Original Article

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Prevalence and Pattern of Congenital Musculoskeletal Anomalies: A Single Centre Study

Obstetrics and Gynaecology Section

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ABSTRACT

Introduction: Musculoskeletal defects are second most common birth defects after central nervous system anomalies. Only very few studies from India concentrates on the pattern of patients specifically dealing with congenital musculoskeletal anomalies.

Aim: To determine the pattern and birth prevalence of musculoskeletal congenital malformations among the subset of neonates and infants born in a single tertiary care centre of north India and referred to Orthopaedic Department from Obstetrics and Gynaecology and Paediatric Department.

Materials and Methods: A retrospective analysis of all the referrals made from Obstetrics as well as Paediatric Department to Orthopaedic Department from September 2014 to August 2016 for assessment of any visible congenital limb defect in a neonate was done. Only those infants were included in the study who were born in the same hospital during the study period. A record of all the deliveries conducted during the study period was also analysed. Pattern of anomalies in upper limb, lower limb and axial skeleton was noted. Birth prevalence of all the musculoskeletal anomalies was also determined.

Results: During the study period a total of 10,126 births were recorded and 71 infants were found to have some form of musculoskeletal defect with prevalence of 7.01/1000 births. Out of 71 cases, 43 (60.56%) were males and 28 (39.44%) females. Ten (14%) children had the anomalies affecting the upper limb with a prevalence of 0.99/1000 births while in 49 (69%) the lower limb was affected with a prevalence of 4.84/1000 births. Twelve (16.9%) children had anomalies affecting the axial skeleton (birth prevalence 1.18/1000). The most common anomalies were Congenital Talipes Equinovarus (CTEV) (33 cases; 46.48%), Developmental Dysplasia of Hip (DDH) (10 cases; 14.08%) and spina bifida (seven cases; 9.86%) with a birth prevalence of 3.25/1000, 0.99/1000 and 0.69/1000 respectively.

Conclusion: Congenital musculoskeletal anomalies has a prevalence of 7.01 per 1000 live births. CTEV and DDH are one of the commonest anomalies which are easy to treat if recognised early. Screening of every newborn by an Orthopaedic specialist is warranted.

Keywords: Defects, Limb, Orthopaedic

INTRODUCTION

A birth defect is defined as any abnormality affecting the body structure or function that is present since birth and may be clinically obvious or may be diagnosed only later in life [1]. The frequency of the occurrence of congenital anomalies is usually quoted as birth prevalence rather than incidence [2]. This is because many foetuses affected by a congenital anomaly will miscarry, the miscarriage may occur before the pregnancy is confirmed and even if the miscarriage occurs later, the anomaly may not be diagnosed. Birth prevalence is the number of infants affected by any congenital anomaly per 1000 live births. The birth prevalence of congenital anomalies present at birth or detected later in childhood is actually underestimated in the developing countries due to lack of diagnostic techniques and their reliability [1,2]. According to March Of Dimes (MOD) Global Report on Birth Defects, worldwide, 7.9 million births occur annually with serious birth defects and 94% of these occur in the middle and low income countries [1]. According to joint World Health Organisation (WHO) and MOD meeting report, birth defects account for 7% of all neonatal mortality and 3.3 million under five deaths [3].

Congenital anomalies are not considered to be a priority health problem in India in spite of the fact that it has the highest number of children with birth defects. In India, birth defects prevalence varies from 61 to 69.9/1000 live births [3]. India being the second most populous country in the world has a high birth rate but no proper screening program for recognition of Congenital Limb Defects (CLD).

Of the congenital birth defects, the musculoskeletal defects seen in childhood occur when a part of, or the entire limb fails to form normally during pregnancy. The aetiology of congenital abnormalities is multifactorial and may be genetic or environmental with a complex interaction of these in many cases [4]. Disruptive events such as amniotic band or vascular disruptions appear to be the most common cause of CLD [5,6]. One of the important cause of CLD is prenatal exposure to different teratogens, the best known example of which is thalidomide, which caused a wide range of deformities in the 1960s [7,8].

According to a study done by WHO, musculoskeletal defects are second most common birth defect after central nervous system anomalies with a prevalence of 51.12/1000 population [9]. India having a large number of infants born annually with birth defects needs a strict surveillance for the same and separate registry for keeping updated about prevalence of these congenital anomalies.

