The Epidemiology of Congenital Upper Limb Anomalies (CULA) In Population of D. I. Khan Division, Pakistan

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Each author of this article fulfilled ALL 04 Criteria of Authorship:
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ABSTRACT

Objective: To find out the Prevalence of Congenital Upper Limb Anomalies by its distribution according to Type, Gender, Family history & Cousin marriages in one calendar year.
Study Design: cross sectional study.
Place and Duration of study: Department of Orthopaedics Medical Teaching Institute Dera Ismail Khan, Pakistan from January 1, 2022 to December 31, 2022.
Materials & Methods: Infants (from birth to one year age) of both genders having congenital anomalies of upper extremities were included in this study. Infants with cerebral palsy, birth palsy and birth trauma were excluded. Swanson's classification was used for classification and types of congenital upper limb anomalies. All cases were analyzed for the type of CULA, gender, family history and parents as first-degree relatives i.e., cousin marriages. Categorical variables were measured as count and percentages while numeric variable was measured as mean and standard deviation, with 5% margin of error and 95% confidence interval, p-value < 0.05 was set as significant.
Results: The overall prevalence of congenital upper extremity anomalies was 142(0.26%), which is similar to International ratios. Polydactyly was most common (51 cases) followed by syndactyly (39 cases) and radial club hands (17 cases). The ratio among boys and girls was 52% and 48% respectively. Forty (28%) patients had a positive family history for CULA as compared to 102 (72%) patients, with p-value <0.05 which was significant. One hundred and twelve (79%) patients had their parents as first degree relative i.e., cousin marriages as compared to 30 (21%) patients with no history of cousin marriages, with p-value <0.05 which was significant.
Conclusions: In our study it was found that Polydactyly is the commonest congenital upper limb anomaly. There was increased incidence of CULA in Cousin marriages & patients with positive family history for CULA. Increased incidence of CULA was found in boys as compared to girls.
Key words: Congenital upper limb anomalies; epidemiology; OMT classification; Polydactyly; Orthopedics; Syndactyly; Hand.

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INTRODUCTION

Congenital anomalies are the structural or functional abnormalities that are present at the time of birth.[1] Congenital abnormalities affect between 3-4% of live births & is the fourth leading cause for infant mortality.[2,3] Among these 10% patients have congenital upper limb deformities.[4] The incidence of congenital upper extremity anomalies is 2 in 1000 live births world-wide.[5] In USA congenital anomalies is the leading cause of infant's mortality with approximately 20.3% of all deaths during infancy.[6] Some of these anomalies occur in isolation but there are associations with systemic syndromes that may involve cardiopathies, CNS malformation or delayed neuropsychomotor development e.g., Fanconi Anemia, TAR (Thrombocytopenia Absent Radius) etc.[7,8]

Approximately 66% of congenital anomalies have unknown etiology. The risk factors for congenital anomalies are the various genetic & environmental factors such as exposure to radioactive materials or teratogens, diabetes, consanguineous marriages, mother addicted to alcohol & malnutrition.[9,10] Anomalies are divided into two categories based on the severity i.e., major & minor. Major anomalies affect the individual's life & performance while minor anomalies don't require treatment or these can be improved through simple methods.[11] Swanson & Oberg-Manske-Tonkin (OMT) classification are commonly used for the classification of congenital upper limb anomalies.[12,13] Swanson's system of classification is accepted by IFSSH & ASSH.[14,15]

Goldfarb et al.[16] from New York, USA in a study which included children born between 1992 to 2010, found that overall prevalence of CULA in New York State & City was 0.27% i.e., 27.2 cases per 10,000 live births. Shin et al.[17] from the Republic of Korea in a Prospective Cohort study for the period from 2007 to 2016 found that the mean annual incidence of CULA was 0.23% i.e., 23.5 per 10,000 live births. The incidence was significantly higher i.e., 0.26% in boys as compared to 0.20% in girls. Polydactyly was the most common anomaly in population. Ogino et al.[18] from Japan in a sixteen years duration study found that the distribution of CULA by sex was 51% (491) in boys as compared to 49% (552) in girls. 3.6% (34) patients had a positive family history for CULA. Trigger finger was the most common anomaly, followed by polydactyly & camptodactyly. Al-Gazali et al.[19] from United Arab Emirates in a 2 years study found that among 5 patients having Congenital upper limb anomalies (i.e., 3 having Polydactyly & 2 patients with Clinodactyly), three patients had consanguineous marriages & 2 patients had non-consanguineous marriage.

