Analysis of Congenital Hand Anomalies at a Specialist Hospital in a Developing Country

U C Mba, I S Ogbonnaya, I I Onah

Citation

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Abstract
Introduction: Congenital hand anomalies constitute a significant work load in plastic surgery practice in this environment. There is however a dearth of information on the presentation and management of these anomalies in our environment. This study was designed to investigate the pattern of presentation and management, determine prenatal factors associated with the condition and review the literature.

Methods: Medical and theatre records of patients that presented at National Orthopaedic Hospital, Enugu, Nigeria, with congenital hand anomalies between July, 2002 and June, 2012 were examined. Information obtained was analyzed using (SPSS) version 17 and presented in tables and charts.

Results: A total of 43 patients were recorded. They were made up of 16 males and 27 females. The age range was 2 days to 26 years with a peak age group of 0 – 11 months. There was a positive history in first degree relatives in 14% of cases. Syndactyly was the most common observed constituting 37.2% in isolated forms and 25.7% in association with other anomalies and syndromes followed by polydactyly which presented in isolated form in 25.7% of cases and in association with syndactyly in 4.7% of cases. Syndromic forms constituted 14% of cases. 25.6 % of mothers had febrile illness while 9.3% used native medication during early pregnancy. Most patients (93%) had various surgical procedures determined by the type of deformity.

Conclusion: Congenital hand anomalies are not uncommon. Those that got to us received standard treatment as documented in the literature.

INTRODUCTION
Congenital hand anomalies are hand deformities that are present at birth. These conditions are common but are relatively minor. Some are severe and can be a disabling challenge when the child starts to interact with the environment. They are also a source of apprehension to the parents requiring early consultation with a hand surgeon. [1, 2, 3] These anomalies constitute a significant work load in plastic surgery practice. There is however a dearth of information on these anomalies in our environment. This study was designed to investigate the pattern and management of congenital hand anomalies, determine prenatal factors associated with the condition and review the literature.

MATERIALS AND METHODS
The medical and operating theatre records of the hospital were examined. The case notes and photographs of patients that presented to the hospital with congenital hand anomalies between July, 2002 and June, 2012 were retrieved and reviewed. Information obtained included; biographic data, diagnoses, antenatal and family history, involved hand and digits, type of operative procedure and complications. Results obtained were analyzed using Statistical Package for Social Sciences (SPSS) version 17 and presented in tables and charts.

RESULTS
A total of 43 patients presented to the hospital with congenital hand anomalies during the period under review. They were made up of 16 males (62.8%) and 27 females (37.2%) representing a male to female ratio of 1:1.69. The age at presentation ranged from 2 days to 26 years with a peak age group of 0 – 11 months when about 50% of the patients presented (Table. 1). Only about 19% presented within one month of birth, while more than 25% presented after the age of five years.
Table 1
Showing age at presentation

<table>
<thead>
<tr>
<th>AGE IN MONTHS</th>
<th>FREQUENCY</th>
<th>PERCENTAGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 – 11</td>
<td>21</td>
<td>48.8</td>
</tr>
<tr>
<td>12 - 23</td>
<td>7</td>
<td>16.3</td>
</tr>
<tr>
<td>24 - 35</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>36 – 47</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>48 - 59</td>
<td>2</td>
<td>4.7</td>
</tr>
<tr>
<td>≥ 60</td>
<td>11</td>
<td>25.6</td>
</tr>
<tr>
<td>Missing</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>Total</td>
<td>43</td>
<td>100</td>
</tr>
</tbody>
</table>

Many cases were bilateral, while in those that were unilateral, the left hand was slightly more frequently involved than the right (fig. 1).

Figure 1
Bar chart showing the distribution of involved hand

The mean paternal age was 35 ± 7.9 while mean maternal age was 31 ± 8.5. There was a history of hand anomalies in first degree relatives in 14% of cases while 79% had negative family history. Information on family history of hand anomalies was missing in 3% of cases.

Syndactyly was the most common type of anomalies observed constituting 37.2% in isolated forms and 25.7% in association with other anomalies and syndromes. This was followed by polydactyly which presented in isolated form in 25.7% of cases and in association with syndactyly in 4.7% of cases. Syndromic forms constituted 14% of cases seen.

