

Trends of Congenital Anomalies in Paediatric Orthopaedic Surgery in Pakistan Emirates Military Hospital Rawalpindi

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ABSTRACT

Objective: To demonstrate the trends regarding the congenital disease burden in the paediatric orthopaedic surgery and explore the importance of enhancing paediatric orthopaedics in Pakistan.

Study Design: Retrospective, Cross-Sectional Study

Place and Duration of Study: Department of Orthopaedic Surgery, Pakistan Emirates Military Hospital Rawalpindi, Pakistan from Jan 2022 to Dec 2024.

Methodology: Paediatric patients with age range of 0-12 years, both genders suffering from a congenital disorder with complete medical record of diagnosis and patient information were included in the study. A total of 840 patients who had a complete medical record were included in the study. After the screening through administration, the patient medical profiles in Health Information Management System were accessed. After comprehensive evaluation and assessing the medical files regarding the patients visiting Orthopaedic Surgery Department, the relevant patients' data was obtained, and data extraction sheets were made. Frequencies and percentages were calculated regarding the type of congenital anomalies.

Results: In 2022, 204(24.3%) patients with different congenital anomalies reported to the orthopaedic surgery department. 275 (32.7%) of congenital anomalies were seen in 2023 and increased percentage of 361(42.9%) was demonstrated in 2024 with DDH being most common with 100 patients.

Conclusion: Increased number of patients with congenital disorders were seen in the Orthopaedic Surgery Department of PEMH over the last 3 years. Increased frequency of developmental dysplasia of hip being 26.7% and clubfoot being 26.4% were noted, highlighting the need to acknowledge paediatric orthopaedic surgery in Pakistan.

Keywords: Cerebral Palsy, Clubfoot, Congenital Anomalies, Orthopaedic Surgery, Pediatrics

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INTRODUCTION

Over the last decade the field of orthopaedic surgery has subspecialized in the area of paediatric orthopaedic surgery providing standardized care to the children around the world. The analysis of database of American Board of Orthopaedic Surgery showed that paediatric cases were more performed by the paediatric orthopaedic specialists as compared to the orthopaedic surgeons.¹ Paediatric orthopaedics include various conditions related to the children like fracture, trauma, spine, scoliosis, tumor, sports medicine, knee problems and congenital disorders including developmental dysplasia of the hip (DDH), Legg-Calve-Perthes, osteogenesis imperfecta and clubfoot.²⁻⁴ In Pakistan the horizon of paediatric orthopaedics is very limited. Epidemiology of the congenital anomalies in paediatric orthopaedics is an important public health concern.

There are numerous congenital conditions which are frequently encountered in paediatric orthopaedics. Metatarsus Adductus is the most common foot abnormality, but talipes equinovarus or clubfoot, calcaneovalgus deformity, and congenital vertical talus may also be encountered.⁵ Joint contractures that spontaneously improve are normal in the newborn, but it is important to identify and institute proper treatment for early developmental dysplasia of the hip, congenital knee dislocation, and torticollis. Clavicular pseudarthrosis and periosteal reactions may be discovered on radiographic examination.⁶ A study in UK documented that the incidence of DDH was 4.9 per 1000 live births and one in 200 infants require treatment of DDH.⁷ A local study showed 6000 to 7000 cases per year of clubfoot in Pakistan.⁸ Another study in Khyber Pakhtun Khwa also reported high frequency of clubfoot that is 36.6%.⁹

Paediatric orthopaedic care is a new branch in the healthcare landscape of Pakistan. In order to significantly improve the overall health outcomes for

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children, this field needs to be promoted and flourish. Monitoring the changes in the incidence and patterns of congenital anomalies proves essential in order to generate more research in the field.¹⁰ The information regarding the epidemiological studies of the congenital anomalies provide the basis for estimating the national healthcare costs and the number of specialists required. There are fewer local studies regarding the epidemiology of the congenital anomalies encountered in paediatric orthopaedic surgery. This study aimed to demonstrate the trends regarding the congenital disease burden among paediatric population in the department of Orthopaedic Surgery in Pak Emirates Military Hospital (PEMH) highlighting the importance of enhancing the branch of paediatric orthopaedics in Pakistan.

METHODOLOGY

A retrospective cross-sectional study was conducted in the department of Orthopaedic Surgery in Pak Emirates Military Hospital Rawalpindi in December 2024. The ethical approval was granted by the Ethical Committee of the Pak Emirates Military Hospital (Ref No. 13046/1-12-2024). The sample size of 1025 was calculated using the World Health Organization (WHO) calculator keeping confidence level at 95%, anticipated population proportion for neurological disorders among congenital anomalies in Pakistan at 40% and absolute precision at 4%.¹¹ Convenience sampling technique was used.

