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The Prevalence of Congenital Hand and Upper Extremity Anomalies based upon the New York Congenital Malformations Registry

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Abstract

Background—There have been few publications regarding the prevalence of congenital upper extremity anomalies and no recent reports from the United States. The purpose of this investigation was to examine the prevalence of congenital upper extremity anomalies in the total birth population of New York State over a 19-year period utilizing the New York Congenital Malformations Registry (NYCMR) database.

Methods—The NYCMR includes children with at least one birth anomaly diagnosed by 2 years of age and listed by diagnosis code. We scrutinized these codes for specific upper extremity anomalies, including polydactyly, syndactyly, reduction defects, clubhand malformations, and syndromes with upper limb anomalies. We included children born between 1992 and 2010.

Results—There were a total of 4,883,072 live births in New York State during the study period. The overall prevalence of congenital upper extremity anomalies was 27.2 cases per 10,000 live births. Polydactyly was most common with 12,418 cases and a prevalence rate of 23.4 per 10,000 live births. The next most common anomalies included syndactyly with 627 cases affecting the hands (1498 total) and reduction defects (1111 cases). Specific syndromes were quite rare and were noted in a total of 215 live births. The prevalence of anomalies was higher in New York City compared to New York State populations at 33.0 and 21.9 per 10,000 live births respectively.

Conclusions—The NYCMR data demonstrates that congenital upper extremity anomalies are more common than previously reported. This is in large part due to the high prevalence of polydactyly. While registries are imperfect, such data are helpful in monitoring prevalence rates over time, identifying potential causes or associations, and guiding health care planning and future research.

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Introduction

There have been few reports on the prevalence of congenital upper extremity anomalies. These epidemiology data are important to allow the identification of risks to the public health.[1–10] The most striking example is that of thalidomide,[1] the medication used in the late 1950s and early 1960s for pregnancy-related nausea, which caused severe birth anomalies of the extremities. Another example, especially pertinent for New York State, is Love Canal, a neighborhood in Niagara, New York. Love Canal gained attention in the 1970s when previously buried toxic waste leaked and created a public health ‘emergency’ and, eventually, was associated with birth anomalies. Ultimately, these public health catastrophes prompted the development of many of today’s state and national registries. Appropriate funding and monitoring of registries have the potential to avoid or minimize the adverse affects from such risks to the public health. Additionally, an understanding of prevalence allows appropriate resource utilization including training and research expenditures.

Three studies inform our understanding of upper extremity birth anomalies. Giele, et al utilized a total population study of Western Australia over 11 years and reported an upper limb anomaly prevalence of 1 in 506 live births (19.8 per 10,000 live births).[2] Koskimies, et al evaluated the Finnish Registry of Congenital Malformations over 13 years and found an incidence of upper limb abnormalities of 5.25 per 10,000 live births.[3] And Ekblom, et al, utilizing multiple registries in Sweden, documented a national upper limb anomaly incidence of 21.5 in 10,000 live births.[4] The variability is based on differing populations, registry differences, and different inclusion criteria. Nonetheless, these data help frame any discussion of the impact of congenital upper limb anomalies on society.

The prevalence figures noted above share a commonality of being products of well-developed national or regional registry systems and patient populations with decreased mobility. Prevalence data in the United States has been challenging as the registries are less well developed and more focused on severe, systemic conditions rather than upper extremity anomalies. The limited data that is available is state-based only. Additional challenges for a registry in the United States include a much larger and more mobile population. Commonly referenced prevalence figures for the United States are most often clinic visit data or local population data[5–7] these have provided estimates of prevalence at 11.4 to 16 per 10,000 births.

The purpose of this investigation was to utilize the New York Congenital Malformation Registry (NYCMR) to assess the prevalence of congenital upper extremity limb anomalies in the defined populations of New York City and New York State.

