

Musculoskeletal Birth Defects at a Tertiary Centre and Associated Maternal Risk Factors

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Abstract: This study was aimed at determining the incidence and risk factors of musculoskeletal birth defects in the newborns delivered at a tertiary centre. This was a prospective, observational study in a tertiary care hospital for a period of one year from 1st June, 2015 to 31st May, 2016 in Gauhati medical college and hospital. All newborns delivered in the obstetric department were screened for musculoskeletal birth defects. Clinical examination, skeletal survey and laboratory work up were performed, and data were analyzed. A total of 37 newborns were found to have skeletal abnormality with overall incidence of the musculoskeletal birth defect being per 1000 live births.. Male and Female ratio was 1.7:1. Among the 37 cases of musculoskeletal defects, the most common was congenital talipes equinovarus (15.22%). Primiparity, lower socioeconomic status, rural habitation were the associated maternal risk factors found.

Keywords: Birth defect, CTEV, Musculoskeletal

1. 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 definition; the two terms are used interchangeably. The musculoskeletal birth defects cause long-term disability in the survivors as well as an economic burden to the families and society at large. 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. Early referral to concerned specialists and appropriate treatment of musculoskeletal birth defects can prevent disabilities and reduces permanent morbidities among the survivors. According to National Neonatology Database, the primary cause of stillbirths and neonatal deaths (9.6%) is contributed by malformations¹. 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 2012². To detect the musculoskeletal birth defects, a careful clinical examination by the primary care paediatrician could be important. A complete physical examination should be performed to rule out co-existing musculoskeletal and neuromuscular problems³.

2. Materials And Methods

The present study is the single-center, prospective, observational study done in a tertiary care institute. All the newborns with birth defects who were born during a period of one year from 1st June, 2015 to 31st May, 2016 were enrolled after approval from the Institutional Ethical Committee and system wise classification of birth defects

were done. Newborns were examined for presence of birth defects immediately after birth by the on duty doctors and were registered in birth defect register.

Inclusion criteria

- 1) Both live and stillborn babies were included in the study.
- 2) Newborns with birth defects involving multiple systems were included in the study.
- 3) Only those cases which were detected during the immediate post delivery hospital stay were included in the study.

Exclusion criteria

- 1) Multiple pregnancies were excluded from the study.
- 2) Those newborns which were delivered at institution other than Gauhati medical college were excluded from the study.

The overall incidence, socio-demographic data, paternal age, maternal risk factors such as age, antenatal screening, drug and radiation exposure, previous anomalous baby, history of tobacco and alcohol exposure etc were documented after interviewing the respective parents. Follow-up plan of the cases was not included in the study. All the data were calculated and analyzed.

3. Results

A total of 16044 newborns were delivered during the study period and out of them, 149 cases were diagnosed to have congenital anomaly. 37 cases of musculoskeletal system birth defects were selected for the study after fulfilling the inclusion criteria and exclusion criteria. The overall incidence of musculoskeletal birth defects was 24.83% of total birth defects. Among the 37 cases of musculo skeletal defects 15 cases of CTEV, 7 cases of Omphalocele, 3 cases of Gastroschisis, 3 cases of Congenital diaphragmatic hernia, 2 cases of polydactyly, 1 Syndactyly, 1 case of

reduced upper limb, 1 case of Poland syndrome, 1 case of Congenital Constriction band, 1 case of skeletal dysplasia, 1 case of Thanatophoric Dwarfism and 1 case of lobster clawhand were observed. Thus CTEV was the most common musculoskeletal defect accounting for 40.54%.

Table 1: Showing Distribution of Birth defects of musculo skeletal system

Type of defect	No of cases	Percentage (%)
CTEV	15	40.54
Omphalocele	7	18.92
Gastroschisis	3	8.11
Congenital diaphragmatic hernia	3	8.11
Congenital constriction band	1	2.7
Congenital absence of right hand and fingers	1	2.7
Polydactyly	2	5.4
Lobster claw hand	1	2.7
Syndactyly	1	2.7
Skeletal dysplasia	1	2.7
Thanatophoric dwarfism	1	2.7
Poland's Syndrome	1	2.7

Among the study group, male were 23 (62.16%) and female were 12 (32.43%) and there were two cases with ambiguous genitalia (5.40%). Male to female ratio was 1.91:1.

