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Frequency and Variation of Congenital Anomalies of the Upper Limb at Liaquat University Hospital Jamshoro

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Abstract

Congenital anomalies (also referred as birth defects) can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth. 1-2 % of newborns have congenital anomalies. Out of them 10% are anomalies of upper limb especially hand and causing disfigurement and disability in the newborn. This descriptive study was conducted in department of Plastic and Reconstructive Surgery, Liaquat University Hospital Jamshoro, Sindh Pakistan to determine the frequency and variations of congenital upper limb anomalies (CULA) seen in our institution. 30 patients with (CULA) were included in the study. All were admitted through the outpatient department. The type of anomaly was recorded with other biodata. Surgery was performed. The most common anomalies were syndactyly, polydactyly and cleft hand. Such type of study in our setup is conducted for the first time which shows that different varieties of (CULA) are seen commonly in our setup and successful surgical repair is also possible. Further research is also required in order to diagnose them in uterus, intra uterine surgical intervention and possible causative factors needs elaboration.

Keywords: congenital; upper limb; anomalies; syndactyly.

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1. Introduction

A congenital anomaly is an abnormality of structure, function or metabolism which is present at time of birth and results in long-term disability. The exact cause of birth defects is not known it may be genetic, infectious or environmental in origin. The congenital upper limb anomalies (CULA) constitute approximately 10% of the total 1-2% of congenital anomalies of newborns but actual number may vary widely in different regions. Although different studies have been undertaken in different parts of world but no such study has been undertaken in this hospital to best of our knowledge. LUH is the biggest public sector tertiary care unit serving health facilities to many districts of lower Sindh. Thus this study was designed to document the pattern of congenital anomalies of upper limb in patients in this region [1, 2].

The upper limb begins to develop in 4^{th} week after fertilization and completed in 8^{th} week with formation of all upper limb structures. Embryogenesis begins with the formation of upper limb bud an out pocketing from ventrolateral body wall opposite to lower five cervical and upper two thoracic segments. The majority of congenital anomalies of the upper extremity occur during this period of rapid limb development. During the period of development of upper limb other organ systems are also developing so terotogenicity may effect the other organ systems as well [3].

So patient with congenital anomalies of upper limb must be carefully examined as some of the (CULA) occurs in isolation while others may be associated with musculoskeletal or systemic disorders. The upper limb anomaly may be part of a wider anomaly, like VATER association (vertebral, anal, tracheo-oesohageal, radial club hand), or more expanded to VACTERL (vertebral abnormalities, anal atresia, cardiac abnormalities, tracheoesophageal

fistula, esophageal atresia, renal defects, radial dysplasia, and lower-limb abnormalities) in such cases proper examination of patient is required to rule out associated cardiac and renal anomalies. Radial club hand is associated with cardiac septal defect and with rare hematological conditions so presence of hand anomaly in infants may indicate significant anomalies in cardiovascular, neurological and hematopoietic systems [4]. The benefit of early surgery provide maximum potential for growth and development, improved scarring, early incorporation of the reconstructed part; and reduced psychologic affect.The best time for surgery is as earlier as possible within first 2 years of life with integration of child's other medical and surgical problems. As early surgery is associated with increased technical difficulties in microsurgeries and anesthetic risks so may be troublesome in some cases [5].

2. Methodology

This descriptive study was conducted in department of plastic and reconstructive surgery at Liaquat University hospital Jamshoro from July 2013 to July 2014. A total 30 patients were studied in this study. All are admitted throughout patient department. A detailed history for any risk factor along with biodata was taken. Then thorough physical examination was performed. Confirmation of internal defect was done by various investigations like radiography, ultrasound, echocardiography and CT scan and pattern wise distribution of the anomalies was recorded according to classification given by Swanson in 1983 [6].

3. Results

During study period total 30 patients were examined, 12 males and 18 females of different age group between 04 months to 19 years, type of anomaly was noted with other details as shown in table 1 frequencies of each anomaly is shown in table 2. No any associated systemic anomaly was found in these patients except constriction bands in 04 cases. Some of the figures of typical cases are shown subsequently.