Table 2
Type of anomalies

<table>
<thead>
<tr>
<th>DIAGNOSIS</th>
<th>FREQUENCY</th>
<th>PERCENTAGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Syndactyly</td>
<td>16</td>
<td>37.2</td>
</tr>
<tr>
<td>Polydactyly</td>
<td>11</td>
<td>25.6</td>
</tr>
<tr>
<td>Syndromes</td>
<td>6</td>
<td>14</td>
</tr>
<tr>
<td>Syndactyly with constriction band</td>
<td>3</td>
<td>7</td>
</tr>
<tr>
<td>Syndactyly with polydactyly</td>
<td>2</td>
<td>4.7</td>
</tr>
<tr>
<td>Macroactyly</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>Hypoplasia</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>Constriction bands</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>Cleft hand with syndactyly</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>Atypical cleft hand</td>
<td>1</td>
<td>2.3</td>
</tr>
<tr>
<td>TOTAL</td>
<td>43</td>
<td>100</td>
</tr>
</tbody>
</table>

Figure 2
Atypical cleft hand
Figure 3
Cleft hand with syndactyly

Figure 4
Congenital constriction band

Figure 5
Macrodactyly

Figure 6
Postaxial polydactyly
Most mothers (55%) had uneventful pregnancy while a reasonable percentage had health challenges during early pregnancy. The most prominent among these was febrile illness but the possible aetiologies of the febrile illness were not documented. Ingestion of native medication also featured prominently in the antenatal histories (Table 3).

Those patients that presented early and required surgery had surgery between six months and two years of age. Those that presented late had surgery as soon as they were fit for surgery. Most patients (93%) had various surgical procedures determined by the type of deformity. Interdigitating zigzag incisions with flaps for the web space and full thickness graft for residual defect were the most common surgical procedures carried out for the release of syndactyly. Excisions were used for polydactyly while multiple Z plasties were used to release constriction bands.

Postoperatively, 28% of patients had some complications while 72% of patients were free from complications. 16.3% had wound breakdown while 4.7% had gangrene (Table 4). The mean follow up period was $10 \pm 26.9$ weeks with a range of 1 to 156 weeks. However, 18.8% of patients were lost to follow up immediately after discharge and additional 48.8% after three weeks of follow up.
DISCUSSION

Congenital hand anomalies have an estimated incidence of 2.3 per 1000 live births. [4] They are a genetically and clinically heterogeneous group of disorders that may present in isolation, in combination or as a syndrome. Individually they are rare but collectively, and due to severity they are of clinical relevance. [5] The relatively low number of cases recorded in this series is probably due to poor medical records. In addition, minor malformations are more common than major malformations, [1,6] and as such many may not get to the plastic or hand surgeon. This may explain the greater incidence of syndactyly over polydactyly in our series. Simple polydactyly frequently is tied off at birth by the midwife and left to drop off.

Though it has been documented that congenital hand anomalies are associated with parental anxiety,[2, 3] late presentation was a common finding in this series as only 19% of patients presented within one month of age while more than 25% of patients presented after five years of age. This is similar to findings by Adewole et al, [7] in a study of congenital club foot in a Teaching Hospital in Lagos, Nigeria. They noted that only 29% of patients presented in the first month of life. It has been noted that the reasons adduced for late presentation include poverty, ignorance as well as activities of unorthodox practitioners. It may also be from late response to referrals to our centre for financial reasons.

There is no sex predilection in congenital anomalies of the hand as male to female ratios vary from study to study. Goldfarb et al,[8] in a study of the epidemiology of congenital upper limb anomalies in a Midwest United States population; an assessment using the OMT classification found that 51% were males while 49% were females representing an approximate ratio of 1:1. Jain et al, [9] in their study of the profile of congenital transverse limb deficiencies noted male to female ratio of 3.5:1. The male to female ratio in this study was 1:1.69. However, some specific congenital hand anomalies such as syndactyly have sex predilection as it is twice more common in males than in females. [6,10,11]

