia, arthrogryposis multiplex congenita, trigger fingers & thumb & radial club hand. In most of the population or institute based studies in literature, syndactyly or polydactyly is the most common congenital anomaly of hand.^{8,14,15,18,19} A population based study, "Congenital upper limb deficiencies and associated malformations", conducted in Finland¹² by Koskimies E, Lindfors N, Gissler M, et al had similar observations. Comparable results are found in other studies also.^{8,14,15,18,19}

CONCLUSION

The intention of this study was to evaluate the cross section of epidemiology of congenital deformities of hand at our institute for one & a half years.

Although rare, congenital anomalies of hand include a wide variety of disorders. Significant cosmetic & functional disability is seen only in 10% cases. The actual incidence may be higher in population as patients with minor deformities do not seek treatment. Most of the patients included in our study either visited for disability certificate or were incidentally discovered while seeking treatment for other condition.

It is more common in boys. Approximately one third of the patients have bilateral involvement. About 15% patients have associated non hand anomalies, most commonly involving the lower limb. In 11% cases there is positive family history.

Polydactyly, symbrachydactyly, syndactyly, clinodactyly, camptodactyly, trigger fingers & thumb, radial club hand, ulnar dysplasia & arthrogryposis multiplex congenita was observed

We may conclude that although rare, congenital deformities of hand include a wide variety of disorders.

- Actual incidence may be higher than observed.
- 10% patients have significant cosmetic or functional deficit.
- Most common disorder found in our institute is polydactyly & symbrachydactyly. It is more common in boys.
- There is slight right side preponderance. Bilateral involvement is seen in about one fifth cases.
- Associated anomalies seen in 15% cases & mostly involve the lower limbs.
- Positive family history is seen in 11% cases.

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