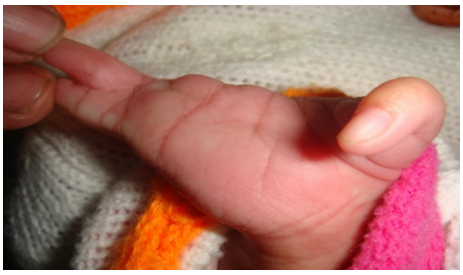

Figure 1 Cleft hand

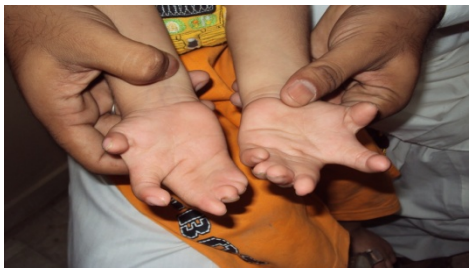

Fig.2 Complex syndactyly of right and polydactyly and syndactyly of left

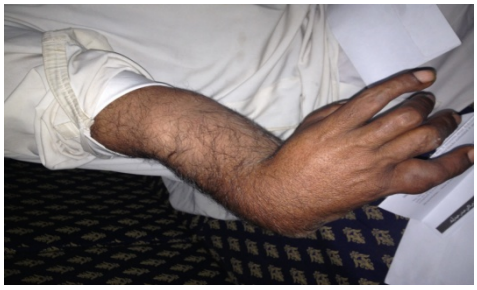

Fig.3 Radial club hand

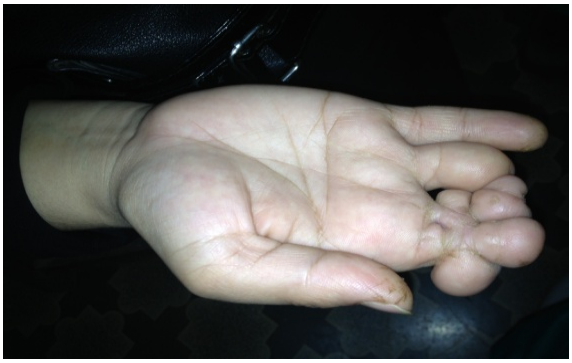

Fig. 4 Complex polydactyly and symbrachydactyly

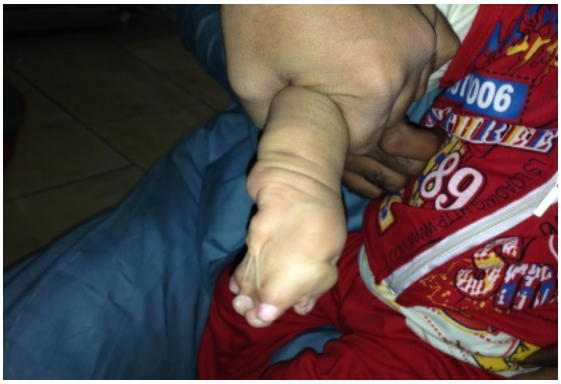

Fig.5 Apert hand

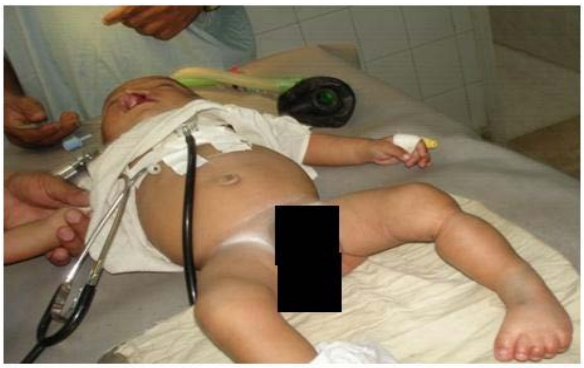

Fig.6 Constriction bands involving left leg with club foot and cleft lip deformity