Table 2: Showing Sex Distribution Among Newborns with Musculoskeletal System Defect

Sex distribution	No. of cases	Percentage (%)
Male	23	62.16
Female	12	32.43
Ambiguous genitalia	2	5.40

The majority of cases 25 (67.56%) belonged to the lower socioeconomic class. Majority of cases, i.e. 22 mothers were in the age group of 20 to 25 years (59.45%). 25 cases were from rural habitation. 24 cases were of term gestation and rest 13 cases were born before 37 weeks of gestation. 26 cases were booked and 11 cases were unbooked.

Only 9 cases were detected by antenatal ultrasonography and rest were detected after delivery. 2 cases were detected to have oligohydramnios and another 2 cases were associated with polyhydramnios. 19 cases (51.35%) were associated with maternal anaemia. None of the cases had a family history of the similar defect, and there was no history of consanguineous marriage among the parents. 19 cases (51.35%) were born to Primigravida. 27 cases were liveborn and rest 10 cases were stillborn with a ratio of 2.7:1. None of the cases took pre-conceptional folic acid.

Out of 37 cases, isolated musculoskeletal birth defects were detected in 30 cases (81.08%), and multiple defects were noted in 6 cases (16.21%) and one case comprised of a syndrome. Among the multiple musculoskeletal birth defects, one case was diagnosed as Polands syndrome. The most common association was seen to be with central nervous system birth defects.

Table 3: Showing Distribution of Isolated and Multiple Birth Defects

Type of musculoskeletal defect	No. of cases	Percentage (%)
Isolated	30	81.08
Multiple defects	6	16.21



Achondroplasia Amputated right hand



CTEV Omphalocele



4. Discussion

There were 37 cases of musculoskeletal defects accounting to 24.83% of total birth defects which were the second most common birth defects in the present study. In most of the studies for birth defects in India, the predominant system involved was musculoskeletal system. Shatanik Sarkar et al ;(2014), did a study on prevalence of congenital anomalies in newborn in a tertiary centre where he found musculo skeletal defects to be 33.2% of total birth defects.⁴ El Koumi MA et al; (2013), found 23% of total birth defects to be of musculoskeletal system in his study on pattern of congenital malformation.⁵ Krikunova N I et al⁶ in a hospital based study of birth defects found highest cases of birth defects were of Musculoskeletal system comprising of 37.68% of total birth.

In our study of 37 musculoskeletal birth defects, male children were more affected than female . Bakare T et al; (2009)⁷, Taksande Amar et al; (2010)⁸, Hossein et al; (2014)⁹, in their study of congenital anomaly, found highest prevalence of birth defects amongst male newborns. The majority of cases belong to the lower socioeconomic group. In a study conducted by Vrijheid M et al; (2001), it was found that risk of structural anomalies were more in population with increased socio-economic deprivation¹⁰.

The same clinical sign or malformation may be caused by a variety of genetic defects in addition to the environmental causes. None of the mothers in the present study had a history of teratogenic drug intake during the antenatal period. There was no similar musculoskeletal birth defect among the family members and siblings.

CTEV was the most common musculoskeletal birth defect found in the present study. In the general population, the incidence of congenital talipes equinovarus (CTEV) is 1 in 1000 live births and there is a 1:800 chance of having this deformity; 1:3.5 chance in sibling and 1:3 chance in an identical twin.¹¹

Omphalocele was the second most common birth defect found in the present study. CDC (centres for disease control and prevention) researchers have reported about some factors like Alcohol and tobacco consumption, certain medications like selective serotonin-reuptake inhibitors (SSRIs) intake during pregnancy-which can increase the risk of having a baby with an Omphalocele.^{12,13} However in the present study none of the cases gave history of exposure to alcohol, tobacco or SSRIs. The causes of most of the birth defects remained unknown. Most of the cases had no obvious risk factors.

5. Conclusion

Raising awareness regarding antenatal care, improving socioeconomic status, use of periconceptional folic acid, early diagnosis by anomaly scan, Neonatal screening for birth defects and early referral to specialists are the major requirements to prevent and reduce this burden of musculoskeletal deformities among our society.

Conflict of interest-nil

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