Original Article

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Incidence and Profile of Neonatal Musculoskeletal Birth Defects at a Tertiary Hospital in North East India

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Abstract

Background: Musculoskeletal birth defects happen when bone and muscle tissue develops abnormally in the newborn during fetal development. The resulting deformities are a burden to the family and associated with substantial morbidity and mortality.

Objective: To determine the incidence and clinical profile of the musculoskeletal defects present at birth in the newborn.

Materials and Methods: This is a single-center, prospective, observational study in a tertiary care hospital between September 2010 and August 2015 in North East India. All newborn admitted to neonatal care unit were screened for musculoskeletal birth defects. Newborn with congenital defects of other systems was excluded. Clinical examination, skeletal survey and laboratory work up were performed, and data were analyzed.

Results: A total of 3120 newborns were admitted during the study period. The overall incidence of the musculoskeletal birth defect was 13.46 per 1000 live births. 42 patients with a total of 93 variants of musculoskeletal birth defects, 46 major, and 47 minor, were analyzed. Male and Female ratio was 1.1:1. Multiple defects were detected in 57.14% with 35.71% involvement of the lower limb. Among the 46 major variants of the musculoskeletal defect, the most common was congenital talipes equinovarus (15.22%). The commonest risk factor associated was neonatal jaundice 15 (35.71%), and only 2 cases (4.76%) were admitted for multiple musculoskeletal defects. Out of 42 cases, 54.76% needed orthopedic consultation. The average duration of hospital stay was 6 days. 4 cases (9.5%) expired following medical condition.

Conclusion: Neonatal musculoskeletal birth defect is under reported though the incidence is not low as our finding show 13.46 per 1000 live births. These may be presenting as multiple or isolated musculoskeletal defect at birth with minor or major variants and needs orthopedic consultation to treat early and prevent long-term disability.

Key words: Birth defect, Disability, Early detection, Musculoskeletal, Prevention

INTRODUCTION

Globally, the congenital defects contribute to a large fraction of childhood morbidity and mortality. Birth defects can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.[1] The musculoskeletal birth defects cause long-term disability in the survivors as well as an economic burden to the families and society at large. There is poor information regarding the prevalence of musculoskeletal birth defects since many countries does not have national representative data. The causative mechanism of birth defect is heterogeneous, but genetic aberrations play a significant role.[2] The musculoskeletal birth defects may have a genetic, infectious, or environmental origin. Some deformities like multiple joint contractures may indicate an underlying serious neurologic malformation.

Antenatal screening and examination of newborn infants for musculoskeletal birth defects facilitates early detection, treatment, and care. Neonatal screening program should include musculoskeletal examination in addition to the

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existing system of screening. Early, referral to concern specialist and appropriate treatment of musculoskeletal birth defects can prevent disabilities and reduces permanent morbidities among the survivors. Considering the inadequacy of reporting system and lack of data regarding musculoskeletal birth defects, we take up this study to detect incidence and clinical profile in all the neonates admitted to our tertiary care institute.

MATERIALS AND METHODS

The present study is the single-center, prospective, observational study done in a tertiary care institute. All the newborns with musculoskeletal birth defect who were admitted to neonatal care unit during September 2010-August 2015 were enrolled after approval from the Institutional Ethical Committee. A written patient consent form was maintained and explained the outcome measures. All the children 0-28 days of age were eligible for the study after they were diagnosed clinically with musculoskeletal birth defects. Congenital malformations involving other systems were excluded from the study. The overall incidence, socio-demographic data, the reason for hospitalization, and duration of hospital stay were analyzed in addition to the clinical examination profiles. The musculoskeletal birth defects were categorized according to the site involves, severity in terms of major or minor and whether isolated or multiple were also recorded. The defects that had referral services for further management were identified. Data regarding maternal age, antenatal screening done, and neonatal risk factors were also recorded. Laboratory investigation, skeletal survey, and recording images were done as indicated. Follow-up plan of the cases was not included in the study. All the data were calculated and analyzed.

