An 8-year review of upper limb congenital differences at a teaching hospital in Jordan

A retrospective study

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ABSTRACT

الأهداف: مراجعة نوع الاختلافات الخلقية في الطرف العلوي لدى المرضى الذين يتم فحصهم في مستشفى ثالثي في الأردن وتقييم فعالية نظام OMT.

المنهجية: تمت مراجعة الملفات الطبية والأشعة السينية لـ 222 مريضاً يعانون من اختلافات خلقية في الأطراف العلوية. تم تصنيف جميع هذه الحالات باستخدام نظام تصنيف OMT.

النتائج: حددت المراجعة الدقيقة للملفات الطبية 222 مريضًا يعانون من 295 حالة اختلافات خلقية في الطرف العلوي. وكان معدل الانتشار 45/10000 مريض. كان متوسط عمر المرضى 6.18±5.5 سنوات، مع معدل انتشار أعلى عند الذكور 54.1%. كانت معظم الحالات عبارة عن تشوهات 176 (79%) وكان تعدد الأصابع الوحشي هو الأكثر شيوعًا (18.5%). تم تحديد 28 حالة خلل التنسج. تم تشخيص 15 مريضا (6.7%) بالمتلازمات، والتي أظهرت متلازمة بولند كأعلى معدل تكرار لها، بنسبة 2.3% من المجموع. أقل عرض كان للتشوهات الشاذة (1.4%).

الخلاصة: شكلت فئة التشوه معظم الاختلافات الخلقية في الطرف العلوي في هذه الدراسة. كان تعدد الأصابع الوحشي هو الأكثر شيوعًا في هذه الفئة. وبالنظر إلى الاختلافات الإقليمية، فإن الحاجة إلى بنية تحتية راسخة للرعاية الصحية تعد خطوة حيوية نحو إدارة وتحسين نتائج هؤلاء المرضى.

Objectives: To review the types of upper limb congenital differences in patients seen in a tertiary hospital in Jordan and to evaluate the effectiveness of the Oberg-Manske-Tonkin (OMT) system.

Methods: The medical charts and the X-rays of 222 patients with upper limb congenital differences were reviewed. All these cases were categorized using the OMT classification system.

Results: A careful review of the medical charts identified 222 patients with 295 upper extremity anomalies. The prevalence was 45/10000 patients. The mean age of the patients was 6.18±5.5 years, with a higher prevalence in males 54.1%. most cases were malformations 176 (79%) and of these the radial polydactyly was the most common (18.5%). A total of 28 cases of dysplasia were identified. A total of 15 (6.7%) patients were diagnosed with syndromes, of which Poland syndrome showed the highest frequency at 2.3% of the total. The least presentation was for the deformation anomalies (1.4%).

Conclusion: The malformation category constituted most of the congenital upper extremity difference in this study. Radial polydactyly was the most common in this category. Considering the regional differences, the need for a well-established healthcare infrastructure is a vital step toward managing and improving the outcomes of these patients.

Keywords: upper extremity deformities, prevalence, OMT classification, malformation, polydactyly

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Studies on the prevalence of congenital deformities contribute to the planning of health services by monitoring the occurrence of such defects, estimating the cost incurred by treatment, and the availability of specialized multidisciplinary teams to treat and follow up with affected children.[1,2] Upper limb congenital deformities are rarely encountered.[3] They have a wide range of clinical presentations from accessory nubbins to the complete absence of an extremity.[4,5] These birth defects can result in severe loss of upper limb function.[6] Classification systems are essential for establishing

a common language to describe a disorder. This makes these classifications necessary for comparative investigations of epidemiology, etiology, and for promoting improvements in treatment protocols.[7]

The incidence of congenital upper limb abnormalities has been studied and recorded in several countries and publications. In the United States, this figure was 3.64 cases per 10,000 live births; in Stockholm, 21.5 cases per 10,000 live births; and in Finland, 5.25 cases per 10,000 live births.[1,7,8] However, due to the absence of a national registry program in Jordan, we could not assess the true incidence of upper limb congenital anomalies.