On thorough search of the literature many studies were found depicting the prevalence of various congenital anomalies among all the in-hospital live births [1,3,4,5,9]. However, only one was found on the pattern of patients specifically dealing with musculoskeletal anomalies [10]. Infants and children with a congenital musculoskeletal defect needs specialised Paediatric Orthopaedic care facility which can provide comprehensive care, right from diagnostic evaluation and surgery to prosthetic rehabilitation. It also calls for close coordination among Obstetric, Paediatric and Orthopaedic Departments for

early recognition and comprehensive care of musculoskeletal defects under one roof. Keeping this fact in mind, the present study was carried out with the aim to determine the pattern and birth prevalence of musculoskeletal congenital malformations among the subset of neonates and infants born in a single hospital and referred to Orthopaedic specialist from Obstetrics and Gynaecology as well as Paediatric Department of a tertiary care centre of north India. The data and results from this study can help government in policy making, planning and allocation of resources and funds for development and need of an appropriate Paediatric orthopaedic care facility at designated tertiary referral centres.

MATERIALS AND METHODS

This hospital based retrospective observational study was conducted at SRMS Institute of Medical Sciences, Bareilly, a tertiary referral centre in northern state of India between September 2014 to August 2016. All live born infants from birth to one year of age were included while all still births, even if born after 28 weeks of gestation were excluded. Only those infants were included in the study who were born in the same hospital during the study period. An analysis of all the referrals made from Obstetrics Department to Orthopaedic Department for assessment of any visible congenital limb defect in a neonate was done. Analysis of all the referrals from Paediatric Department to Orthopaedic Department for assessment and management of all the children from birth to one year of age was also done. A record of all the deliveries conducted during the study period was also analysed. During the study period a total of 10,126 births were recorded and 71 infants were found to have some form of musculoskeletal defect detected by Obstetrician, Paediatrician or Orthopaedic surgeons.

Anomalies affecting the upper limb were classified according to Swanson's classification into six groups as failure of formation, failure of differentiation, duplication, overgrowth, undergrowth and miscellaneous anomalies [11]. Separate column was made for lower limb and axial skeleton anomalies. The data was collected, entered and tabulated into excel data sheet and appropriate analysis were performed. All children were treated by standard treatment protocol for the given condition and are under long-term follow up of Orthopaedic department.

STATISTICAL ANALYSIS

The data was statistically analysed by using the Chi-square test and probability test (ANOVA). The software used was SPSS 16.0 (Chicago, Illionis).

RESULTS

A total of 71 cases with congenital musculoskeletal anomalies were detected out of 10,126 births patients during the study period with prevalence of 7.01/1000 births. This study does not reflect the exact incidence or birth prevalence in the population as it is a hospital based retrospective study based on infants born in a single centre and also high-risk cases are referred for delivery from distant places of western Uttar Pradesh and Uttarakhand. Out of 71 cases, 43 (60%) were males and 28 (40%) females [Table/Fig-1]. Ten (14%) children had the anomalies affecting the upper limb while in 49 (69%) the lower limb was affected. Twelve (16.9%) children had anomalies affecting the axial skeleton. Incidence of all anomalies as well as each individual anomaly per 1000 births was also calculated. The pattern and distribution of all the anomalies is depicted in [Table/Fig-2,3 and 4].

Congenital anomaly	Males	Females
Lower limb		
CTEV	22	11
DDH	4	6
AMC	1	1
Fibular hemimelia	1	0
Tibial bowing	1	0
Tibial hemimelia	1	0
PFFD	0	1
Upper limb		
Radial clubhand	2	0
Macrodactyly	1	0
Cong Trigger thumb	1	0
Lobster hand	1	0
Sprengel shoulder	0	1
Hypoplastic thumb	1	1
Cong.torticollis	1	1
Axial skeleton		
Meningomyelocele	3	2
Spina bifida	3	4
Total	43 (60.56%)	28 (39.43%)

[Table/Fig-1]: Sex distribution of various musculoskeletal anomalies.
CTEV (Congenital Talipes Equino Varus); DDH (Developmental Dysplasia of Hip); AMC (Arthrogryposis Multiplex Congenital); PFFD (Proximal Femoral Focal Deficiency)