Materials and Methods

Institutional Review Board approval was obtained for this investigation. We reviewed the New York Congenital Malformations Registry (NYCMR) from 1992 to 2010 to identify cases of upper extremity congenital anomalies; all data were blinded. The NYCMR was begun in 1982 funded in response to the thalidomide disaster and the Love Canal pollution

disaster.[8] It is one of the largest such registries in the country and annually includes approximately 11,000 children with birth anomalies among 270,000 live births.[9]

The registry identifies patients with congenital anomalies among live births in the state of New York. Hospitals and health care providers are required to report specific malformations identified in children up to 2 years of age. There are specific requirements for physician and hospital reporting and the NYCMR staff fills information deficits with the patient medical record; additional information may be requested and the Medical Director may become involved if there is uncertainty with diagnosis.

Each report is compared against previous entries to prevent duplication. Periodic audits and regular assessments of accuracy and comprehensiveness are performed including a review of discharge data, discharge summaries, and on- site audits.[9] The data ascertainment for this registry is considered, therefore, a combination of active and passive. The sensitivity of the NYCRM data has been estimated at 86.5%, [11] similar to other registries including the Metropolitan Atlanta Congenital Defects Program (MACDP), considered the gold standard registry in the United States.

Trained NY state officials code each of the diagnoses according to the 1979 British Pediatric Association (BPA) Classification of Diseases and the International Classification of Diseases, 9th revision, Clinical Modification (ICD-9-CM). The BPA codes are used for greater specificity and due to its wider adoption in similar registries. Staff members search for duplicate entries and address incomplete records using birth certificate data and direct contact with reporting hospitals.

We searched the data sets for the following general BPA codes and included subcategories:

- 755.0, polydactyly
- 755.1, syndactyly
- 755.2 (including 755.250, 755.260, 755.270, among others, representing reduction defects of upper limb: split hand malformation, split hand and foot malformation, absent upper limb, absent upper arm and forearm, absent forearm only or upper arm only, absent forearm and hand, absent hand or fingers, pre-axial longitudinal reduction defect of upper limb, post-axial longitudinal reduction defect of upper limb, and unspecified reduction defect of upper limb
- 759.840, and 759.890 Syndromes and associations with common upper extremity anomalies including: Holt- Oram, thrombocytopenia absent radius, VACTERL, Nail Patella, and Poland Syndromes.

Each clinically identified upper extremity case included at least one (and often several) of the above BPA codes together with a written description. We manually sorted these data to best classify each case. To assist with this process, we established certain rules for the diagnoses to address potential confusion and to prevent repetitive data counts:

- For reduction defects, if more than one code was utilized for a particular patient, we only included the patient under the code representing the most proximal/most severe anomaly

- For patients coded under syndactyly and fused digits, we only counted the entry as syndactyly
- For patients coded as missing fingers and split hand, we only counted them as split hand (i.e., cleft hand)
- For patients with a Not Otherwise Specified (NOS) code and a more specific, overlapping code, we only counted the patient's anomaly as the more specific code only.
- Patients with a syndrome diagnosis and a specific upper limb anomaly were only counted one time, as a syndrome but only if the syndrome could be readily identified from the narrative information or the constellation of anomalies. For example, a patient with a diagnosis of Holt Oram syndrome was only counted as a syndrome and not counted in the reduction defect category.
- Patients with a syndrome diagnosis and a specific upper limb anomaly in which the syndrome could not be readily identified were only counted as the specific upper limb anomaly.

The New York City and the New York State (exclusive of New York City) data were provided to us in separate data sets. The New York City information was provided in 4 sets: 1992, 1993–2005, 2006–2007, and 2008–2010. The New York State data was provided in 3 sets: 1992, 1993–2003, and 2004–2010. These data sets were searched for each diagnosis code to determine prevalence for New York City, New York State, and combined. Due to the report variability by year, we did not assess change over time but we were able to compare overall New York City vs New York State data utilizing the chi-square test (or Fisher exact test for smaller sample sizes). Significance was set at $P < .05$ and 95% confidence intervals were computed.

Results

There were a total of 4,883,072 live births between 1992–2010 in New York State. This includes 2,314,661 births in New York City and 2,568,411 in New York State. We identified 13,278 upper extremity anomalies, including those patients with syndromes affecting the upper extremities. Therefore, 0.272% of newborns (95% confidence intervals 0.267%–0.277%) were affected, a prevalence rate of 27.2 per 10,000 live births (Table 1).

