

## A Study of Patterns of Congenital Malformations of Musculoskeletal System in Perinates

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### ABSTRACT

**Background:** The term congenital malformation signifies disruption in the normal process of organogenesis before birth. This may result in complete or partial absence of a structure or in alteration of its normal configuration. The knowledge of prevalence of congenital malformations help in establishment of preventable programmes. Hence the present study was carried out to find out the frequency of congenital malformations of musculoskeletal system both in live and still born perinates.

**Methods:** All live and still born babies of both sexes, with congenital malformations were collected from the Department of Obstetric and Gynecology, Assam Medical College and Hospital, Dibrugarh, Assam, from July 2022 to June 2023 and were examined and epidemiological information were collected from parents in a pretested structured proforma.

**Results:** Out of 7985 births, 38 cases of congenital malformations were found, 31 (17.41%) were stillbirth and 7 (0.09%) were live birth. The musculoskeletal system involvement was seen in 8 cases with congenital talipes equinovarus (CTVS) deformity and polydactyly being the commonest malformation followed by rudimentary forearm and a case of achondroplasia. Central nervous system anomalies were mostly associated with musculoskeletal system anomalies.

**Conclusion:** The findings of the present study will help to plan future strategies for prevention, early diagnosis and timely management of cases of congenital malformations of musculoskeletal system.

**KEY WORDS:** congenital talipes equinovarus (CTEV), polydactyly, achondroplasia.

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### INTRODUCTION

Malformations occur during formation of structures, for example, during organogenesis. Most malformations have their origin during the third to eight weeks of gestation [1].

According to WHO, perinatal mortality is defined as the late fetal death (28th weeks of

gestation or more) plus early neonatal death (1st week) in year per thousand live birth, in the same year. It has been reported that in India congenital malformations have emerged as the 3rd commonest cause of perinatal morbidity and mortality, next only to malnutrition and infections [2]. So, congenital

malformations are one of the major childhood health problems. Treatment and rehabilitation of children with congenital malformations is costly and complete recovery is usually impossible [3]. Congenital anomalies of the musculoskeletal system refer to the anomalies of the muscular and skeletal system present at birth of the child. Approximately, 66% of major malformations have no recognized etiology and most of them have multifactorial inheritance [4].

The proved causes of congenital anomalies of the musculoskeletal system are:- 1. Hereditary factors (chromosomal defect), 2. Nutritional factors (folic acid deficiency) and 3. Environmental factors e.g. German measles, and some of these are preventable [5]. The magnitude of the problem of musculoskeletal system anomalies in Northeast India is still not known [6]. Also, most of the studies had been done on congenital anomalies of the musculoskeletal system with very few studies being done on association with other anomalies.

Hence, the present study was carried out (1) to find out the frequency of congenital malformations of musculoskeletal system and (2) to find out the frequency of associated anomalies both in live and still born perinates in our region.

## METHODS

**Place of study:** Department of Anatomy, Assam Medical College & Hospital, Dibrugarh, Assam, India.

**Type of study:** Cross sectional observational study.

**Study population:** All consecutive born perinates (total number is 7985) from 28th weeks of gestation to 7 days after birth (both live and still born) in the Department of Obstetrics and Gynaecology, Assam Medical College and Hospital, Dibrugarh, Assam.

**Period of study:** One year (July 2022 to June 2023).

**Exclusion criteria:**

1. Fetuses born less than 28 weeks,
2. Abortus and macerated fetuses.

**Procedure:** All live and still born babies of both sexes, with congenital malformations within the period of one year (July 2022 to June 2023) of study were collected from the Department of Obstetric and Gynecology, Assam Medical College and Hospital, Dibrugarh, Assam. The babies were examined and epidemiological information were collected from parents in a pretested structured proforma, after taking written consent from them.

The autopsy of the still born fetuses (after fulfillment of all official procedures) was done in the Department of Anatomy, Assam Medical College and Hospital, to detect presence of congenital malformations of musculoskeletal system. When malformation was detected, information regarding birth order, sex, birth weight, H/O consanguinity, H/O teratogenic drug exposure were obtained by systematic maternal and paternal interviews, and information were noted in the proforma.

The findings were noted and presented in tabular form. Statistical calculations were done in microsoft excel sheet.

