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Review

The Operative Incidence of Syndactyly in Northern Ireland. A 10-Year Review.

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ABSTRACT

BACKGROUND

Syndactyly is a common congenital condition that can present sporadically or in relation to an underlying genetic condition. Little contemporary published data exists detailing specific rates of presentation and surgical intervention, especially in Western European population. This is the first published review of operative intervention rates for the condition over time in Northern Ireland.

METHODS

A ten-year retrospective review of electronic operative records from January 2007 – October 2017 was carried out within Northern Ireland's regional tertiary centre Royal Belfast Hospital for Sick Children (RBHSC). All congenital hand surgery in the country was performed here during the period reviewed, by a single surgeon. Patient age at surgical intervention, their sex, digits involved and clinical grade of syndactyly was recorded.

RESULTS

One hundred and twenty four cases were returned following the review. On individual analysis 22 cases were excluded as they were not primary congenital syndactyly. The remaining 102 cases were all Caucasian. Six cases were toe syndactyly while 96 cases involved the upper limb digits. The group consisted of 70 males and 32 female infants. Age range at time of surgical intervention was 8 months to 14 years with a median age of 26 months. For clinical grade of upper limb syndactyly; 35 cases in the data set were classed as simple incomplete, 34 cases as simple complete, 17 as complex and 5 cases as complicated syndactyly. The remaining 5 cases lacked clear documentation. The most common site of syndactyly was between the ring and middle finger (40/102). Annual frequency of operative intervention has trended upwards in the period studied.

CONCLUSION

This case review adds epidemiological data on the operative incidence of syndactyly cases in Northern Ireland - a relatively isolated genetic population. Overall rates of incidence have increased over the past 10 years. It remains unclear if this is due to new environmental influences on the developing population or increased referral for surgical intervention over time.

Levels of evidence - IV (Case Series)

INTRODUCTION

Syndactyly (Greek Syn=together; Dactylos=digit) is a digital malformation in which adjacent fingers and/or toes are joined by soft tissue or bony bridges beyond the normal web due to failed separation during embryonic limb development[1]. It is one of the most common hereditary limb malformations in the western world, with a prevalence ranging from 3–10 per 10 000 births to 10–40 per 10 000 births in specific settings[2]. The syndrome is twice as likely to occur in males and most commonly found in caucasian populations. The third, fourth, second and first web spaces are affected in decreasing frequency respectfully, and the condition is bilateral in half of cases[3,4].

Published geographic variations in frequency of syndactyly suggest the possibility of environmental as well as genetic influence on the disorder[5]. For syndactyly with or without defined genetic causation, as discussed below, perinatal maternal exposure to noxious stimuli[6] may provoke lack of normal apoptosis of interdigital spaces[7] during development[8,9] leaving the fingers fused, and the hand lacking its normal profile. Maternal health is an independent risk factor for development of the disorder; obesity[10] along with smoking, nutritional status[11] and even viral infection[12] have been suggested as possible aetiological factors for syndactyly development.

At least nine syndactylous genetic entities with various subdivisions have also been classified. These include patients with Mendelian autosomal dominant inheritance, recessive inheritance and even X-linked recessive inheritance[13]. The majority of these cases are non-syndromic. Syndromic syndactyly is a recognised separate genetic entity e.g. Acrocephalosyndactyly, Apert and Pfeiffer Syndrome[14,15].

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Sporadic cases that are non hereditary and non syndromic however globally are the most commonly reported form[16]. Present at birth and lacking a clear genetic association these lie apart from postpartum causes e.g. trauma or burns[13].

Despite environmental risk factors and multiple underlying genetic causes being identified in the literature a lack of published epidemiological reports into the disorder remains. This has previously been identified as a limiting factor for further research into certain related issues such as the skewed sex ratio of presentation[17].

In this review we present the number of syndactyly cases operated on in over a 10-year period in Northern Ireland's single paediatric tertiary surgical centre, by a single surgeon, and discuss changes in operative incidence within our population.

MATERIALS AND METHODS

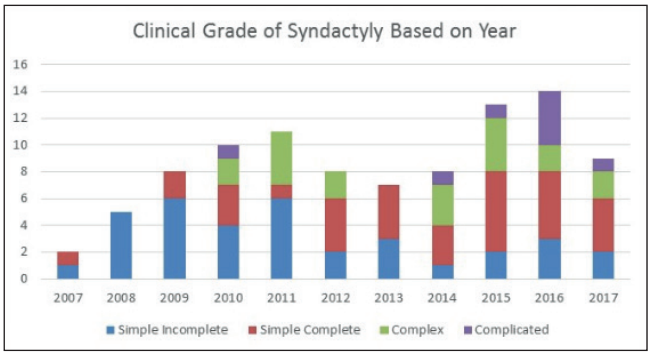
We conducted a ten-year retrospective review of operative records from January 2007 – October 2017 using the Electronic Theatre Management System (TMS) in the Royal Victoria Hospital for Sick Children. This is the only unit in the region where operative intervention for syndactyly occurs thus the data set reflects the totality of operative cases within Northern Ireland within the defined period. Searching for, "Syndactyly," produced a return of 124 cases including the same patient requiring bilateral procedures, digital release or further staged procedures. Cases that were carried out for reasons other than primary congenital syndactyly release e.g. burns were then excluded from the annual totals.