Inclusion Criteria: Paediatric patients with age range of 0-12 years, both genders suffering from a congenital disorder with complete medical record of diagnosis and patient information were included in the study.

Exclusion Criteria: Patients having incomplete medical record and vague diagnosis were excluded from the study.

A retrospective data of 1000 patients were obtained from January 2022 to December 2024. After applying the exclusion criteria 840 patients were included in the study. To access the patient record consent was taken from the competent authorities. Patient confidentiality was maintained. After the screening through administration, the patient medical profiles in Health Information Management System (HIMS) were accessed as all the detailed information about the patients can be retrieved through it. After comprehensive evaluation and assessing the medical files regarding the patients visiting Orthopaedic

Surgery Department, the relevant patients’ data was obtained, and data extraction sheets were made.

The data was entered in Statistical Package for Social Sciences (SPSS) version 26. Frequencies and percentages were calculated regarding the type of congenital anomalies and gender. Descriptives were calculated for age. Chi square test was applied to find any association of the congenital anomalies regarding gender and year. A *p*-value equal to or less than 0.05 was considered significant at 95% confidence interval. Results were presented as graphs and tables.

RESULTS

A total of 840 patients who had a complete medical record were included in the study. Mean age of the patients was 5.61±3.40 years. 398(47.4%) were females and 442(52.6%) were males. Clubfoot was the most common congenital anomaly seen in males with 122(27.6%) followed by DDH being highest found in females with 117(29.4%) as shown in Figure. In 2022, 204(24.3%) patients with different congenital anomalies reported to the orthopaedic surgery department. 275(32.7%) of congenital anomalies were seen in 2023 and increased percentage of 361(43.0%) was demonstrated in 2024. Increase in number of patients was seen over the years.

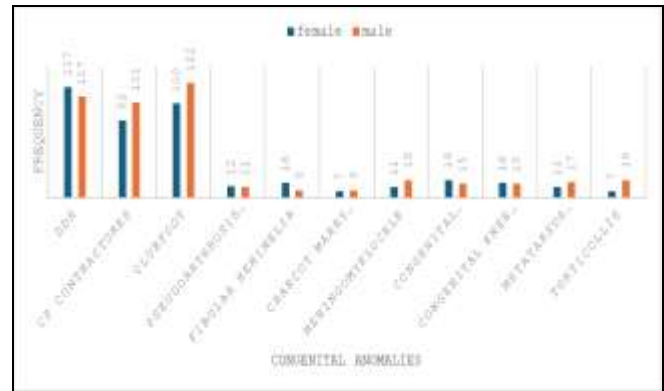


Figure: Frequency of Congenital Anomalies among Genders

Regarding frequency of different congenital anomalies over the years, DDH was the most common followed by clubfoot and Charcot Marry tooth disease was the least common as shown in Table-I.

No statistically significant association was found between the congenital anomalies regarding males or females over the years as presented in Table-II. However, the number of patients increased over the

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years with DDH being most common with 100 patients in 2024.

a recent metanalysis the prevalence of DDH was 1 in 100 with increased rate of prevalence seen over the last

Table-I: Frequency of Congenital Anomalies Over Three Years

Congenital Anomalies	2022n(%)	2023n(%)	2024n(%)	Totaln(%)
DDH	60(29.4)	64(23.3)	100(27.7)	224(26.7)
Clubfoot	45(22.0)	84(30.5)	93(25.8)	222(26.4)
CP contractures	50(24.5)	55(20.0)	78(21.6)	183(21.8)
Pseudoarthrosis tibia	5(2.5)	9(3.3)	9(2.5)	23(2.7)
Fibular hemimelia	5(2.5)	8(2.9)	11(3.0)	24(2.9)
Charcot Marry tooth disease	4(1.9)	5(1.8)	6(1.7)	15(1.8)
Meningomyelocele	8(3.9)	9(3.3)	13(3.6)	30(3.6)
Congenital vertical talus	8(3.9)	13(4.7)	13(3.6)	34(4.0)
Congenital knee dislocation	4(1.9)	11(4.0)	16(4.4)	31(3.7)
Metatarsus Adductus	8(3.9)	9(3.3)	11(3.0)	28(3.3)
Torticollis	7(3.4)	8(2.9)	11(3.0)	26(3.1)

*DDH: Developmental dysplasia of Hip; CP: Cerebral palsy

Table-II: Distribution of Congenital Anomalies by Genders over the Years

Years		Gender		p-value
		Males n (%)	Females n (%)	
Years	2022	103 (50.5)	101 (49.5)	0.758
	2023	145 (52.7)	130 (47.3)	
	2024	194 (53.7)	167 (46.3)	

*Chi Sq Test was applied

DISCUSSION

There is high representation of neurological defects followed by limb defects and musculoskeletal defects.¹² In Pakistan 58% of the congenital anomalies are seen because of the parental consanguinity. A local study highlights that among the limb defects, polydactyly and talipes were most prevalent while, among neurological disorders, intellectual disability and cerebral palsy were more frequent.¹³ This study highlights the prevalence of the second most common congenital disorders that is limb defects and musculoskeletal defects.