RESULTS

The total of 3120 newborns were admitted to neonatal care unit during the study period. 42 cases of musculoskeletal birth defects were selected for the study after fulfilling the inclusion criteria. The overall incidence of musculoskeletal birth defects was 13.46 per 1000 live births. 23 birth defects involving other major systems were excluded from the study. Among the study group, male were 22 (52.38%) and female were 20 (47.62%). Male to female ratio was 1.1:1. The highest number of admission belongs to age group below 24 h of life (52.38%). The majority of cases 30 (71.43%) belong to the lower middle socioeconomic class. 32 mothers were in the age group 25 to 35 years (76.19%). Antenatal sonography reports were recorded in 25 cases, and 2 cases were detected with moderate to severe oligohydramnios. Not a single mother had undergone antenatal screening with serum biochemical markers. None of the cases had a family history of the similar defect, and there was no history of co-sanguineous marriage among the parents. 6 cases (14.29%) were born to Primigravida. Only 3 cases (7.14%) were born before 37 weeks of gestation. Out of 42 cases, isolated musculoskeletal birth defects were detected in 18 cases (42.86%), and multiple defects were noted in 24 cases (57.14%) Among the multiple musculoskeletal birth defects, one case was diagnosed as arthrogryposis multiplex congenita (distal type) which is shown in Figure 1. Four cases had Downs's syndrome (9.5%). Figure 2 shows another case of multiple defects with the absence of right foot, the absence of 2^{th}, 3^{rd}, 4^{th} distal, and middle phalanges with cutaneous syndactaly of 2^{nd}, $3,^{rd}$ and 4^{th} proximal phalanges of both hands. Among the 42 cases, a total of 93 (2.2 per patient) variants of musculoskeletal birth defects were analyzed. 35.71% had involvement of lower limb birth defect. Out of 93 variants of musculoskeletal birth defects, the major

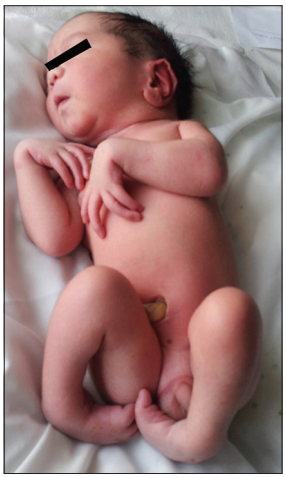

Figure 1: A child of arthrogryposis multiplex congenita

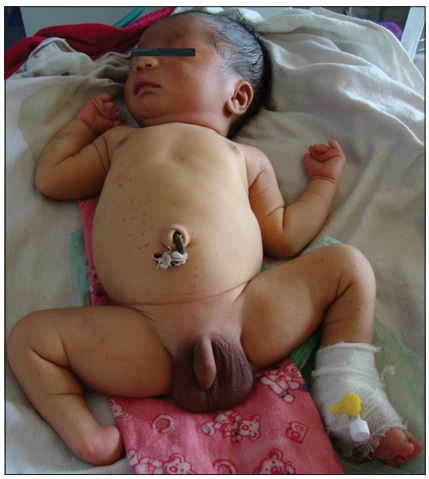

Figure 2: A child with absence of right foot and absence of 2^{nd}, 3^{rd}, 4^{th} distal and middle phalanges with cutaneous syndactaly of 2^{nd}, 3^{rd}, 4^{th} proximal phalanges of both hands

defect was 46 (49.46%), and the minor defect is 47 (50.54%) cases. The most common major musculoskeletal defect was congenital talipes equinovarus, 7 cases (15.22%). (Table 1)

Out of total 42 cases of musculoskeletal defects, 23 cases (54.76%) needed consultation with an orthopedic surgeon for the correction of deformity. The commonest risk factor for admission to neonatal care unit was neonatal jaundice (35.71%). Only 2 cases (4.76%) were admitted for multiple musculoskeletal birth defects. Table 2 The average duration of hospital stay was 6 days ranging from 6 h to 22 days. Out of 42 cases included in the study, 4 patients had expired (9.5%) resulting from associated medical conditions.

DISCUSSION

Musculoskeletal birth defects remain public health burden because most of them remain hidden. The congenital malformations are seen in 3% of all newborn.[3] The musculoskeletal defect is one of the common congenital malformations present at birth in a newborn. In the present study, the incidence of musculoskeletal birth defect is 13.46 per 1000 live birth. In India, the prevalence of birth defect is 64.3 per 1000 live birth according to the data given in Birth Defects prevalence estimates in the South- East Asia region, 2006.[4] Nationally representative data on overall prevalence of musculoskeletal birth defect needs to be studied as it contributes to significant proportion of neonatal and child morbidity and mortality. In most of the studies for birth defects in India, the predominant system involved was musculoskeletal system. In a study done by Muranjan and Vijayalakshmi, musculoskeletal birth defects among older children (0.08-11 years.) was found to be 16.3% which is quite high in comparison to the present study.[5] Suresh *et al.* studies show that with an estimated 25 million births every year, the absolute number of children born with birth defect in India would be in excess of 500,000.[6] In our study of 42 musculoskeletal birth defects, male children were more affected than female with male and female ratio of 1.1:1. The majority of cases (71.43%) belong to the lower middle-income group. In a study, about 94% of serious birth defects occur in middle and lower income countries.[1]