Congenital hand abnormalities may be caused by genetic, non-genetic or environmental factors disturbing the regular developmental program during embryogenesis. Multiple genes are involved in the control of growth, polarization as well as skeletal development of the hand. Homeobox (hox) genes in the 5’ region of chromosomes 2 and 7 are involved in the determination of shape and identity of skeletal elements of the limb. Sonic hedgehog (Shh) has been identified as the molecule responsible for the morphogenetic properties of the zone of polarizing activities (ZPA) that controls the anterior-posterior patterning of the limb. Similarly Wnt 7a and Lmx1 genes controls the dorsal identity whereas the En1 gene is responsible for the ventral identity of the hand. Malformations occur as a result of abnormal intrinsic embryonic development due to genetic defects. Such defects can be inherited, or result from exposure to teratogens or maternal diseases. It is of note that 14% of patients in this series have positive family history of congenital hand anomalies.

Hyperthermia was the first teratogen in animals that was subsequently proven to be teratogenic in humans. [12] Hyperthermia refers to elevated body temperature and has many causes including febrile infections. About 26% of mothers in our series had febrile illness during pregnancy. Though the aetiologies of the febrile illness were not documented, it is known that malaria is a very common cause. It is estimated that 40% of the world’s pregnant women are exposed to malaria and about 45% of pregnant women who did not receive intermittent preventive treatment had clinical malaria in a study done in Ado-Ekiti, Nigeria. [13]

One of the mechanisms of heat damage to embryo is vascular damage resulting in microvascular insufficiency due to endothelial damage with leakage, perivascular and interstitial oedema. There is resultant haemorrhage and hypoplasia in the limbs and digits, as well as other defects.[12] Vascular damage may also interfere with normal equilibrium of the preset program of mitosis and apoptosis within the mesenchymal cells of the inter-digital spaces.[14]
Increased mitoses instead of apoptosis is observed and result in failure of separation.

Another possible teratogen is the use of herbal drugs. According to Singh S. as cited by Goel RK et al,[15] there are four criteria for a drug to be called a teratogen: I) Foetal resorption or death, II) Stunting in size or growth retardation. III) Malformation – gross or microscopic. IV) Functional disorder or behavioral changes. Some herbal extracts have been shown to contain oestrogen-like substances and steroidal glycosides among others substances, [15, 16] that may account for any of above effects.

Generally, the use of herbal products as dietary supplement or “neutraceuticals” is rapidly increasing. [16] These herbs may be used when the woman does not even know she is pregnant which is also the period when the fetus is more sensitive to teratogens, 3 – 8 weeks.[17] It is of note that about 9% of mothers in this series used herbal medication during pregnancy. Similarly, in a survey in Benin, Nigeria, Gharoro,[18] found that 12% of women attending ante natal clinic use herbal drugs. The percentage may be higher if those in rural areas are considered. Though firm conclusions are difficult to make, available evidence implies that use of herbal products during pregnancy is associated with some risks. [16] Establishing direct causal relationship between the use of herbs and congenital hand anomalies requires more studies.

Congenital hand anomalies were grouped into seven based on the aetiopathogenic classification propounded by Swanson et al in 1976 which was modified and adopted by the International Federation of the Societies for the Surgery of the Hand (IFSSH) in 1983. [19] These are: I) Failure of formation; a) Transverse, b) Longitudinal. II) Failure of differentiation. III) Polydactyl; a) Pre axial, b) Post axial, c) Central. IV) Overgrowth. V) Undergrowth. VI) Constriction band syndrome. VII) Generalized skeletal abnormalities. Though this classification attempts to group anomalies on the bases of similarity in aetiopathogenesis, closer analysis will reveal some heterogeneity even within groups. Based on the perceived challenges with this classification, several modifications or even new classifications have been proposed. Ogino, [20] proposed a modification which was adopted by the Japanese society for the surgery of the Hand and named Japanese modification of IFSSH classification. Similarly, Chung, [21] proposed his modification on the bases of which he discussed the principles of management. Another new classification by Oberg, Manske and Tonkin (OMT) has been used in some epidemiologic studies, [8, 22] and was found to be useful though not without flaws. The IFSSH scientific committee on congenital condition has in 2014,[23] recommended the adoption of the OMT system of classification which they believe has the ability to be flexible and respond to newer developments and additional conditions with a recommendation for reviews at 3 years interval. However, the Swanson’s classification was used in this study as it was the bases of diagnoses in the cases being reviewed. It is also simple, internationally accepted and most widely used classification. Besides, in the everyday management of congenital hand anomalies, each case needs to be analysed in its own right, the anomaly described and treatment planned on the bases of this analysis, [24] as no classification system is as yet perfect.