The prevalence figures of the anomaly types depend on the presence of a well-developed registry system. This is well expressed by a different presentation of the most common anomalies in different societies.[9]

The International Federation of Societies for Surgery of the Hand (IFSSH) recommended the use of the Oberg-Manske-Tonkin (OMT) classification in 2014 that provides several advantages that allow for the correct identification and classification of upper limb congenital differences.[10] The OMT classification has been updated over the years. The update for 2020 took into consideration the respective diagnoses on the upper-extremity congenital anomalies, timing of insult and its relation to the developmental errors and dysmorphology, and improved terminology for the clarification of several diagnoses.[11] It is divided into 4 major categories: malformations, deformations, dysplasias, and syndromes. It uses dysmorphological descriptions to determine the limb axis, whether the anomaly affects the whole upper limb or the hand plate alone.[8]

This study aimed to assess and evaluate the pattern of distribution of different congenital differences in a tertiary center in Jordan, along with the implementation of the OMT classification system.

Methods. We carried out a retrospective review of the medical charts and the X-rays of all patients with congenital upper limb differences who were evaluated at the Orthopedic Division, Hand and Pediatric Orthopedic Specialty Clinics, University of Jordan, Amman, Jordan, between 2014-2022. We used the ICD 10 codes of musculoskeletal congenital anomalies (Q65-Q79) to extract the patients' records from our hospital registry system. A total of 222 patients were identified and reviewed. Each patient's diagnosis had been confirmed by a senior consultant in the clinic at the first visit and documented in the patients' medical charts. Those charts were reviewed again to check and confirm the diagnosis depending on the relevant clinical and operative notes in case these patients were operated on, in addition to the radiographic images when available. A second check on the data to confirm the data documented was carried out. The findings were discussed with the help of the online application developed by Leon[12] to reach a final decision. We included all syndromes associated with upper limb anomalies. We excluded patients with only lower limb deformities.

This study was carried out after obtaining an ethical approval from the institutional review board of the University of Jordan, Amman, Jordan, with approval reference number 10/2021/5892. The study was carried out according to Helsinki's declaration. No patients' consent was needed, due to the retrospective nature of this research.

All identified cases were categorized using the OMT classification system.[11] The OMT system has 4 general categories that depend on the framework of dysmorphology. These are malformations, deformations, dysplasias, and syndromes.[8,11] If the patient had symmetrical bilateral anomalies, then he was counted as one case for analytical purposes, but if he had different anomalies in both upper limbs, then he was counted as 2 cases. Syndromic patients with upper limb differences were counted once under the "category of syndromes" and never repeated under other categories. The application OMT medical reference, developed by Leon LW, was utilized secondarily during this study to ease the diagnosis process.[12]

Statistical analysis. We used the Statistical Packages for the Social Sciences, version 18.0 (SPSS Inc., Chicago, Ill, USA) for the analysis in this research. The descriptive analyses used for the categorical data (literality, gender, and age) to present the sample and then to present their distribution according to the OMT classification.

The prevalence of the most common congenital upper limb differences presented with its frequency to illustrate the exact diagnosis before the OMT classification.

Results. Among 48978 patients who were seen in the specialty clinics for the same period of the study, 222 patients with 295 upper extremity anomalies were identified. The risk was 45/10000 patients for the same period. Patients' age ranges from 0-28 years. The mean age was 6.18±5.5 years. The right upper extremity was affected in 78 (35.1%) patients, the left in 71 (32.0%), and both limbs in 73 (32.9%). In these 222 patients,

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the prevalence of upper limb anomalies was higher in males (Table 1).

All patients included in the study were classified according to the OMT classification. There were 176 malformations; 54.5% of them were in males. Dysplasia was seen in 28 cases, again with males' predominance (53.6%). Contrary to that, syndromes were seen slightly more in females' patients. The least presentation was for the deformation anomalies (Table 2).

Regarding the laterality, the majority of the malformation category, 63 (35.8%) cases, were in the right upper limb, and the least were the bilateral cases (30.1%). Among the dysplasia cases, there were 13 (46.4%) bilateral cases, and only 4 (14.3%) cases were left laterality. The syndromic cases showed equal distribution between the left-sided and the bilateral cases with 40.0% each (Table 3).

Table 4 shows the relative frequencies of all cases distributed based on their anatomical location. The radial polydactyly was constituting most of the malformation category with 41 (18.5%) cases out of 222 patients. While Poland syndrome showed the highest frequency among syndromic patients in our series 5 cases and formed 2.3% of the total (Table 4).

Table 1 - Demographic characteristics of patients with congenital hand anomalies.

Variables	n (%)
Laterality	
Bilateral	73 (32.9)
Left	71 (32.0)
Right	78 (35.1)
Gender	
Female	102 (45.9)
Male	120 (54.1)

Values are presented as numbers and percentages (%).

Table 2 - Gender distribution of congenital hand anomalies.