Antenatal diagnosis of birth defects is possible with antenatal sonography and biochemical screening. The facilities for both the diagnostic procedure are costly and not universally available. The same clinical sign or malformation may be cause by a variety of genetic defects in addition to the environmental causes. In the present study, none of the cases were detected by antenatal ultrasonography and biochemical enzyme studies were not done for all cases. Only 2 cases had oligohydramnios and one case with twin pregnancy. None of the mothers had a history of drug intake during the antenatal period. There was no similar musculoskeletal birth defect among the family members and siblings. Only 6 cases were first born child.

The present study also shows 24 cases of multiple musculoskeletal birth defects and 18 cases of isolated

Table 1: Variants of musculoskeletal birth defects

Major musculoskeletal birth defects	*n*=46 (%)	Minor musculoskeletal birth defects	*n*=47 (%)
Absence (2, 3, 4) distal and middle phalanges both hand	1 (2.17)	Bilateral anonychia of hand	1 (2.13)
Absent multiple ribs (left) 3rd to 9th	1 (2.17)	Bilateral hammer toe	3 (6.38)
Apodia (right lower limb)	1 (2.17)	Bilateral wide gaping of 1st and 2nd toe	9 (19.15)
Absent right thumb	1 (2.17)	Cleinodactyly	4 (8.51)
Cleft palate	3 (6.52)	Depress nasal bridge	10 (13.51)
Cleft lip	5 (10.87)	Low set ear	5 (10.63)
Congenital calcaneovalgus	1 (2.17)	Metatarsus adductus	4 (8.51)
CTEV	7 (15.22)	Overlapping of 1st and 2nd toe	2 (4.23)
Cutaneous syndactaly (2, 3) proximal phalanges of both feet	1 (2.17)	Rudimentary ear lobe (both)	2 (4.23)
Cutaneous syndactaly (2, 3, 4) proximal phalanges both hand	1 (2.17)	Short and broad finger of both hands	4 (8.51)
Flexion contracture of knee (both)	1 (2.17)	Sacral dimple	2 (4.23)
Flexion contracture of wrist (both)	2 (4.35)	Unilateral hammer toe 1	1 (2.13)
Genu recurvatum (both)	2 (4.35)		
Genu recurvatum (left)	1 (2.17)		
Hemi-vertebrae	2 (4.35)		
Hypermobility of the joint	6 (13.04)		
Imperforate (both) external auditory canal	1 (2.17)		
Macrocephaly	1 (2.17)		
Microcephaly	2 (4.35)		
Micrognathia	3 (6.52)		
Osseous polydactyly (L) thumb	1 (2.17)		
Osseous polydactyly (R) thumb	1 (2.17)		
Rudimentary left thumb	1 (2.17)		

CTEV: Congenital talipes equinovarus

Table 2: Reason for indoor admission in neonatal care unit

Condition	*n*=42 (%)
Multiple birth defect	2 (4.76)
Pre-term with low birth weight	3 (7.14)
Poor feeding with sepsis	6 (14.29)
Respiratory distress	6 (14.29)
Low Apgar score	10 (23.81)
Neonatal jaundice	15 (35.71)

musculoskeletal birth defect out of the total 42 cases. In a study done by Aase, the mechanism of isolated lesion is poorly understood but may include phenomena like somatic cell maturation, aberrant cell migration, deficient or excess cell division, and failure of cellular interactions.[3]

The combinations of birth defects appear to be random, but sometimes a specific pattern of defects can be recognized. Two cases of multiple musculoskeletal birth defects were also detected during our study period. One of the defects was distal arthrogryposis multiplex congenita. This could be due to environmental factors occurring in intrauterine life. The congenital contractures involving two or more body areas in extremities may be present as distal arthrogryposis. These congenital contractures tend to improve with age but do not completely disappear.[7] Another multiple defect was the absence of right foot (Apodia) with bilateral absence of middle and distal phalanges of 2,nd 3,rd and 4th and cutaneous syndactaly of 2,nd 3,rd and 4th proximal phalanges of both hands. In this case, only the limb defects were detected with no other systemic involvement. With proper prosthesis and other rehabilitation, baby may live a normal life. According to International Standard (ISO 8540 - 1: 1989) classification absence of foot including skeletal deficiency is a transverse tarsal and metatarsal total deficiency. It is a form of "congenital amputation" implying that a limb segment has been lost before birth which is partial failures of the formation.[8] The etiology could be genetic, teratogens, vascular disruptions, chemical, and radiation exposure.[9] In our study, the etiology of the defect could not be revealed.