Upper Limb Anomalies			Prevalence/1000 births*	% of all anomalies
Failure of part formation	Radial club hand	2	0.20/1000	2.82%
Failure of differentiation	Cong Trigger Thumb	1	0.10/1000	1.41%
Duplication	Lobester Hand	1	0.10/1000	1.41%
Overgrowth	Macrodactly	1	0.10/1000	1.41%
Undergrowth	Thumb Hypoplasia	2	0.20/1000	2.82%
Miscellaneous	Torticollis	2	0.20/1000	2.82%
	Sprengels shoulder	1	0.10/1000	1.41%
Total		10	0.99/1000	14%

[Table/Fig-2]: Distribution, pattern and birth prevalence of upper limb anomalies.
*Prevalence calculated as total number of live birth infants with a congenital anomaly per 1000 live births.

Lower Limb Anomalies		Prevalence/1000 births*	% of all anomalies
CTEV	33	3.25/1000	46.48%
DDH	10	0.99/1000	14.08%
AMC	2	0.20/1000	2.82%
Fibular Hemimelia	1	0.10/1000	1.41%
Tibial bowing	1	0.10/1000	1.41%
Tibial Hemimelia	1	0.10/1000	1.41%
PFFD	1	0.10/1000	1.41%
Total	49	4.84/1000	69%

[Table/Fig-3]: Pattern, distribution and birth prevalence of lower limb anomalies.
*Prevalence calculated as congenital anomaly present in a live birth infant per 1000 live births
CTEV (Congenital Talipes Equino Varus); DDH (Developmental Dysplasia of Hip); AMC (Arthrogryposis Multiplex Congenital); PFFD (Proximal Femoral Focal Deficiency)

Upper Limb Anomalies

Ten (14%) children had the anomalies affecting the upper limb and the incidence was found to be 0.99/1000 births. The most common congenital malformations affecting upper limb and proximal appendicular skeleton were radial club hand, congenital muscular torticollis and thumb hypoplasia. Radial club hand was found in two patients (2.82%), both in males. Congenital muscular torticollis and thumb hypoplasia was also found in two patients (2.82%) each one in male and female. Sprengel shoulder, congenital trigger thumb, lobster hand, macrodactyly were found in one patient (1.41%) [Table/Fig-2].

Axial malformations		Prevalence/1000 births*	% of all anomalies
Spina bifida	7	0.69/1000	9.86%
Meningomyelocele	5	0.49/1000	7.04%
Total	12	1.18/1000	17%

[Table/Fig-4]: Pattern, distribution and birth prevalence of axial malformations. *Prevalence calculated as congenital anomaly present in a live birth infant per 1000 live births.

Lower Limb Anomalies

Forty nine (69%) children had anomalies affecting the lower limb with birth prevalence of 4.84/1000. The most common anomaly was CTEV followed by DDH. CTEV was found in 33 patients (46.48%), 22 males and 11 females. DDH was found in 10 patients (14.08%), four males and six females [Table/Fig-3].

Arthrogryposis multiplex congenita was found in two patients (2.82%), one each in a male and female. Fibular hemimelia, congenital bowing of tibia, tibial hemimelia and Proximal Femoral Focal Deficiency (PFFD) were found in one patient (1.41%) each.

Axial Malformations

Twelve (17%) patients had congenital axial malformations and the birth prevalence was found to be 1.18/1000. Spina bifida was found in seven patients (9.86%), three males and four females. Meningomyelocele (MMC) was found in five patients (7.04%), three males and two female [Table/Fig-4].

DISCUSSION

Congenital musculoskeletal anomalies are among the major causes of childhood morbidity around the world and many large studies are being conducted for surveillance of the same [12-15]. In India, there is no national surveillance system to measure the magnitude of these anomalies, and few cross-sectional studies have yielded wide differences in overall prevalence rates in different parts of the country [16-18].

In this retrospective analysis of Orthopaedic referrals for congenital limb defects by Obstetrics and Paediatrics Department, we specifically studied the birth prevalence of various musculoskeletal anomalies, their pattern of involvement in the upper/lower limb or axial skeleton and their sex distribution. We have included only those cases which were delivered in the same centre during the study period so that exact birth prevalence of these anomalies can be calculated. The cut-off age of one year was kept as many anomalies remain undetected by obstetricians at time of birth but later detected by paediatricians during regular follow up visits or for immunisation.