Table 1 summarizes our findings. Polydactyly was the most common diagnosis but the data did not allow a reliable separation of preaxial and postaxial polydactyly. Reduction defects considered together were next most common, including transverse deficiency, longitudinal deficiency and split hand anomaly. Syndactyly of the hand and foot were less frequently noted.

There were 215 children identified with a syndrome or association, a prevalence of 0.44 per 10,000 live births. The most common of these was Amniotic Constriction Band with 78 cases. Table 1 provides further details on the cohort as a whole including confidence intervals and prevalence breakdowns.

The data were divided into City and State cohorts allowing data comparison for the 19 years in total. Table 2 provides a breakdown with a comparison of anomaly prevalence between the two groups. There were significantly more patients with polydactyly in the New York City group, with a prevalence of 29.9 compared to 17.5 per 10,000 live births in the New York State group, $P < .0001$. However, for all other diagnoses and syndromes, there were significantly more anomalies in the New York State group.

Discussion

In the United States, data from the National Birth Defects Prevention Network (NBDPN) have been utilized to assess major birth anomalies and are tracked annually. To date, there have been few assessments of specific limb anomalies with the available surveillance data; our understanding of extremity congenital anomalies has the potential to be greatly increased with further study. Two recent works highlight the benefits of assessing registry data. Chen, et al assessed the risk of maternal caffeine consumption and congenital limb deficiencies.[12] A weak increased risk of limb deficiency related to caffeine consumption was identified but this did not vary based on the amount of caffeine consumed. Parker, et al reported the prevalence of clubfoot using several surveillance programs and confirmed associations with maternal age, parity, education, and marital status.[13]

The utility of the NBDPN has been demonstrated with a number of assessments of major birth anomalies. There are 30 population based surveillance programs that comprise the NBDPN and report data on 45 major birth anomalies. Parker, et al reported the prevalence of selected upper extremity birth defects from 2004–2006 using 14 of these programs to assess 21 birth anomalies and to report estimated national prevalence data.[14] The prevalence of upper extremity reduction defects was estimated to be 3.49 per 10,000 live births with an estimated 1454 annual cases. In a more recent study focusing on race/ethnicity utilizing 12 of the surveillance programs (approximately 1 in 3 United States births), the prevalence of upper extremity reduction defects was found to be 2.85 per 10,000 live births.[15] These population- based studies provide a useful but basic view of congenital anomalies with only broad information on the prevalence of reduction defects. There are no granular data on different types of reduction defects, such as radial longitudinal deficiency or cleft hand, or information on other upper extremity anomalies such as polydactyly or syndactyly.

A comparison of three of the large ‘State’ registries in the United States demonstrates grossly similar findings despite differences in the registry programs.[8] Using prevalence data from 1995 and 1996, upper extremity reduction defects varied from 2.7 (NYCMR) to 3.9 (Metropolitan Atlanta Congenital Defects Program - MACDP) to 4.1 (California Birth Defects Monitoring Program- CBDMP) per 10,000 births. The NYCMR includes only live births and is a mixed active and passive case ascertainment program, two factors likely accounting for at least some of the difference compared to the other two registries.

In the most specific prior report, Correa, et al assessed the MACDP data between the years of 1968–2003 to assess a variety of congenital anomalies.[16] This population- based surveillance of 5 counties around Atlanta, Georgia is highly regarded given its durability and

data acquisition methods. There is multisource active case ascertainment in children up to 6 years of age with data and clinical review of all cases.

In this report, there were 32,938 total anomalies among the 1,232,191 births for an anomaly prevalence of 26.7 per 10,000 live births. An assessment of upper extremity anomalies identified 1913 children with polydactyly (a prevalence of 15.53 per 10,000 live births), 377 with a transverse limb deficiency (3.06/10,000), 176 with a longitudinal limb deficiency (1.43/10,000), 49 with a split hand and/or split foot anomaly (0.40/10,000), and 40 with an intercalary limb deficiency (0.32/10,000). These data vary somewhat from our findings as summarized in Table 1.