From the present study, 93 variants of musculoskeletal birth defects were identified of which 46 (49.46%) major defects were detected and minor defects were 47 (50.54%). Out of total major variants, the most common defect was congenital talipes equinovarus, 7 cases (15.22%). Congenital talipes equinovarus is a deformity in which the foot is turned inwards to a varying degree. In the general population, the incidence of congenital talipes equinovarus (CTEV) is 1 in 1000 live births. There is a 1:800 chance of having this deformity; 1:3.5 chance in sibling and 1:3 chance in an identical twin.[10] In our study, the incidence of CTEV was 2.2 per 1000 live birth admitted to neonatal care unit which shows a higher incidence in a high-risk group of neonates. There are inherited neonatal orthopedic conditions like CTEV present at birth, which may be underdiagnosed.[11] An equinovarus deformity present at birth should be brought to the notice of an orthopedic specialist at the earliest.[12] All our cases were referred to an orthopedic specialist for immediate management.

We also detected a case of mild Calcaneo-valgus deformity in one of the twins, whereas the another twin was normal in all the four limbs. Due to crowding of the uterine environment there is a greater risk of limb deformities in multiple pregnancies. During antenatal period, twin to twin transfusion syndrome may occur in monochorionic twins due to the arteriovenous anastomosis. One of the complications in the donor twin includes limb deformities.[13]

Of the total study patients, 23 (54.76%) cases were needed an orthopedic consultation for further management. Referral to the specialist concern is required for all the musculoskeletal birth defects to start timely treatment and achieve reasonable functionality. Treatment should be started as soon as possible after birth to prevent lifelong morbidity. During the first 3 weeks of life relaxin hormone remains in circulation that keeps the ligament relaxed and stretching treatment is successful.[14]

In a study done by Colvin and Bower, children with birth defects were also more likely to hospital admission for reasons other than birth defects.[15] In the present study, a neonatal risk factor which required hospital admission were calculated, and the most common risk factor was neonatal jaundice 15 cases (35.71%) followed by birth asphyxia 10 cases (23.81%). Only two cases of multiple musculoskeletal defects were admitted without any risk factor. This study indicates that one must not overlook the presence of musculoskeletal defect at the birth of a newborn.

Children with genetic diseases and chromosomal disorders had an average hospital stay of 7.1 days as compare to 3.5 days average stay for children without any pre-existing medical disorder.[16] In our study, the average duration of hospital stay was 6 days. Out of total 42 cases detected as musculoskeletal birth defects, 4 cases (9.5%) were expired. In developing countries like India, congenital malformations were the third commonest cause of perinatal mortality. According to National Neonatology Database, the primary cause of stillbirths and neonatal deaths (9.6%) is contributed by malformations.[17] This finding is near to our finding of neonatal deaths during the study period. WHO estimates that birth defects accounted for some 556,000 deaths worldwide and for 145,611 out of 1,564,530 neonatal deaths in the year 2012.[18]

Birth defects are an emergent health priority in developed countries. From the findings of the present study, it is evident that musculoskeletal birth defects are common. They account for 1.35% cases admitted to neonatal care unit (13.46 per 1000 live births). The limitation of the present study is that it being from a single resource limited center; the sample size is small, but the findings are quite remarkable. Birth defects remain under-recognized owing to poor information on prevalence, and many countries do not have nationally representable data. To detect the musculoskeletal birth defects, a careful clinical examination by the primary care pediatrician could be important. A complete physical examination should be performed to rule out co-existing musculoskeletal and neuromuscular problems.[19] The need for a thorough systematic physical examination at birth cannot be over emphasized. Non-operative treatment should be started as soon as possible following birth. Services and intervention for prevention, treatment, and care of children with musculoskeletal birth defects must be integrated into the existing health care services.