We searched local and national literature regarding our topic, but couldn't found, so this will be the first study from Pakistan to show the epidemiology of congenital anomalies of upper limb and hence will provide data to our National health officials to allocate funds to treat these anomalies accordingly and efficiently. The objective of this study was to find Prevalence of CULA and the Distribution of CULA by Sex, Family history & Cousin marriages in one calendar year.

METHODOLOGY

This cross-sectional study was conducted in the department of Orthopedics, DHQ Teaching Hospital, D.I.Khan, Pakistan from January 01, 2022 to December 31, 2022. Approval for the research was taken from hospital ethical committee (No.256/GJMS) & informed consent was taken from patients or attendants. There were expected 54,600 live births in D.I.Khan division during the study period, which was calculated from the birth rate of Pakistan i.e., 2.7034 births per 100 people in 2021. Patients having age less than 1 year & presented to OPD in DHQ Teaching Hospital & Mufti Mehmood Memorial Teaching Hospital Dera Ismail Khan with CULA were included in the study. Infants with cerebral palsy, birth palsy and birth trauma were excluded. Swanson's classification was used for categorization of anomalies. Patients were divided on the basis of family history for the presence of CULA & also on the basis of Parents as a first-degree relative i.e., cousin marriages.

All CULA patients having age less than 1 year were assessed for the type of anomaly on the basis of Swanson's classification. Multiple categories of anomalies in one patient were considered as a single case. Data was collected in our out-patient department by specifically designed proforma under the supervision of consultant orthopaedic surgeon. All information's regarding type of anomaly, family history of each patient for the presence of CULA & also for the parents of patients as a first-degree relative i.e., cousin marriages were recorded. Gender (male/female), family history of CULA & Parents as first degree relative i.e., cousin marriages were our independent variables while the presence of CULA was our research variable. The data for the sample was described by counts & percentages and for the population as confidence interval at 95% confidence level. Chi-square goodness of fit test was used to test for the hypotheses & to compare categorical variables.

RESULTS

The overall Prevalence of congenital upper extremity anomalies in population of D.I.Khan division, Pakistan was 142(0.26%) (Table 3.1). Polydactyly was most common (51 cases) followed by Syndactyly (39 cases) and Radial club hands (17 cases). Arthrogryposis (12), Macrodactyly (7), Trigger fingers (5), Camptodactyly (5), Radio-Ulnar synostosis (4) and Cleft hand (2).

The distribution of CULA by Gender was 74 (52%) males as compared to 68 (48%) females, (Table 3.2) and the results were statistically significant (p-value<0.05).

The distribution of CULA by family history was that 40 (28%) patients had positive family history for CULA as compared to 102 (72%) patients with no family history of CULA, (Table 3.2) and the results were statistically significant (p-value<0.05).

The distribution of CULA by cousin marriages was that 112 (79%) patients had their parents as first degree relative i.e., cousin marriages as compared to 30 (21%) patients with no history of cousin marriages,(Table 3.4) and the results were statistically significant (p-value<0.05).

Table 3.1: The prevalence of CULA in population of D.I.Khan division, Pakistan.

Variables		Observed	Adjusted Expected	Difference	Difference Square	Chi-square value	d.f	p-value
Presence of CULA	Yes	26	23.5	2.50	6.25	0.267	1	>0.05
	No	9974	9976.5	-2.50	6.25			
Total		10,000	10,000					

Table 3.2: The distribution of CULA by gender in population of D.I.Khan division, Pakistan.

Variables		Observed	Adjusted Expected	Difference	Difference Square	Chi-square value	d.f	p-value
Presence of CULA	Yes	74	86.43	-12.43	154.50	4.568	1	<0.05
	No	68	55.57	12.43	154.50			
Total		142	142					

Table 3.3: The distribution of CULA by family history in population of D.I.Khan division, Pakistan.

