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Original Research Article

Clinical profile of congenital limb anomalies in neonates

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ABSTRACT

Background: Reported birth prevalence of congenital limb defects vary from country to country. Epidemiological studies permit the timely detection of trends in congenital limb anomalies and the associations with other birth defects. The objective of this study was to know the prevalence and the variety of congenital limb anomalies and their association with other anomalies.
Methods: A prospective observational study in which all newborns reported to the neonatal department were surveyed to find out the incidence, diversity and association of limb anomalies. Age of the mother, risk factors and other demographic data of the newborns was analyzed. Babies born less than 28 weeks were excluded from the study.
Results: The prevalence was 6.34 per thousand live births. The majority of the neonates were term babies and female. 28 % of the babies were born out of consanguineous marriage. The mean age of the mother was 25. Median parity was 1. 9.52 % of the babies had a history of anomaly in the sibling. 4.76 % had a history of still birth. The most common congenital anomaly was congenital talipo equino varus. The other congenital limb anomalies observed were polydactyly, syndactyly, bifid thumb and absence of the radius. There were multiple congenital anomalies in 28.57 % of the babies.
Conclusions: The study has given us an insight into the variety of congenital limb malformations and also into the possible etiological factors. A study done on a larger population would probably help us assess the incidence and pattern of occurrence of limb anomalies and help us counsel the parents.

Keywords: Congenital limb anomalies, Neonates, Risk factors

INTRODUCTION

Birth defects can be defined as structural or functional abnormalities, including metabolic disorders, which are present from birth. The term congenital disorder is considered to have the same meaning and two terms are used interchangeably.[1]

Congenital abnormalities are not uncommon among newborns and contribute to neonatal and infant morbidity and mortality. The prevalence and pattern of presentation vary from place to place. Many a time the exact etiology is unknown but genetic and environmental factors tend to be implicated.

Congenital limb malformations rank behind congenital heart disease as the most common birth defects observed in infants.[2] One in 506 newborns has congenital malformation of the upper limb.[3] These malformations can occur as isolated malformations, in combination with another hand and/or foot, or as part of a syndrome.

The etiology can be divided into environmental and genetic causes. Early intrauterine period (between the 3rd

and the 8th week of gestation) is the vital period of life for the normal development of organs.[4]

The field of congenital anomalies of the limbs has been inundated by many classifications. More than 50 classification schemes have been drafted over the past 150 years, each claiming some special merit not possessed by others.[5]

A workable classification should employ a simple, easily remembered and descriptive terminology. It should allow the recording of common clinical entities with minimal confusion yet permit the full categorization of complex cases. An ideal classification should be made by grouping malformations according to morphogenesis or cause.[6]

Various limb anomalies are recognized at birth. Some of these include metatarsus adductus, congenital talipes equino cavo varus (CTEV/club foot), polydactyly, syndactyly, congenital pseudarthrosis of the tibia and fibula, a constriction band of the legs and limb length discrepancies.[7]

Timely identification of these anomalies is of great importance for a Pediatrician. The early identification not only prevents further complications, it also ensures a better future for the newborn.

The aim of present study was to assess the prevalence of congenital limb anomalies among the newborns in one setting and recognize the risk factors associated for these abnormalities and to compare our results with other surveys conducted earlier. Analyzing the risk factors would help us prevent and care for these risk factors. With the improvement in ultrasonography techniques and prenatal diagnosis techniques it has become easier to study and detect various congenital anomalies and necessary action could be taken at the earliest.

METHODS

A prospective observational study was conducted in a neonatal unit at a tertiary care hospital in India after due ethical clearance was obtained from the institution. The study was conducted over a period of 10 months, from January 2016 to October 2016, a total of 3310 neonates were examined for the presence of congenital limb anomalies. Of these, 21 (0.63%) neonates with congenital limb anomalies were identified and examined further. Only the neonatal unit admissions were included in the study. Those neonates with gestational age less than 28 weeks were also excluded.

Details about the mother such as maternal age at delivery, obstetric history of the mother, supplements whether taken or not during pregnancy, complications during pregnancy and the type of delivery were recorded. History of limb anomalies in siblings was asked for. The fetal parameters studied included gender distribution, birth weight; the details of the limb anomalies present and the presence of other congenital anomalies were recorded.