CONCLUSION

The present study shows the incidence of musculoskeletal birth defects as 13.46 cases per 1000 live births admitted to our neonatal care unit indicating one of the common birth defects present in newborn. Multiple defects (57.14%) were more affected than isolated musculoskeletal birth defects. It is estimated that 49.46% of the cases were having major variants of musculoskeletal defects present at birth. Though the commonest reason for admission was neonatal jaundice, 54.76% of the cases required orthopedic consultation for the correction of deformity. Musculoskeletal birth defects are an emerging burden of tertiary care center and screenings of newborn infants for musculoskeletal birth defects by careful physical examination is needed. A multidisciplinary approach to identify, treatment and prevention of permanent disabilities are highly recommended. National data on musculoskeletal birth defects are under reported from resource-poor health care centers in India. Maintenance of musculoskeletal birth defects registries might play a vital role to understand the epidemiology and could be useful for policy makers to tackle the problem of birth defects. To conclude more studies on etiology, pathogenesis, early detection and treatment of musculoskeletal birth defect is an important area of pediatric health care research.

REFERENCES

1. Rajesh M, Neena R. Prevention and control of birth defects. J Neonatol 2014;28:23-8.
2. Rajwanti KV, Sandipan S. Approach to a malformed infant. In: Keya L, Mamta NM, editors. Clinical Genetics. 1st ed. New Delhi: Jaypee; 2005. p. 42-9.
3. Aase JM. Dysmorphologic diagnosis for the pediatric practitioner. Pediatr Clin North Am 1992;39:135-56.
4. March of Dimes. Global Report on Birth Defects. The Hidden Toll of Dying and Disabled Children. White Plains, NY: 2006. Available from: http://www.marchofdimes.org/materials/global-report-on birth defects-the hidden-toll-of-dying-and-disable-children-full-report.pdf. [Last accessed on 2014 May 19].
5. Muranjan M, Vijayalakshmi P. The unforeseen toll of birth defects and their economic burden at a tertiary care public institute in Mumbai. Indian J Pediatr 2014;81:1005-9.
6. Suresh S, Thangavel G, Sujatha J, Indrani S. Methodological issues in setting up a surveillance system for birth defects in India. Natl Med J India 2005;18:259-62.
7. Beals RK, Weleber RG. Distal arthrogryposis 5: A dominant syndrome of peripheral contractures and ophthalmoplegia. Am J Med Genet A 2004;131:67-70.
8. Day HJ. The ISO/ISPO classification of congenital limb deficiency. Prosthet Orthot Int 1991;15:67-9.
9. Moore KL, Persaud TV, Torchia MG. The musculoskeletal system. Text Book of Essentials of Embryology and Birth Defects. 7th ed. Philadelphia: John F. Kennedy; 2008. p. 243.
10. Robert BD. Congenital malformations. In: Robert BD, George B, editors. Mercer's Orthopaedic Surgery. 9th ed. London: Arnold; 1996. p. 178-80.
11. Mazumder ND, editor. Importance of genetics in neonatal orthopaedic diseases. In: Neonatal Orthopaedics. 1st ed. New Delhi: Jaypee; 2003. p. 13-6.
12. Ramesh A, Vinod KP, Ashok KD. Newborn infants, evaluation of newborn. In: Vinod KP, Arvind B, editors. Ghai Essential Pediatrics. 8th ed. New Delhi: CBS; 2013. p. 135-8.
13. Meharban S, editor. Miscellaneous conditions, twins. In: Care of the Newborn. 8th ed. New Delhi: CBS; 2015. p. 548-50.
14. Singh NV, editor. Congenital diseases. In: Fundamentals of Orthopaedics. 1st ed. New Delhi: New Age International; 2011. p. 177-85.
15. Colvin L, Bower C. A retrospective population-based study of childhood hospital admissions with record linkage to a birth defects registry. BMC Pediatr 2009;9:32.
16. McCandless SE, Brunger JW, Cassidy SB. The burden of genetic disease on inpatient care in a children's hospital. Am J Hum Genet 2004;74:121-7.
17. Verma IC. Burden of genetic disorders in India. Indian J Pediatr 2000;67:893-8.
18. World Health Organization: Disease and Injury Regional Mortality Estimates 2012. Available from: http://www.who.int/healthinfo/global-burden- disease/estimates/en/index1.html. [Last accessed on 2014 May 20].
19. Lawrence W, Kriti S, John PD. Bone and joint disorders, evaluation of the child. In: Kliegman RM, Stanton B, Game SJ, Schor NF, Behrman RW, editors. Nelson Textbook of Pediatrics. 19th ed. Philadelphia: Saunders; 2013. p. 2331-5.

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