We then analysed each primary syndactyly case individually using electronic or case notes to define age at surgical intervention, sex, digits involved and clinical grade of syndactyly (13). Clinical grade was defined using clinical and x-ray findings to produce; simple incomplete (partial skin bridge between digits), simple complete (complete skin bridge length of digit), complex (where bony and/or bony cartilage bridges are present), complicated (complete bony disruption / interconnection of phalanges). After interrogation of data, cases were presented graphically by intervention rate per year and type of case in an attempt to see if a trend in frequency of syndactyly surgery in the province was present, and if the grade of syndactyly has changed over time.

RESULTS

Of the 124 results returned during the 10 year analysis all the patients were from a Caucasian background and native to the region. Four cases were excluded for syndactyly occurring secondary to burns. A further 18 cases were excluded as these were delayed surgery performed for scar contracture release following the primary syndactyly intervention. The remaining 102 cases were carried out for primary syndactyly release (including staged bilateral release). Six cases were toe syndactyly while the remaining 96 cases involved the upper limb digits. The group consisted of 70 males and 32 female infants. Age range at time of surgery was 8 months to 14 years with a median age of 26 months. For clinical grade of syndactyly 35 cases in the data set were classed as simple incomplete, 34 cases as simple complete, 17 as complex and 5 cases as complicated (figure 1). Annual frequency of operative intervention displayed in figure 2. The most common site of syndactyly was between the ring and middle finger (40 / 102 cases.)

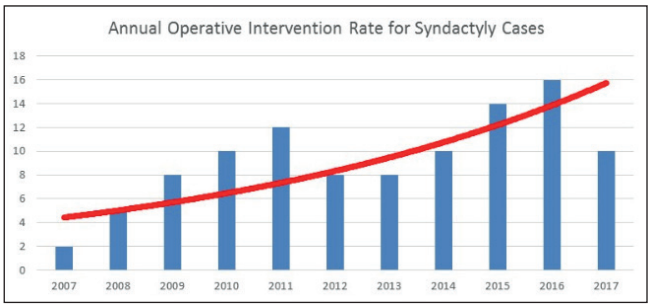
Figure 1



Simple incomplete: partial skin bridge 35/102
Simple complete: full length skin bridge 34/102
Complex: complete bony disruption or interconnection of phalanges 17/102
Complicated: cartilaginous or bony connection of digits 5/102

While the majority of cases had not undergone formal genetic testing, some were found to be syndrome related. Twelve cases presented in patients with a background of Down's Syndrome, five cases with known Poland syndrome, one with Greig cephalopolysyndactyly, one with Phelan-McDermid syndrome and one with Disorganisation Like Syndrome. One patient is additionally undergoing testing for Miller syndrome. Additionally we noted 23 of the patients had been referred for tertiary ENT follow up for a variety of issues such as recurrent tonsillitis or middle ear infections. Frequency of operative intervention increased over the 10 year period analysed (figure 2).

Figure 2



Over the period analysed our operative workload has increased both in number of cases and complexity of those undertaken.

DISCUSSION

The results demonstrate the operative incidence of syndactyly cases in Northern Ireland population has increased over the past 10 years. Unlike previous studies[2] our data highlights that it is upper limb rather than lower limb syndactyly more commonly encountered within the N. Ireland population. However, this could be due to presenting bias of parents requesting surgical intervention on hand syndactyly being a more publicly visible anomaly. Additionally the senior author is reluctant to separate otherwise uncomplicated toe syndactyly due to the risk of developing hypersensitive scar in a weight bearing area.

Our study suggests that in the Northern Ireland population the male: female skew of cases is even greater than previously reported in US populations[17]. Given there is no other recent epidemiological studies of UK syndactyly presentations it is not possible to say if this is due to unexplained genetic pathways or as yet undiscovered environmental factors.

Medical comorbidity of some syndactyly cases was also noted. The link between syndromic syndactyly patients and upper airway issues has previously been widely reported[16]. In syndromes such as Apert where significant associated craniofacial abnormalities occur cocordinaly anaesthetic and airway issues are part of standard preoperative planning[18]. Less clear is the link between sporadic syndactyly and upper airway conditions. Eight referrals to tertiary paediatric Otorhinolaryngology were carried out in our cases series for syndromic patients with recognised upper airway symptomatology. However in 15 cases, no underlying related genetic syndrome was previously diagnosed, prior to an Otorhinolaryngology referral. The literature to date is sparse on the relationship between upper airway conditions and sporadic syndactyly presentations[19]. This is a reminder for all clinicians involved with management of infants with congenital hand to screen for other systemic morbidity as part of their general pediatric examination and work-up.

CONCLUSION

This study provides the first epidemiological review of the operative incidence of syndactyly in the Northern Ireland region over time. Syndactyly continues to present as one of the most common conditions referred to the paediatric hand surgery service in the region. The data presented over the last 10 years highlights an increasing incidence of surgical intervention in the Northern Ireland population, in more complex syndactyly cases.

A number of these patients had additional associated established medical conditions or syndromic causes however the majority of cases we reviewed were sporadic and simple in nature. Medical teams reviewing and managing these conditions regardless should be aware of the possible need for referral to other services should recurrent symptoms in other systems be identified regardless of the underlying genetic cause of syndactyly. This study confirms that although increasing in number with time the general trends displayed e.g. male sex, type and location (middle and ring finger most commonly) are in keeping with other contemporary studies seen elsewhere in both Western Europe and America[20].

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