## RESULTS

Out of 7985 births, 38 cases (0.47%) of congenital malformations were found, 31 (17.41%) were stillbirth and 7 (0.09%) were live birth (Table—1). Among these 38 cases, 20 (52.64%) were male and 18 (47.36%) were female. The distribution of congenital malformations were seen to be high among male children with a male female ratio of 1.11:1 (Table—2). Congenital malformations were seen to be more common in the preterm babies than the term babies (Table-3). The musculoskeletal system involvement was seen in 8 cases with congenital talipes equinovarus deformity and polydactyly being the commonest malformation followed by rudimentary forearm and a case of achondroplasia as shown in Table—4 (Fig 1).

Involvement of musculoskeletal system anomalies with anomalies of other systems was seen in six cases. The distribution of the cases is shown in Table-5 (Fig 2). Involvement of few syndromes with anomalies of musculoskeletal system is shown in Table-6.

**Table 1:** Showing frequency of congenital malformations.

Birth	No. of cases	No. of congenital malformations	Percentage %
Total	7985	38	0.47%
Still born	178	31	17.41%
Live born	7807	7	0.09%

**Table 2:** Showing distribution of cases of congenital malformations in relation to sex.

Sex	No. of congenital malformations	Percentage %	Ratio
Male	20	52.64%	
Female	18	47.36%	1.11:1
Total	38	100%	

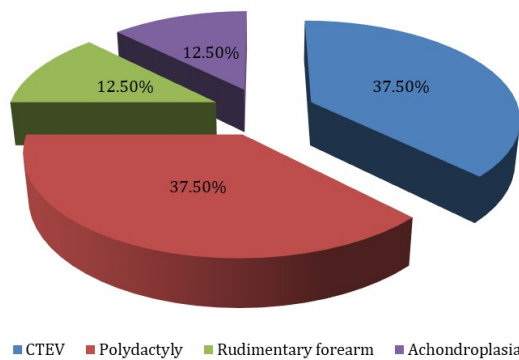
**Table 3:** Showing distribution of cases in relation to period of gestation.

Sl. No.	Gestation period	Total No. of cases of cong. malformations	Percentage %
1	Preterm	25	65.79%
2	Term	13	34.21%

**Table 4:** Showing distribution of congenital malformations of musculoskeletal system.

Malformations	No. of cases	Incidence per 1000 births
CTEV	3	0.375
Polydactyly	3	0.375
Rudimentary forearm left	1	0.125
Achondroplasia	1	0.125

**Figure 1:--Musculoskeletal system malformations .**

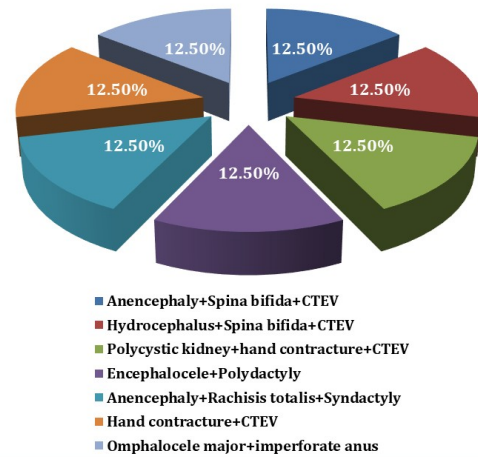


**Fig 1:** Musculoskeletal system malformations

**Table 5:** Showing involvement of multiple systems.

Sl. No.	Malformations	No. of cases	Incidence per 1000 births
1	Anencephaly, Spina Bifida, CTEV	1	0.125
2	Hydrocephalus, Spina bifida, CTEV	1	0.125
3	Hand contracture, CTEV	1	0.125
4	Polycystic kidney, Hand contracture, CTEV	1	0.125
5	Encephalocele, Anencephaly, Polydactyly	1	0.125
6	Anencephaly, Rachis totalis, Syndactyly	1	0.125
7	Omphalocele major, imperforate anus	1	0.125

**Figure 2:--Multiple system malformations .**



**Fig 2** Multiple system malformations

**Table 6:** Showing involvement of miscellaneous syndromes.

Sl. No.	Malformations	No. of cases	Incidence per 1000 births
1	Prune belly syndrome	1	0.125
2	Frontonasal hypoplasia	1	0.125
3	Fetal hydrops	1	0.125



**Fig. 3:** photograph showing bilateral CTEV.



**Fig. 4:** photograph showing polydactyly



**Fig. 5:** Photograph showing rudimentary forearm (left).



**Fig. 9:** photograph showing fronto-nasal hypoplasia



**Fig. 6:** photograph showing achondroplasia.



**Fig. 10:** photograph showing fetal hydrops



**Fig. 7:** photograph showing hand contracture with CTEV.



**Fig. 8:** photograph showing omphalocele major with imperforate anus

## DISCUSSION

Most children who are born with major congenital anomalies and survive infancy are affected physically, mentally or socially and can be at increased risk of morbidity due to various health disorders [7].