In this study, the prevalence of major musculoskeletal anomalies was found to be 7.01/1000 births. According to a study conducted by WHO, musculoskeletal anomalies were seen as second most common cause of congenital anomalies with prevalence rate of 5.1/1000 population [9]. In a study from Uttarakhand region of north India, musculoskeletal anomalies were recognised as third most common cause of congenital anomalies with 0.94% of total births [16]. In a study done in central India, the prevalence of musculoskeletal malformations was 3.9/1000 population [17]. A study done in northeast India revealed the prevalence of musculoskeletal anomalies as 4.36/1000 population [10]. The prevalence of congenital limb malformations in a long-term population based study in Netherlands was found to be 21.1/10000 population [14].

On comparison of results of this study with the available international, national and regional data it was found that western literature shows less prevalence of congenital limb malformations while other studies from India shows almost similar prevalence of limb malformations. Although, the studies in question are not directly comparable and the data from this study could be generalised to the larger population to some extent.

In our study, out of 71 cases, 43 (60.56%) were males and 28 (39.43%) females with male: female ratio of 1.5:1. Other national and international studies shows male: female ratio of 1.2:1 [10,14-16,19]. Increased involvement of male sex has been reported previously, but its aetiology remains unclear [20,21].

In this study, upper limb anomalies were seen in 10 (14%) cases with a prevalence of 0.99/1000 births. The most common malformations affecting upper appendicular skeleton was radial club hand and congenital muscular torticollis. A study done in USA found more longitudinal (3.5/10,000 births) than transverse reduction defects (1.9/10,000 births) [5]. However, various other studies have found transverse reduction defects to be more common. The prevalence of such upper limb defects was found to be 3.9/10,000 population in Netherlands, 4.3/10,000 births in France, 2.6/10,000 in Italy, and there was a prevalence of 4.0/10,000 in six combined EUROCAT registries (Strasbourg, Belfast, Emilia Romagna, Odense, Groningen, Basque Country) [14,20,22]. The differences in pattern of anomalies among various countries can be contributed to the various environmental and genetic factors playing role in their occurrence.

In this study, lower limb anomalies were found to be the most common. 49 (69%) cases had anomalies affecting the lower limb with clubfoot being the most common (46.48%; 3.25/1000 births) followed by DDH (14.08%; 0.99/1000 births). In a WHO sponsored Global study the incidence of clubfoot was found to vary from 3.42/1000 in Kolkata to 10.95/1000 in Panama City as reported by Stevenson AC et al., [23]. The main reason for this variation could be that in some hospitals, any malposition of feet was recorded as Talipes. In most of the Indian studies lower limb anomalies were the most common, and clubfoot was the commonest of these [9,16-18].

In western literature, upper limbs were more commonly affected, as compared to lower limb, in contrast to our study and other Indian studies [14,22,24]. This could be attributed partly due to different sampling techniques, different inclusion criteria and partly due to the different genetic makeup and environmental factors which play an important role in the etiogenesis of congenital anomalies [7,8]. However, this contrast needs to be further investigated at the multicentre level in India to establish the pattern of these musculoskeletal anomalies, which would play an important role in the formulation of the healthcare policies at the national and the regional level.

In this study, we also included and studied the pattern of congenital malformations of the axial skeleton which was lacking in the previous studies [17,18,20,21]. In the present study, 12 (17%) patients were affected by congenital axial malformations and spina bifida was the most common anomaly (seven cases, 9.86%) followed by MMC (five cases, 7.04%). Congenital anomalies of skull were excluded. Axial malformations were found to be the second commonest after lower appendicular skeleton. Some studies from India have reported central nervous system malformations as most common congenital anomaly [9,18,19], but they cannot be compared to our study as they have included all the congenital malformations of central nervous system.

LIMITATION

This study is the first of its kind in India which has analysed solely the pattern of congenital musculoskeletal anomalies. Some cases may have been missed due to non-detection at time of birth and which never returned for follow up either in Paediatric or Orthopaedic OPD, so the results could not be generalised to the whole population.

CONCLUSION

Congenital musculoskeletal anomalies has a prevalence of 7.01 per 1000 live births. Lower limbs anomalies (69%) are more common than upper limb and axial skeleton anomalies. In lower limb the most common anomalies are CTEV and DDH which are easy to treat if recognised early by an Orthopaedic specialist with no long-term disability to child. Screening of every newborn in hospital setting by an Orthopaedic specialist is warranted.

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