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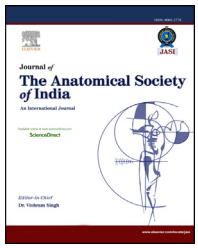

Original Article

Congenital upper limb anomaly as a cause of physical handicap

Sharmistha Biswas [a,*], *Panchanan Kundu* [b], *Rudradev Meyur* [c], *Anjan Adhikari* [d], *Gopal Chandra Mondal* [a]

[a] *Associate Professor, Department of Anatomy, N.R.S. Medical College, Kolkata, West Bengal, India*
[b] *Prof., Medical Superintendant, Vice Principal, Bankura Sammilani Medical College, Bankura, West Bengal, India*
[c] *Associate Professor, Department of Anatomy, R.G. Kar Medical College, Kolkata, West Bengal, India*
[d] *Associate Professor, Department of Pharmacology, R.G. Kar Medical College, Kolkata, West Bengal, India*

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ABSTRACT

Introduction: Congenital anomalies are most common causes of handicap in developing and developed countries. There are many approaches to classify congenital limb defects, especially upper limb.

Our study aimed to present a profile of the cases of handicap due to congenital anomalies of upper extremity encountered in B. S. Medical College & Hospital, Bankura, West Bengal. This may be helpful to the practicing orthopedic surgeons to assess and treat congenital anomalies of the upper extremity.

Methods: All the cases of congenital orthopedic anomalies affecting the upper limbs who attended the B. S. Medical College & Hospital in Bankura, West Bengal, for the purpose of obtaining physically handicapped certificate during a period of 1 year were included as subjects of the present study. All cases were subjected to clinical and radiological examination. Complete history was taken.

Results: Different types of deformities noted in our series, like transverse arrests at different levels, longitudinal arrests of preaxial variety, central longitudinal arrest (cleft hand) and intercalated longitudinal arrest or phocomelia. Overgrowth (macrodactyly), undergrowth (radial hypoplasia) both was noted. There was a case of constriction band syndrome.

Discussions: This study comprised of cases who attended hospital to obtain physical handicap certificate only and none had any therapeutic intervention. There was no case of total handicap. Incidence of congenital upper limb anomalies was higher in this series. More males were affected than females; there were more right sided defects than left. No definitive causes for the deformities could be isolated, chromosomal abnormality studies might have revealed etiology.

* *Corresponding author*. BJ 145, Sector II, Salt Lake, Kolkata 700091, West Bengal, India. Tel.: +91 9903408977.
E-mail addresses: drsharmisthabiswas@rediffmail.com, drsarmisthabiswas@gmail.com (S. Biswas).

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1. Introduction

Congenital anomalies are abnormalities of structures of body parts arising at the time of conception, or during intra-uterine period. They are the most common causes of handicap in developing and developed countries.

Handicap is the disability which adversely affect normal growth, development & adjustment of life for a substantial period of life, if not permanently.

Congenital anomalies affect 1%–2% of new-borns, and approximately 10% of those children have upper extremity abnormalities.[1,2]

Limb malformations can be categorized into 3 major groups:

1) A genetically-determined group, 2) An environmentally-induced group, 3) A multifactorial group.

There are many approaches to classify Congenital Limb Defects, especially upper limb. The most widely accepted classification of congenital limb anomalies was proposed by Frantz and O'Rahilly (1961)[3] and presented by Swanson (1968).[4] This work eliminated much of the confusing Greek and Latin terminology and has been accepted by the American Society for Surgery of the Hand (ASSH) and the International Federation of Societies for Surgery of the Hand (IFSSH) [1983], and the International Society for Prosthetics and Orthotics. This system defines the anomalies according to the embryonic failure during development and relies on the clinical diagnosis for categorization. Each limb malformation is classified according to the most predominant anomaly and is placed into one of seven categories, viz:

1.1. *Type I – Failure of formation*

1) Transverse arrest – Can be at any level, shoulder to phalanx
2) Longitudinal arrest – Preaxial – Varying degrees of hypoplasia of the thumb or radius Central – Divided into typical and atypical types of cleft hand Postaxial – Varying degrees of ulnar hypoplasia to hypothenar hypoplasia
3) Intercalated longitudinal arrest – Various types of phocomelia

1.2. *Type II – Failure of differentiation*

1) Soft tissue – Syndactyly, trigger thumb, poland syndrome, camptodactyly
2) Skeletal – Various synostoses and carpal coalitions
3) Tumorous conditions – Include all vascular and neurologic malformations

1.3. *Type III – Duplication*

May apply to whole limb, mirror hand, polydactyly.