The first group of anomalies i.e. failure of formation include transverse and longitudinal arrest. Transverse arrest commonly occurs at the level of the proximal forearm, wrist, metacarpal and the phalanges. Longitudinal arrest includes phocomelia, radial/ulnar club hand and cleft hand. Cleft hand was the only anomaly in this group that was recorded in this study. These constituted 4.6% out of which half was typical and the other half atypical. Congenital cleft hand is a rare hand anomaly and was historically divided into typical and atypical. While typical cleft hand show absence of parts in the centre of the hand and dominant inheritance pattern, atypical cleft hand is now recognized as symbrachydactyly as it is a whole range of degrees of absence and hypoplasia, and is typically sporadic in occurrence.[24,25,26] In this study, typical cleft hand was associated with positive family history of cleft hand.

The second category of anomalies is failure of differentiation which includes: syndactyly, synphalangism, synostosis, radial head dislocation and contracture. Contracture can be soft tissue as in camptodactyly or skeletal as in clinodactyly. Syndactyly is the only anomaly in this group that was recorded and was the most common congenital hand anomaly in this study. Similarly syndactyly has been documented by many authors as the most common congenital hand anomaly.[1,11, 27] Typically syndactyly occur in isolated form but can also occur in association with other anomalies like polydactyly or syndromes such Apert’s which is the most common acrocephalosyndactyly, Poland’s syndrome, Holt Oram’s etc.[10, 11, 25, 28] In this study, syndactyly was mostly an isolated finding representing 25.6% while it occurred in association with polydactyly in 4.7 %, constriction band in 7% and Apert’s syndrome in
Polydactyly was a common finding in this study and was second only to syndactyly. While this is in keeping with what some authors documented, [1, 11, 27] others find polydactyly as the most common congenital hand anomaly. [29, 30] Syndactyly appear to be the commonest congenital hand anomaly in hospital based studies such as ours while polydactyly is commoner in population based studies. Ekblom [22] in a thesis reviewing the studies of epidemiology of congenital hand anomalies found in the population based study, that polydactyly was much more common than syndactyly. Congenital trigger thumb was the commonest in that study. Many ulnar polydactyly are minor and may be tied off in new born nurseries, [30, 31] and hence may not present to specialized centres.

Macroductyly is a rare congenital hand anomaly that represents an overgrowth of all the structures of the involved digit and is different from isolated enlargement of bone or vessels. [32, 33] We recorded only 2.3% in this study reflecting the rarity of the condition. It usually occurs in isolation but can occur with neurofibromatosis or Klippel-Trenaunay-Weber syndrome. It has been described as either static where it is present at birth and grows proportionately with other body tissues or progressive where it occurs in early childhood and grows progressively till skeletal maturity.

Category V is undergrowth or hypoplasia. It represents incomplete or under development of the fingers or the thumb. [1, 20, 34] Some authors consider it as a form of category I [9,20, 22] It is rare, commonly affecting the thumb and is about 3.5% of congenital upper limb malformations. [34, 35] In this study, we recorded 2.3%.

The sixth category of anomalies is congenital constriction band syndrome. The syndrome constitutes a heterogeneous group of disorders including constriction of extremities, oligodactyly, acrosyndactyly, cleft lip and palate, tallipes equinovarus and abdominal wall defects with multiple varied names used to describe it. These include amniotic band syndrome, ring constriction syndrome, Streeter’s dysplasia, annular groove, amniotic rupture sequence and, amniotic deformity, adhesion and mutilation (ADAM) sequence. Most of these terminologies typically relate to the extremity manifestations, [36,37] and reflect lack of consensus on the aetiology. The term congenital constriction band syndrome is most appropriate in view of current understanding of the pathophysiology of the condition. [38] A number of theories have been made regarding the aetiology of this syndrome each explaining some but not all the manifestations. The Streeter’s intrinsic theory of germline developmental abnormality being responsible for the syndrome often explains major craniofacial, body wall and internal organ malformation while the Tropin’s extrinsic theory of amniotic sac rupture leading to formation of chorioamniotic mesodermal bands explains extremity anomalies. [36] Only extremity anomalies were recorded in this study and represented 9.3% of all the anomalies. 2.3% was type I (simple) while 7% was type II (associated with acrosyndactyly).