A comparison of New York City and New York State anomaly prevalence over the 19-year study period demonstrated that polydactyly was more frequent in New York City while all other anomalies were more common in New York State. This difference in polydactyly prevalence is likely explained by the racial disparity between the population of New York City and New York State. In 2013, 25.5% of the population of the New York City was African American whereas New York State (exclusive of New York City) was only 8.7% African American.[17] Given that postaxial polydactyly is found predominately in African Americans, the higher prevalence of polydactyly in New York City is not surprising. The MADCP data also confirmed notable differences along racial lines for polydactyly. The prevalence of polydactyly was 11.61 per 10,000 for whites, 23.18 per 10,000 in African Americans, and 12.16 per 10,000 in Hispanics. Furthermore, the higher percentage of African Americans in both the NYCMR and the MACDP populations likely account for prevalence differences compared to the Scandinavian (5.9 per 10,000)[4] and Australian data (Table 3).

The NYCMR population differed from previous reports for two other conditions. Longitudinal deficiency was present in 0.5 per 10,000, notably less than the Swedish and Finnish populations at 3.1[4] and 2.1[3] per 10,000 respectively. In addition, there were 172 cases of radial longitudinal deficiency and 36 cases of ulnar longitudinal deficiency, a ratio of approximately 5:1. While traditional reports have cited an even higher difference in prevalence, Ekblom et al's recent Swedish epidemiological study demonstrated a prevalence of 1.3 and 1.0 for RLD and ULD per 10,000 births respectively.[4] In addition, there is a seemingly low prevalence of syndactyly at 1.3 per 10,000. This is lower than expected and likely excludes partial syndactyly and may relate to underreporting of complete syndactyly; however, the prevalence is remarkably similar to the Swedish prevalence of 1.4[4]. This does call into question a long held belief that syndactyly is the most common, or second most common, congenital anomaly.[18–20]

Our report is limited by the weaknesses of any registry report. The NYCMR utilizes a mixed case ascertainment with passive inclusion and active confirmation. It is an imperfect registry for the identification of specific upper extremity congenital anomalies as confirmation of such anomalies often requires clinical and radiographic data assessment by upper extremity experts (i.e., a congenital hand surgeon). Within the data analyzed, we believe that the polydactyly and syndactyly data are the most reliable as these cases were easiest to identify and did not require inclusion “rules”. In contrast, the cases of reduction defects are least

reliable due to variability in coding and the fact that these diagnoses are less straightforward to diagnose.

In conclusion, using the NYCMR database of nearly 5 million live births, we found the prevalence of upper extremity congenital anomalies to be 27.2 per 10,000 births. Polydactyly was the most common anomaly with a prevalence of 23.4 per 10,000 births, and syndactyly and reduction defects were somewhat less common. While registry data are imperfect, we believe these data from New York provide useful information on upper extremity birth anomalies.