1.4. *Type IV – Overgrowth*

Includes conditions such as hemihypertrophy and macrodactyly.

1.5. *Type V – Undergrowth*

Most commonly, radial hypoplasia, brachysyndactyly, or brachydactyly.

1.6. *Type VI – Constriction band syndromes*

Occurs with or without distal lymphedema; may involve amputation at any level.

1.7. *Type VII – Generalized anomalies and syndromes*

Ogino (1997)[5] classified upper limb anomalies into

i) Longitudinal deficiency – radial, ulnar, central deficiency or cleft hand
ii) Transverse deficiency
iii) Constriction band syndrome

The clinician must possess a basic understanding of embryogenesis, limb formation, and inheritance patterns to relay relevant knowledge to the family. Certain upper extremity anomalies occur in isolation, whereas others are associated with systemic conditions. These associated disorders often take precedence over the limb anomaly and must be assessed with appropriate diagnostic testing.[6]

Our study aimed to present a profile of the cases of handicap due to congenital anomalies of upper extremity encountered in Bankura Sammilani Medical College & Hospital, Bankura, West Bengal. This may be helpful to the practicing orthopedic surgeons to assess and treat congenital anomalies of the upper extremity.

2. Methods

All the cases of congenital orthopedic anomalies affecting the upper limbs who attended the Bankura Sammilani Medical College & Hospital in Bankura, West Bengal, for the purpose of obtaining physically handicapped certificate during a period of 1 year (between August, 2008 and July, 2009) were included as subjects of the present study. The study population were residents of western parts of West Bengal and were either originally from western West Bengal or migrants from parts of surrounding states of Jharkhand and Odisha.

Clearance from Institutional Ethical Committee was taken and informed consent of the patients was taken before carrying out the study and taking photographs.

All cases were subjected to a thorough clinical and radiological examination. Complete history like maternal & paternal age, occupation, drug history, disease, injury, exposure to radiation, consanguinity, etc. and personal and family histories were taken. Chromosomal abnormalities were not searched for.

3. Results

In our study, we observed that 430 cases attended B.S.M. College to receive Handicapped certificate. Of them, 291 cases

suffered from congenital anomalies. 37 (8.6%) of these 291 patients were orthopedically handicapped. Out of these 37 cases of congenital orthopedic handicaps, 14 cases were suffering from upper limb anomalies (37.8%). Out of these cases, 9 were male and 5 were female. 6 persons suffered from congenital anomaly of right upper limb, 4 had left sided anomaly and 4 had anomalies of both sides.

Types of deformities noted in our series were the following:

Transverse arrests at the level of metacarpals were present in 3 cases [Fig. 1]; at the level of carpals in 1 case; and at the level of forearm in 1 case.

Longitudinal arrest of preaxial variety – were noted in 3 cases, of them, hypoplasia of the radius was seen in 1 case and absence of radius were found in 2 cases. Radiology of one of these cases shows aplasia of radius, absence of first metacarpal and phalanges of the thumb and radially placed carpal bones and short, hypoplastic and abnormally curved ulna [Fig. 2].

There was central longitudinal arrest (cleft hand) in 1 case [Fig. 3]. We did not come across any postaxial variety of longitudinal arrest. There was one case of intercalated longitudinal arrest or phocomelia.

We found **overgrowth** in the form of macrodactyly in 2 cases, radial hypoplasia (**undergrowth**) was noted in one case.

There was one case of **Constriction band syndrome** in our series [Fig. 4].

One case of bilateral radial aplasia had a sibling having similar defect. Parents of one case of transverse arrest at the level of metacarpals had consanguineous marriage.