OMT classification	Gender			
	Female		Male	
	Row	Column	Row	Column
Malformations	80 (45.5)	80 (78.4)	96 (54.5)	96 (80.0)
Deformations	1 (33.3)	1 (1.0)	2 (66.7)	2 (1.7)
Dysplasias	13 (46.4)	13 (12.7)	15 (53.6)	15 (12.5)
Syndromes	8 (53.3)	8 (7.8)	7 (46.7)	7 (5.8)
Total	102 (45.9)	102 (100)	120 (54.1)	120 (100)

Values are presented as numbers and percentages (%).
OMT: Oberg-Manske-Tonkin classification

Table 3 - Laterality of congenital hand anomalies.

OMT classification	Laterality		
	Bilateral	Left	Right
Malformations	53 (30.1)	60 (34.1)	63 (35.8)
Deformations	1 (33.3)	1 (33.3)	1 (33.3)
Dysplasias	13 (46.4)	4 (14.3)	11 (39.3)
Syndromes	6 (40.0)	6 (40.0)	3 (20.0)
Total	73 (32.9)	71 (32.0)	78 (35.1)

Values are presented as numbers and percentages (%).
OMT: Oberg-Manske-Tonkin classification

Discussion. The population of Jordan was 11.3 million people in 2022. The birth rate for the same year was 21.6 per 1000 people. The urban population constitutes 91.8% of the total population.[13] Since the current study data are confined to a single center, the prevalence of upper limb congenital anomalies per live birth in Jordan cannot be assessed, especially in the absence of a national registry program in Jordan. But the prevalence of the upper limb differences among the total patients seen for the same period in the specialty clinics was 45/10000. Disparities in socioeconomic status present in Jordan worsen the burden of these anomalies. It may be difficult for low-income families, especially those who reside in remote locations far from major cities to access healthcare facilities for the early diagnosis and surgical management of these conditions, which can be costly.

Such cases need a multidisciplinary team including orthopedic and plastic surgeons, pediatricians, and a rehabilitation specialist to provide the best care for these children. Moreover, when we consider syndromic patients, there is a need for geneticists and social services to be involved in the management team.

Congenital anomalies of the upper extremity occur in approximately 0.16-0.18% of live births. Nearly 10% of them will have either partial or complete deficiency of the limb.[6] These anomalies are a major cause of long-term disability. Recognizing the burden of these disabilities is a cornerstone in planning measures for the care and prevention of these anomalies.[14]

Classification systems play a crucial role in assisting with diagnostic and therapeutic procedures when dealing with diseases and syndromes. Classification systems also ease communication between doctors, bypassing the language barrier that may exist and allowing for a pragmatic approach. Information and research surrounding the incidence of congenital upper limb anomalies are extremely scarce in Jordan, as well as the entire Middle Eastern region. These cases were referred to our center directly from obstetric clinics or peripheral hospitals from different cities in Jordan.

Table 4 - Prevalence of the most common different congenital upper limb anomalies based on anatomical location.

Anatomical locations	n (%)
Finger	
Polydactyly radial	41 (18.5)
Syndactyly simple	36 (16.2)
Polydactyly ulnar	19 (8.6)
Symbrachydactyly	9 (4.1)
Syndactyly complex	9 (4.1)
Polysyndactyly radial	7 (3.2)
Camptodactyly	4 (1.8)
Thumb in palm deformity	4 (1.8)
Triphalangeal thumb	3 (1.4)
Brachydactyly	2 (0.9)
Constriction band sequence	2 (0.9)
Macrodactyly	1 (0.5)
Synopolydactyly thumb	1 (0.5)
Thumb hypoplasia	1 (0.5)
Hand	
Cleft hand	7 (3.2)
Enchondromatosis	2 (0.9)
Forearm	
Club hand radial	13 (5.9)
Club hand ulnar	6 (2.7)
Synostosis radioulnar	6 (2.7)
Exostosis forearm	3 (1.4)
Congenital dislocation of the radial head	2 (0.9)
Fibrous dysplasia	1 (0.5)
Madelung deformity	1 (0.5)
Shoulder	
Sprengel's scapula	13 (5.9)
Syndromes	
Poland syndrome	5 (2.3)
Down syndrome	3 (1.4)
Cenani Lenz syndrome	2 (0.9)
Apert syndrome	1 (0.5)
Ellis van creveld syndrome	1 (0.5)
Larsen syndrome	1 (0.5)
Rubinstein Taybi syndrome	1 (0.5)
Vactrels syndrome	1 (0.5)
Whole limb	
Epiphyseal abnormalities	5 (2.3)
Hemihypertrophy	5 (2.3)
Arthrogryposis	3 (1.4)
Neurofibromatosis	1 (0.5)
Total	222 (100)

Values are presented as numbers and percentages (%).