Table 1 showing age, gender and anomaly

S.No	Age of patient	Gender	Type of anomaly
1	16 years	Female	Polydactyly plus Camptodactyly
2	3 years	Female	Cleft hand(fig.1)
3	04 years	Female	showing complex syndactyly of right and polydactyly and syndactyly of left (fig.2)
4	14 years	Female	cleft hand with narrow first web space
5	1year	Female	Complex syndactyly with polydactyly
6	13years	Male	Camptodactyly of ring finger
7	4months	male	Type 3 Apert hand
8	4months	Male	Complex syndactyly
9	15years	Male	Radial club hand (fig.3)
10	8years	Male	Acrosyndactyly
11	19 years	Female	Complex polydactyly and Symbrachydactyly.(fig.4)
12	08 years	Female	Simple and complete syndactyly
13	1 year	Female	Cleft hand
14	04 years	Female	Polydactyly and Symbrachydactyly.
15	9 years	Female	Acrosyndactyly
16	04 year	Female	Apert type 2 mitten or spoon hand
17	14 year	Female	Cleft right hand with index clinodactyly, left hand incomplete syndactyly of third web space.
18	09 year	Male	Thumb polydactyly
19	3 year	Male	Cleft hand
20	5 months	Female	Complex hands and feet
21	8 years	Female	Symbrachydactyly of left hand
22	02 years	Male	Apert type 2 hand (fig.5)
23	03 years	female	Constriction band syndrome with radial nerve palsy
24	02 years	Male	Cleft hands
25	03 years	Male	CBS with aplastic fingers
26	5years	female	Constriction band syndrome with distal lymph edema
27	15years	female	Simple and complete syndactyly of 3rd and fourthweb space.
28	6 months	Male	Constriction band syndrome, cleft lip(fig.6)
29	10 years	Female	Polydactaly
30	15years	Male	Syndactly

Table 2 showing frequency of anomalies

Type of Anomaly	NO.of patients affected	Frequency
Failure of formation of parts		
Radial club hand	01	2.8%
Cleft hand	05	14.2%
Failure of differentiation		
Syndactyly	10	28%
Apert syndrome	03	8.57%
Camptodactyly	02	5.71%
Clinodactyly	01	2.8%
Symbrachydactyly.	03	8.57%
Duplication		
Polydactyly	06	17.14%
Congenital constriction band syndrome	04	11.4%

4. Discussion

Limb malformation occur in approximately 6/10,000 live birth with 3.4/10,000 affecting the upper limb. Congenital anomalies of the hand and upper extremity constitute one of the most frequent disorders that afflict newborns. The cumulative incidence is often underappreciated because of the large variety of anomalies, methodology issues, variability in reporting, and also the racial variations of the individual disorders. The most commonly occurring congenital anomalies of the upper limb are polydactyly, syndactyly, and camptodactyly. The incidence of syndactyly is estimated to be approximately 1 in 2,000 births. Polydactyly shows considerable racial variation and is autosomal dominant in many African and Americans. Familial polydactylies are often bilateral and symmetrical in presentations. However, if one assumes a relatively similar incidence of syndactyly, camptodactyly, and polydactyly across the broader population, then the combined incidence of these three congenital anomalies of the upper limb would be approximately 1 in 750 births. The more complex congenital hand anomalies are less frequent. As a point of reference, Apert syndrome, one of the less frequent

anomalies, has an approximate incidence of approximately 1 in 100,000 births, there would only be expected to be 40 children born per year with Apert syndrome in the entire United States [7].

Detailed classification criteria have been developed for congenital hand anomalies. The most recognized comprehensive classification developed by the International Federation of Societies for Surgery of the Hand (IFSSH) classifies congenital hand anomalies in seven categories: (1) failure of formation; (2) failure of differentiation; (3) polydactyly; (4) overgrowth; (5) undergrowth; (6) amniotic band syndrome; and (7) generalized skeletal syndromes.

The purpose of this study is to get an idea about the frequency and variation of different congenital hand anomalies in our set up. According to this study most commonly seen anomaly is syndactyly seen in 10 cases out of 35(28%), next commonest is polydactly seen in 06 cases out of 35 (17%) Trent and Stephen also find these anomalies quite frequently in their study on limb defects in large fetal collection [8].

Munisamy and Uppalu also found polydactly and syndactly as commonest anomalies with cleft hand numbered next in this study we found cleft hand in 5 cases (14%) [9].

Abu saleh find cleft lip in association with constriction band syndrome, Obdeijn in 2010 also report facial Clefts in association with limb anomalies we also find one case of cleft lip with constriction band and club foot in six months old baby [10,11].

The surgeon treating children with upper limb anomalies must offer surgery to improve the child's function and cosmesis, at appropriate time after counseling parents about possible outcomes. Sucessful surgical repair is also possible in our setup depending highly on the timing of surgery. Good awareness to general population is play vital role in bringing their siblings for proper management at proper time and at proper place.

5. Conclusion

Congenital hand surgery is one of the most challenging aspects of surgery and the hand surgeon is routinely confronted with significantly aberrant anatomy, The '"impact factor" of successful management yields a lifetime of rewards for the afflicted child.

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