The diagnosis of each condition was made after a thorough examination and investigations were conducted under the supervision of a senior pediatrician. The identified abnormalities were categorized. The results were analyzed using Statistical Package version 21 for Windows.

RESULTS

3310 neonates were studies over a period of 10 months. 21 neonates were found to have congenital limb anomalies. Ten (47.62%) males and 11 (52.38%) females were found to have congenital limb anomalies. The difference was insignificant.

Table 1: Demographic data of the study.

Demographic data	Neonates with congenital limb anomalies	Percentage
Sex distribution of neonates		
Male	10	47.61
Female	11	52.38
Birth weight		
<2.5 kg	14	66.67
>2.5 kg	7	33.33
Type of delivery		
Normal vaginal delivery	16	76.19
Lscs	5	23.8
Maternal age		
Upto 30	15	71.4
30 and above	6	28.57
Parity of the mother		
Primigravida	9	42.9
Multipara	12	57.14
Consanguinity	6	28.57

There was a great variation in the birth weight of the neonates with congenital limb anomalies. Lowest weight being 930 g and the highest being 3.8 kg, the mean birth weight was 2.28 kg. Six (28.57%) of the affected neonates were preterm babies.

14 (66.67%) neonates weighed <2.5 kg. The maternal age range was variable with the youngest mother being 20 year old and the eldest being 35 year old, the average was 25.52 years. 5 neonates (23.8%) were delivered by LSCS. Median parity was 1. 4.76 % had a history of still birth.

9 (42.9%) deliveries were of primi gravida mothers, 4 (19.04%) where the newborn was a second child, 4 (19.04%) newborns were the third child and 2 (0.09%) were born as the 4th child or more. Consanguineous marriage was observed in 6 (28.57%) of the mothers and

anomalies were observed in other siblings in 2 (0.09%) of the cases.

Supplements such as Iron, Folic acid and Calcium were taken by 14 (66.66%) of the mothers. The complications that were noted in the antenatal and the perinatal period have been detailed in the Table 2.

Table 2: Maternal factors associated with the congenital limb anomalies.

Complication	No. of mothers	Percentage
Hyperemesis	6	28.57
Radiation exposure	1	4.76
Pregnancy induced hypertension	5	23.8
Gestational diabetes mellitus	1	4.76
Oligohydramnios	2	9.52
Antepartum hemorrhage	5	23.8
History of still birth	1	4.76
Hypothyroidism	0	0

A total of 21 congenital limb anomalies were detected in the study group. Upper limb anomaly was noted in 6 (28.57%), lower limb in 10 (47.61%) and both upper limb and lower limb anomalies were found in 1 neonate (4.76%).

The most common limb anomaly detected was club foot, seen in 5 (23.8%) of the neonates. The other identified anomalies included Syndactyly, polydactyly, absence defects, brachydactyly and bifid thumb. Four of the neonates (19.04%) had more than one limb anomaly.

Table 3: Major categories of congenital limb anomalies.

Anomaly detected	Number of neonates	Incidence per 1000 live birth
Club foot	5	1.51
Syndactyly	1	0.3
Polydactyly	4	1.2
Absence defect	2	0.6
Brachydactyly	2	0.6
Bifid thumb	1	0.3
More than limb anomaly	4	1.2

Out of the 21 neonates, one neonate was diagnosed to have Apert's syndrome and another with VACTERL syndrome.

DISCUSSION

The pattern and prevalence of congenital anomalies may vary over time or with geographical location, reflecting on the complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables.[8]

Figure 1: Upper limb hypoplasia.

With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India.[9] The incidence of congenital disorders at birth varies from one territory to another. European Surveillance of Congenital Anomalies has reported a prevalence of 3.8 limb defects per 1000 births.[10] A recent European study reported a prevalence of congenital limb defects as 21.1/10,000.[11] Other studies have reported different incidence of congenital malformations, Perveen and Tyyab reported a prevalence of 11.4/1000 births.[12]