Treatment of upper extremity congenital differences can be challenging but rewarding and should be initiated as soon as possible. Patients should be thoroughly evaluated to rule out associated anomalies such as VACTERL associations. Where present, appropriate consult or referral should be done. Most surgeons wait till the child is about six months when anaesthesia is considered safe and tissues can better be handled. Though there is no definite upper age limit for surgery, most anomalies tend to change with time. For instance, in syndactyly affecting border digits, delay in release may result in deformity relating to forced flexion, angulation, and/or rotation due differential growth between the digits. In addition, the human body is considered to maintain skeletal remodeling power until two to three years of age and it is also important to complete all major reconstructive procedures before school age. [2, 11, 21, 28, 39] In this study, those patients that presented early and required surgery had surgery between six months and two years of age. Those that presented late had surgery as soon as they were fit for surgery.

The aim of treatment is to achieve maximum functional recovery while bearing in mind the importance of appearance. The type of surgery performed is determined by type of anomaly and could be staged. In syndactyly release, we used zigzag incisions with interdigitating flaps for release while triangular or rectangular dorsal skin flaps were used to reconstruct the web spaces. Residual raw areas were covered with full thickness skin grafts. Most surgeons use similar approach though there have been reports of use of split skin graft and tissue expansion for residual raw areas but these methods did not gain popularity due to complications. [11, 24, 27, 28, 39]

Treatment of polydactyly depends on type. Postaxial polydactyly type B can safely be suture ligated in new born nurseries, allowed to turn gangrenous and fall off. [33, 35]
This may be associated with complications such as bleeding and infection. [35] though there are no studies to support this. [31] Watson et al, [31] in a study of the prevalence and treatment of post axial polydactyly type B found that suture ligation was used in twenty-one patients with thirty-seven post axial type B polydactylies with no report of infection or bleeding. Similarly, Maree MN, [40] reviewed the use of vascular clip ligation and excision of type B postaxial polydactyly as done at Red Cross Children’s Hospital, Cape town, South Africa and reported no bleeding or infection. Large postaxial polydactyly (type A), preaxial and central polydactyly require operative ablation with reconstruction of important functional parts. [24, 33, 39] Our patients with polydactyly had operative treatment that involved excision of the extra digit and in some cases reconstruction of collateral ligaments.

In treating constriction band syndrome we used staged excision of the band and multiple Z-plasty reconstruction on one side of the circumference of the involved digit and the other half treated similarly later. This is the method advocated by many authors, [24, 38, 39] as it is believed to give better outcome and avoid scar contraction that will result in recurrence. However, Choulikain et al [41] described the use of straight line closure with improved scar and superior cosmetic result, a technique they noted had been in use for decades in their hospital though the number patients treated with the method was not indicated. Similarly, Larkin M. [42] reported a study done by Esien DB and colleagues which showed that linear scars are more aesthetically pleasing to the lay public than zig zag scars. The use of straight line closure may therefore be worth considering.

CONCLUSION

Congenital hand anomalies are not uncommon in this environment but not all cases get to the plastic surgeons. Those that do get standard treatment as documented in the literature. Further prospective studies are required to eliminate limitations imposed by inadequate medical records, determine the exact causes of febrile illness and its effect and the causal relationship between native medication and congenital hand anomalies.

References


42. Larkin M. Linear scars more aesthetically pleasing than zigzag scars. Medscape.2016.
Author Information

Uwakwe C. Mba, FMCS, Consultant Plastic Surgeon
Department of Surgery, ESUT Teaching Hospital
Enugu, Nigeria

Iheuko S. Ogbonnaya, FWACS, Chief Consultant Plastic Surgeon
Department of Plastic Surgery, National Orthopaedic Hospital
Enugu, Nigeria

Igwilo I. Onah, FWACS, Chief Consultant Plastic Surgeon
Department of Plastic Surgery, National Orthopaedic Hospital
Enugu, Nigeria