Variables		Observed	Adjusted Expected	Difference	Difference Square	Chi-square value	d.f	p-value
Presence of CULA	Yes	40	5.1	-34.90	1218.01	247.723	1	<0.05
	No	102	136.9	34.90	1218.01			
Total		142	142					

Table 3.4: The distribution of CULA by parents as first-degree relatives i.e., cousin marriages in population of D.I.Khan division, Pakistan.

Variables		Observed	Adjusted Expected	Difference	Difference Square	Chi-square value	d.f	p-value
Presence of CULA	Yes	112	85.2	-26.80	718.24	21.075	1	<0.05
	No	30	56.8	26.80	718.24			
Total		142	142					

DISCUSSION

Congenital anomalies are the main cause of disability & mortality among children in developing as well as developed countries. The various surgical procedures required in these children & hospitalization for proper management impose a great load on health system & families.[20,21] Identifying & preventing the incidence of congenital anomalies is more cost effective as compared to treatment or rehabilitation of these patients.[22] The overall Prevalence of congenital upper extremity anomalies in population of D.I.Khan division, Pakistan was 142(0.26%).

Similar to our study Senes et al.[23] from Italy found that the prevalence of Congenital Hand and Upper Limb anomalies was 0.25% with the predominance of right sided anomalies. Goldfarb et al.[16] from New York, USA in a study found that overall prevalence of CULA in New York State & City was 0.27%. Shin et al.[17] from the Republic of Korea found that the mean annual incidence of CULA was 0.23%. Parikh et al.[24] from India found that the prevalence of Congenital Limb Deformity (CLD) in neonates was 0.63% i.e., 63.4 per 10,000 live births.

Polydactyly was most common (51 cases) followed by Syndactyly (39 cases) and Radial club hands (17 cases). Shin et al.[17] from the Republic of Korea in a study also found that polydactyly was the most common anomaly among CULA. Ogino et al.[18] from Japan found that Trigger finger was the most common anomaly, followed by polydactyly & Camptodactyly. Uzun et al.[25] from Turkey in 2020 found that syndactyly was the most common anomaly followed by ulnar club hand.

The distribution of CULA by gender was that 74 (52%) male patients had CULA as compared to 68

(48%) females. Similar to our study Barik et al.[26] from India found that distribution of CULA by gender was 58.2% in boys as compared to 41.8% in girls. Shin et al.[17] from the Republic of Korea found that incidence was significantly higher i.e., 0.26% in boys as compared to 0.20% in girls. Ogino et al.[18] from Japan found that the distribution of CULA by sex was 51% (491) in boys as compared to 49% (552) in girls.

The distribution of CULA by family history was that 40 (28%) patients had positive family history for CULA as compared to 102 (72%) patients with no family history of CULA. Similar to our study Ogino et al.[17] from Japan found that the distribution of CULA by family history was 3.6% (34) patients had a positive family history for CULA. In literature no study was retrieved with a different result.

The distribution of CULA by cousin marriages was that 112 (79%) patients had their parents as first degree relative i.e., cousin marriages as compared to 30 (21%) patients with no history of cousin marriages. Similar to our study Al-Gazali et al.[18] from United Arab Emirates found that among 5 patients having Congenital upper limb anomalies (i.e., 3 having Polydactyly & 2 patients with Clinodactyly), three patients had consanguineous marriages & 2 patients had non- consanguineous marriage.

CONCLUSION

In our study it was found that Polydactyly is the commonest congenital upper limb anomaly. There was increased incidence of CULA in Cousin marriages & patients with positive family history for CULA. Increased incidence of CULA was found in boys as compared to girls.

Ethical Approval:
This study was performed with the approval of the Ethical Committee of Gomal Medical College DIKhan (No.256/GJMS Dated: November 13, 2021).

Patients' Consent:
Informed consent was obtained from all patients and their relatives before the study began.

Competing Interest:
The authors declared no potential competing interest with respect to the research, authorship, and publication of this study.

Authors' Contribution:
All the three authors contributed equally in data collection, compilation, analysis and manuscript writing. All authors have read and approved the final version of the manuscript to be published.

Conflict of Interest: None

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