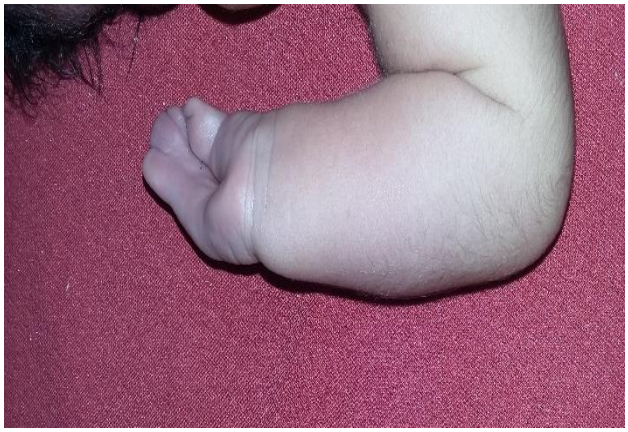

Figure 2: Mitten hand-in Apert's syndrome.

In present study, the overall prevalence found was 6.7 per thousand live births. This is higher compared to other studies from India, which have reported an incidence of 1.9%, 2.72%, and 2.22%.[13-15] Tertiary care hospitals usually do not have a definite catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this study cannot be projected to the total population. A community based study should be ideally conducted for true estimation of incidence of congenital anomalies in a population. Most other studies showed a higher incidence of anomalies in

males in contrast to present study.[14,15] The higher incidence of malformations with birth weight <2.5 kg in our study is similar to some other studies while some studies have reported no relation to low birth weight.[15-18]

Figure 3: Facial features in a neonate with Apert's syndrome.

Bai S et al reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta et al documented statistically insignificant association of increased maternal age and congenital anomalies.[19,20] The relationship between maternal age and babies born with congenital malformations, in our study, revealed that a majority of malformed babies were born of mothers aged 20-29 years; though, it was statistically insignificant.

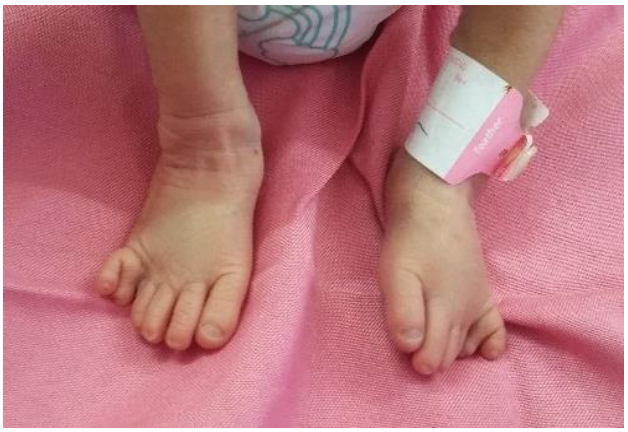

Figure 4: A neonate with bilateral post axial polydactyly.

Other studies have reported significantly higher incidence of malformations among the multiparas.[9] Present result is consistent with this finding, which indicates a positive correlation between the birth order and the incidence of congenital anomalies. Consanguineous marriages are reported to play a major role in the occurrence of congenital malformations.[21] In the present study also, prevalence of malformed babies was more when born out of consanguineous marriages as seen in studies from Kuwait, Arab.[22,23]

Despite the high risk of recurrence of congenital malformations, there are no well-accepted preventive measures in developing countries like India. It indicates that strong preventive measures for congenital anomalies are needed. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies. The congenital limb anomalies should be identified early as corrective measures or surgery when undertaken early can reduce the morbidity and improve the prognosis.

Limitation of the present study was firstly as it is a tertiary care hospital or referral centre, prevalence calculated may be higher than the general population in this hospital-based study hence, the data cannot be projected to the general population, for which population-based studies are necessary. Secondly, we could not include the abortions and stillborns, because often the abnormalities are not obvious or visible externally. In those cases, a pathological autopsy is warranted and in most of the cases, parental consent is not available for pathological autopsy. And third the study was conducted over a short duration of time; the data could be collected for a longer period of time to get a better idea about the morphoetiology of congenital limb anomalies.

CONCLUSION

In conclusion, the etiological study, including the genetic work-up, of morphologically classified cases with congenital limb anomalies is helpful in the prevention of many disorders causing limb defects. Prevention can be achieved via proper genetic counselling, which includes information on the etiology of the disorder, prenatal detection of limb anomalies and risk estimation for later siblings.

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