According to the results of the present study the number of patients increased from 204 in 2022 to 361 in 2024. This increase in patient number over the years in the orthopaedic surgery department in PEMH may be due to the increased awareness about the treatment being provided for the congenital anomalies related to orthopaedics. Successful treatment of congenital orthopaedic problems develops a better doctor-patient relationship. DDH is the most common presentation at PEMH with 26.7%. DDH is a congenital disorder of the hip joint which can lead to severe disability during the critical growth period if not treated correctly and timely. The incidence of DDH ranges from 0.5-30% in different populations around the world. According to

30 years.¹⁴ A local study reported high frequency of neglected cases of DDH as there is a lack of early screening and referral system.^{15,16}

After DDH other common disorders presenting are clubfoot (26.4%) and contractures in Cerebral Palsy children (21.8%). Clubfoot deformity consists of equinus and varus rotations along with the ductus and cavus deviations at the ankle occurring at birth. It is the seventh most prevalent congenital birth anomaly and the most common of the musculoskeletal system. Globally the burden of this birth defect affects more than 150,000 infants every year. In accordance with our results, a study in Khyber Pakhtun Khwa also reported high frequency of clubfoot that is 36.6%.⁸ Similarly, another study by Jarurantanasirikul et al, showed a high frequency of 44.1% of the clubfoot among the lower limb abnormalities.¹⁷ Cerebral Palsy is a congenital neurological disease resulting in pronounced functional disabilities. In developing countries prevalence of CP is 2.5 per 1000 live births. In Pakistan a local survey regarding neurological disorders in children, CP was the third most prevalent anomaly with 12% frequency.¹⁸ In contrast to the present study, a Danish study reveals 44% of knee contractures in CP children which is very high.¹⁹ In accordance with the present study results, a Swedish study reported a high frequency of 22% of contractures in CP.²⁰ 36% of contractures in CP children were demonstrated by Makki D et al. which is also close to our results.²¹ A local study showed 67% prevalence of contractures in CP children which is very high frequency as compared to our results.²² The general population should be made aware of the congenital disorders and the treatments that can be provided and where they can access them to improve

the quality of life of their children. About 69.7% people were unaware about the clubfoot deformity as depicted by a local study.²³

Around the globe due to lack of paediatric surgeons, the children requiring surgery get absorbed in the adult surgical services. Pakistan also faces similar challenges in delivering adequate surgical care to children. There is a high burden of paediatric patients in Pakistan and still the paediatric surgical care remains limited throughout Pakistan.²⁴ A recent study highlights the paediatric surgical crisis in Pakistan where the unmet surgical need was calculated to be 13.5% with the extremities being neglected for surgical treatment in 28% of cases of children.²⁵ There is dearth of paediatric orthopaedic surgeons in Pakistan. It is a need of time to flourish this particular speciality to improve the care provided to the special children so that their quality of life can be enhanced. Healthcare planning in Pakistan needs more research to formulate the required policies to improve the overall system. To decrease the paediatric orthopaedic surgery care gap new programs like fellowships or FCPS should be initiated to produce qualified surgeons in this respective field. International exchange programs in paediatric orthopaedic surgery should be promoted and doctors should be accommodated without any hassle. Moreover, the information regarding the epidemiological studies of the congenital anomalies provides the basis for estimating the national healthcare costs and the number of specialists required.

LIMITATION OF STUDY

This was a retrospective analysis of records only showing glimpse of diagnosis of data available. No associations or temporal causal effect relationship were possible because of cross-sectional design. Each congenital orthopaedic problem requires a separate study as there is so much to address in a single disorder including its further types and presentations and the treatment options being provided.

CONCLUSION

Increased number of patients with congenital disorders were seen in the Orthopaedic Surgery Department of PEMH over the last 3 years. Increased frequency of developmental dysplasia of hip being 26.7% and clubfoot being 26.4% were noted, highlighting the need to acknowledge the need for paediatric orthopaedic surgery in Pakistan.

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Authors' Contribution

Following authors have made substantial contributions to the manuscript as under:

FO & HUH: Data acquisition, data analysis, critical review, approval of the final version to be published.

HFUR & AMK: Study design, data interpretation, drafting the manuscript, critical review, approval of the final version to be published.

MYW & MU: Conception, data acquisition, drafting the manuscript, approval of the final version to be published.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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