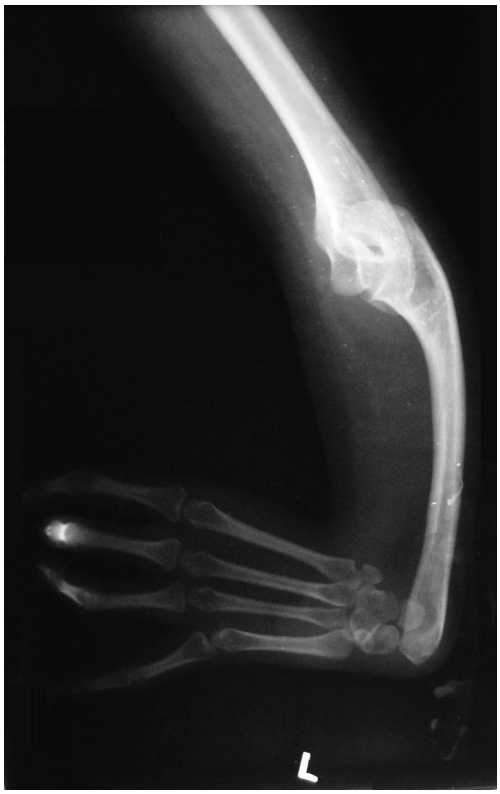

Fig. 2 – X-ray of a case revealed aplasia of radius, absence of first metacarpal and phalanges of the thumb and radially placed carpal bones. Ulna was short, hypoplastic and abnormally curved.

4. Discussions

In our study the incidence of congenital upper limb anomalies is 14 in 430 cases who attended B.S.M. College to receive Handicapped certificate, i.e. 3.2%. This is definitely much higher than previous reports. From the birth census data in Canada and UK, the estimated incidence of congenital upper limb deficiencies was found to be approximately 1:4200 live births. In a series studied by Conway (1956)[7] incidences of upper limb anomalies were 1 in 626 (0.14%). Flatt A.E. (1994)[1] observed that this incidence was 0.16–0.18% in the United States. Since our subjects were chosen from persons attending B.S. Medical College and Hospital for obtaining physically handicapped certificate, incidence of upper limb anomalies might have been higher. According to Bolitho (2006),[8] incidence of limb anomalies exhibit regional and ethnic differences. This also might be a reason of higher incidence of upper limb anomalies in our series, whose ethnicity differ from those of European and white races.

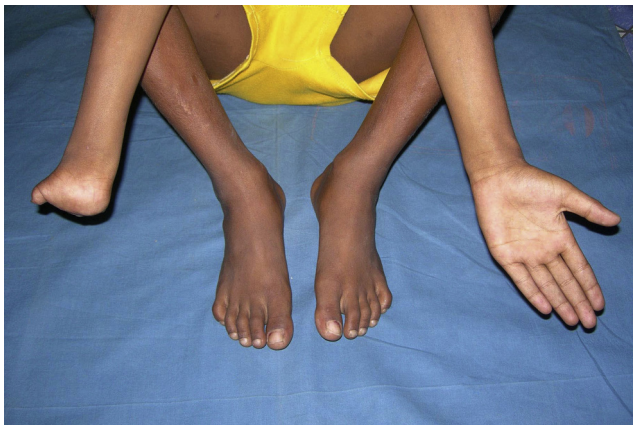

Fig. 1 – Transverse deficiency (Right) – at the level of metacarpals.

Present series revealed that congenital upper limb anomalies were present in more males than females (9 males and 5 females). This was in accordance with previous reports stating such anomalies were more common in males.[9,10]

Our study revealed more right sided upper limb defects than left (6 right sided, 4 left sided and 4 bilateral). This is unlike Canadian Congenital Anomalies Surveillance System (CCASS) data which indicate that congenital anomalies were reported more frequently on the left side of the body. Data from the Alberta Children's Hospital confirmed this left sided bias (McDonell, 1988).[11]

In the present series of cases of congenital upper limb anomalies, no significant prenatal, perinatal, and family histories were found. No histories of exposure to known teratogenic agents, drugs, or maternal diseases were present. One case of bilateral radial aplasia had a sibling having similar defect. Probably it was an autosomal recessive disorder. Parents of one case of transverse arrest at the level of