Analyzing these deformities in a tertiary center in Jordan through a well-designed classification system helps to identify the need for specialized personnel to deal with these cases whilst incurring the lowest possible cost. Both pediatric orthopedic surgeons and plastic surgeons deal with these congenital anomalies in Jordan. A Turkish study published in 2020, with Turkey being the closest country geographically, reported 1050 upper limb anomalies in a time span of 5 years, with malformations being the most recurring (n=865), followed by deformations (n=114), and dysplasias (n=71).[4] The shortage of information in the Middle East and North Africa regions calls for a need to divert attention towards the application of the OMT classification to better document and manage upper limb congenital anomalies.

Looking at a broader scale publication regarding the OMT classification, a Korean nationwide population-based study reported 10,704 patients with anomalies with an incidence of 23.5 per 10,000 live births.[15] Of these, 6,174 (57.7%) males and 4,530 (42.3%) females were affected, which reflects a distribution comparable to that of our own study. Moreover, they showed polydactyly to be the most common anomaly, followed by syndactyly, and limb deficiency.[15] Our study reported nearly double prevalence which is mostly due to the nature of the hospital-based study that was carried out in comparison to this population-based study.

Goldfarb et al[8] examined 3 Midwestern referral centers, including 653 congenital upper limb anomalies. As in our study, malformations were the most common anomaly (74%); however, they cited arthrogryposis as the most common specific anomaly, while in the current study, it was radial polydactyly. Deformation was seen in 13% of patients in a study by Goldfarb et al.[8] By contrast, in the current study, only 2.7% showed deformations. This might be explained by the classification update from 2020, where trigger finger was omitted from the deformation category because many investigators showed that trigger finger was not present at birth, and the name congenital trigger finger is misleading.[11] They had 86 patients with dysplasia and 98 with syndromes.[8]

Ekblom et al[16] reclassified 562 patients from their previous study, which was based on the IFSSH classification, using the OMT classification. Similar to the current study, the most common category documented was malformations (n=429), which comprised 74% of their cohort study, whereas deformations (n=124) made up 22%, dysplasias (n=10) 2%, and syndromes (n=14) made up 2%. Similarly, the use of OMT enabled the inclusion of all patients, showing a very similar data distribution. In contrast to our study, Ekblom et al[16] excluded arthrogryposis from the classification due to differences among the authors regarding the diagnostic accuracy. Odatuwa et al[17] studied the congenital differences of the upper limb in tertiary centers in Nigeria. They reported 46 patients with 53 upper limb congenital anomalies. Contrary to the current study, they reported syndactyly as the most common anomaly.

Modifications that were requested by Ekblom et al[16] and accepted by the IFSSH made it easier to classify anomalies. These modifications included better

clarification surrounding hand plate involvement in brachydactyly, and transverse deficiency without proximal involvement.

Certain anomalies, such as amelia, were not observed in this series of patients. This might have been because few patients were included in this study. In addition to proper care, pregnant mothers received advice regarding multivitamin use early in pregnancy, and warnings on the use of certain teratogenic medications.

The OMT classification system allowed all patients in this study to be classified. The greatest limitation of the use of OMT is the classification of several syndromes under multiple headings, giving room for the inclusion of the same patient twice. However, in the current study, all were classified only under the syndrome category.

Study limitations. The first limitation of this study was noted to be the small sample size, which can be attributed to several factors, the most important being the inadequate registry of patients. The other factor is the presence of 2 other tertiary referral centers in the capital Amman, Jordan; therefore, many patients are referred to those other hospitals. The second limitation was that the study did not reflect the exact incidence of anomalies in the population, since Jordan University Hospital, Amman, Jordan, is a tertiary hospital and does not represent the entire population but rather a small percentage. This might lead to inflation of the prevalence of cases due to the characteristics of the hospital-based studies. A national registry program and future multicentric study might reveal the true incidence of congenital upper limb anomalies. Moreover, it might show the geographic distribution of these cases and emphasize the need to provide proper health care services there.

In conclusion, a high prevalence of the congenital upper limb differences was observed in this hospital-based study. The malformation category formed most of these differences. Radial polydactyly was the most common in this category. A national wise registry program is of outmost importance to identify the true population-based incidence. Taking into consideration regional differences, the need for a well-established healthcare infrastructure is a vital step toward managing and improving the outcomes of these patients.

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