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TABLE 1

TOTAL NEW YORK STATE POPULATION 1992–2010

4,883,072 live births				
Anomaly of Interest	Number of cases	Percentage of total births	95% CI for Percentage	Prevalence (per 10,000 live births unless otherwise indicated)
<i>Polydactyly</i>	11418	0.234%	0.230 – 0.238%	23.4 per 10,000
Polydactyly, hand	10529	0.216%	0.212 – 0.220%	21.6
Polydactyly, hand and foot	783	0.016%	0.015 – 0.017%	1.60
Polydactyly, NOS	106	0.002%	0.002 – 0.003%	0.22
<i>Syndactyly</i>	627	0.013%	0.012 – 0.014%	1.29 per 10,000
Syndactyly, hand	458	0.009%	0.009 – 0.010%	0.94
Syndactyly, hand and foot	115	0.002%	0.002 – 0.003%	0.24
Syndactyly, NOS	19	0.0004%	0.0002 – 0.0006%	0.04
Apert Syndrome	33	0.0007%	0.0004 – 0.0009%	0.068
Carpenter Syndrome	2	0.0000%	0.0000 – 0.0001%	0.004
<i>Reduction Defects</i>	1111	0.023%	0.022 – 0.025%	2.3 per 10,000
Split hand malformation	66	0.001%	0.001 – 0.002%	0.14
Split hand and foot malformation	15	0.0003%	0.0002 – 0.0005%	0.03
Absent upper limb	16	0.0003%	0.0002 – 0.0005%	0.03
Absent upper arm and forearm	39	0.0008%	0.0005 – 0.0010%	0.08
Absent forearm only or upper arm only	59	0.001%	0.001 – 0.002%	0.12
Absent forearm and hand	59	0.001%	0.001 – 0.002%	0.12
Absent hand or fingers	453	0.009%	0.008 – 0.010%	0.93
Preaxial longitudinal reduction defect of upper limb	172	0.004%	0.003 – 0.004%	0.35
Postaxial longitudinal reduction defect of upper limb	36	0.0007%	0.0005 – 0.0010%	0.07
Unspecified reduction defect of upper limb	138	0.003%	0.002 – 0.003%	0.28
VATER/VACTERL	44	0.0009%	0.0006 – 0.0012%	0.090
Thrombocytopenia-absent radius (TAR)	4	0.0001%	0.0000 – 0.0002%	0.008
Holt Oram syndrome	10	0.0002%	0.0001 – 0.0003%	0.020
<i>Other Syndromes</i>				
Amniotic Band Syndrome	78	0.002%	0.001 – 0.002%	0.160
Poland Syndrome	18	0.0004%	0.0002 – 0.0005%	0.037
Klippel-Trenaunay-Weber syndrome	16	0.0003%	0.0002 – 0.0005%	0.033
Rubenstein-Taybi syndrome	8	0.0002%	0.0001 – 0.0003%	0.016
Sirenomelia	2	0.0000%	0.0000 – 0.0001%	0.004
TOTAL # ANOMALIES	13156	0.269%	0.267 – 0.277%	27.2 per 10,000

CI = confidence interval.

NEW YORK CITY VS NEW YORK STATE 1992–2010

Table 2

Anomaly of Interest	NEW YORK CITY Total Live Births NYC 1992–2010 = 2314661				NEW YORK STATE Total Live Births NYS 1992–2010 = 2568411				p-value [*]
	Number of cases	Percentage of total births	95% CI for Percentage	Prevalence (per 10,000 live births unless otherwise indicated)	Number of cases	Percentage of total births	95% CI for Percentage	Prevalence (per 10,000 live births unless otherwise indicated)	
<i>Polydactyly</i>	6938	0.300%	0.293 – 0.307%	29.9	4480	0.174%	0.169 – 0.180%	17.5	<0.0001
<i>Syndactyly (excluding syndromes)</i>	238	0.010%	0.009 – 0.012%	1.03 per 10,000	354	0.014%	0.012 – 0.015%	1.38 per 10,000	0.0005
<i>Reduction Defects (excluding syndromes)</i>	401	0.017%	0.016 – 0.019%	1.68 per 10,000	652	0.025%	0.023 – 0.027%	2.46 per 10,000	<0.0001
<i>Syndromes</i>	67	0.003%	0.002 – 0.004%	0.290 per 10,000	148	0.006%	0.005 – 0.007%	0.576 per 10,000	<0.0001
TOTAL # ANOMALIES	7644	0.330%	0.323 – 0.338%	33.0	5634	0.219%	0.214 – 0.225%	21.9	<0.0001

CI = confidence interval.

^{*} P-value compares the percentage of total births with the specified anomaly in NY City versus NY State by chi-square test, unless otherwise indicated[†] P-value by Fisher's exact test due to small sample sizes in the contingency table.

Table 3

Comparative Prevalence in the Literature

	Current	Eklblom *	Koskimies	Giele	MACDP
Polydactyly	23.4	5.9		7.5	15.5
Syndactyly	1.3	1.4			
Transverse Deficiency	1.3	0.8	2.6		3.1
Longitudinal Deficiency	0.5	3.1	2.1		
Cleft Hand	0.2	0.3	0.5		0.4

* Eklblom, et al 2010