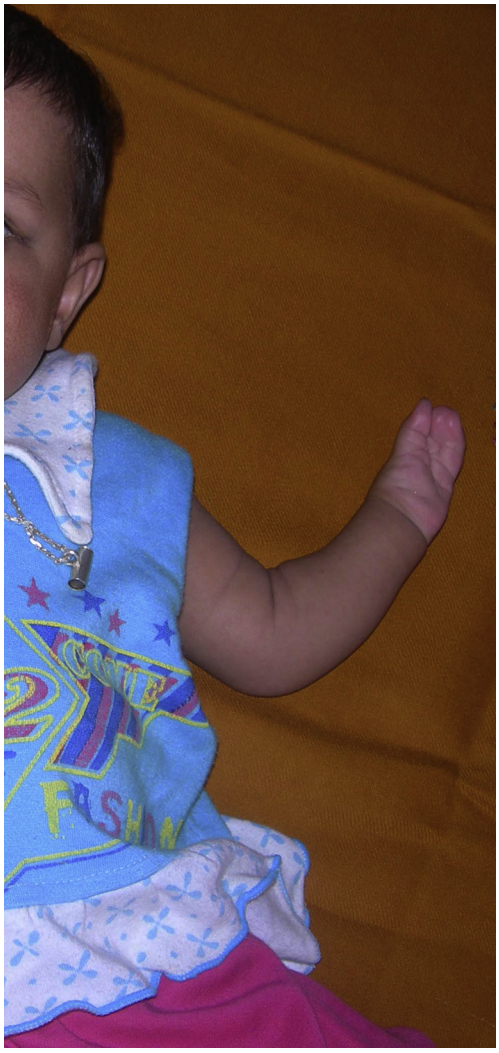

Fig. 3 – Central longitudinal arrest – Cleft hand (left).

metacarpals had consanguineous marriage. Consanguineous marriage may be another important risk factor.[12]

So, in our series definitive causes for the deformities could not be isolated. This observation was in accordance to those of Lamb & Scott (1981),[13] who found that congenital upper limb anomalies had no familial involvement and they were definitely not inherited. The majority of limb defects, especially those affecting one limb only, are sporadic.[14]

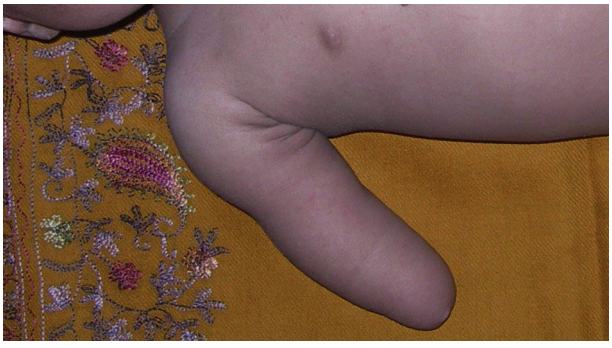

Fig. 4 – Constriction band syndrome.

Surprisingly, many upper extremity malformations cause little functional deficit. Children develop prehension with hands as they are.[1] The hand surgeon must offer surgery to improve the child's function and cosmetic value. Early surgery performed within the first 2 years of life is recommended. Parents should be counseled about what is possible and what not with surgery.

4.1. *Emgryology*

Limb development takes place during 3rd–8th weeks of gestation. Limb buds are formed by condensations of mesenchyme covered with a thick layer of ectoderm called Apical ectodermal ridge (AER). The underlying mesoderm is organized into a zone of polarizing activity (ZPA), which is located posteriorly and a progress zone (PZ) located more anteriorly. Vessels and then nerves subsequently grow into the limb. Then mesodermal differentiation into cartilage and muscle begins to occur. The limb begins to pronate, the elbow flexes, and the hand is flexed and ulnarly deviated. Muscle formation is derived from a dorsal (extensor) and ventral (flexor) muscle blastoma. Mixed motor and sensory nerves enter the limb as a pioneer growth cone. The period of hand differentiation is short, taking place during the 4th–7th weeks.[13] Majority of congenital upper limb defects take place during this period of rapid limb development.[15] Research has proved that there is definite role of HOX genes in development of limb buds.[16] T-box genes, Tbx5 and Tbx4 might be involved in determining limb identity, i.e., upper or lower (Takeuchi et al, 1999).[17]

The causes of congenital anomalies are: i) genetic, ii) environmental, iii) unknown. A minority of congenital anomalies have a single major environmental or genetic cause. Three types of sequences might occur in morphogenesis:

- Malformation sequence: an intrinsic malformation exists in the embryo, resulting in certain other abnormalities (e.g., radial dysplasia).
- Deformation sequence: no intrinsic defect in embryo (e.g., constriction bands).
- Disruption sequence: the healthy embryo is subjected to tissue breakdown or injury (e.g. as in TORCH infection, thalidomide-caused deformities).

When the cause is unknown, the term malformation is preferred. Approximately half of cases with multiple anomalies fall into known syndromes.

5. Conclusions

This study comprised of cases who attended hospital to obtain a physical handicap certificate only and none had any therapeutic intervention. Almost all cases were eligible for the physical handicap certificates. There was no case of total handicap. Incidence of congenital upper limb anomalies was higher in this series. Congenital upper limb anomalies were present more in males than females, there were more right sided defects than left. No definitive causes for the deformities

could be isolated, chromosomal abnormality studies might have revealed etiology. In some cases, autosomal recessive disorder may be responsible, consanguineous marriage is also a risk factor for such defects.

Conflicts of interest

All authors have none to declare.

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