Although the incidence of congenital malformations has been reported to be 3—5% worldwide and 3.65% in infant at birth in India, in Assam overall incidence has been reported to be 0.08% [8,9].

In the present study, the incidence of congenital malformations was found to be 0.47% of the total births. The percentage of malformations observed was quite low when compared with the study reports of Singh A et al [10], Rathod SP et al [11] and Ramakrishna D et al [12].

Several studies have reported higher incidence of malformation such as those of Swain S et al [13] (Table—7).

The rate of malformations in the present study was much higher in still births (17.41%) as compared to live births (0.09%).

Hospital based studies carried out in various cities in India, has also shown that the incidence was significantly higher in still born babies. A study conducted in Ajmer by Gupta BM et al [14] (1971), reported the rate of congenital malformation as 1.9 % in live born in comparison to 20.4% in still born babies.

Study conducted by Chandra P & Harilal KT [15] (1977) at Madras and Mathur BC et al [16] (1975) in Hyderabad observed the rate of malformation to be more in still born than in live born babies (1.9% to 20.4% & 1.4% to 12% respectively).

According to Chaturvedi et al [17] (1989) and Bhat et al [18] (1998) the incidence of congenital malformations in the still born fetuses was 9.8% and 15.7% when compared to live born fetuses 2.4% and 3.2% respectively.

### Musculoskeletal System

The musculoskeletal system is the second commonest congenital malformations. In the present study, 21.05% of the total cases of malformation had musculoskeletal system involvement and this was lower than the findings of Singh A et al (2009) (30.6%) [10].

The incidence of musculoskeletal malformation in the present study was 1/1000 live birth. On being compared, it was seen to be lower in observations of Ghosh S et al [20] in 1963 (14.7/1000), Tiberwala NS et al [22] in 1974 (3.56/1000), Datta V et al [25] in 2000 (3.11/1000), Sugunabai NS et al [23] in 1982 (1.4/1000), as shown in table 7. McIntosh R et al [19] (1954) and Singh A et al [26] (2009) reported it to be much higher 30/1000 and 75/1000 births respectively. In all these studies, the musculoskeletal system anomalies were either heading the list or were second most common type. This was probably because musculoskeletal malformations were externally visible and readily identifiable at birth [27].

**Table 7:** Comparison of musculoskeletal system anomalies with previous studies

Sl. No	Author	Year	Msk	Multiple	Misc.
1	Mcintosh R et al [19]	1954	154(28/1000)	0	5 (01/1000)
2	Ghosh S et al [20]	1963	64 (14.7/1000)	0	0
3	Stevenson AC et al [21]	1966	22 (1.14/1000)	0	0
4	Tiberwala NS et al [22]	1974	44 (3.56/1000)	0	4 (0.32/1000)
5	Sugunabai NS et al [23]	1982	10 (1.4/1000)	30(4.2/1000)	13 (1.8/1000)
6	Choudhury A et al [24]	1984	9 (0.44/1000)	2(0.09/1000)	0
7	Datta V et al [25]	2000	9 (3.11/1000)	0	13 (4.53/1000)
8	Singh A et al [26]	2009	75 (0/1000)	0	0
9	Agarwal et al [27]	2014	26 (3.58/1000)	6(0.83/1000)	0
10	Prashar et al [28]	2016	49 (3.2/1000)	0	0
11	Shylaja et al [29]	2017	37 (2.9/1000)	0	0
12	Kumari et al [30]	2018	71 (7.01/1000)	0	0
13	Akinmoladun JA [31]	2021	14 (4/1000)	0	0
14	Emeka et al [32]	2022	49 (7/1000)	0	0
15	<b>Present study AMCH</b>	<b>2022-23</b>	<b>8 (21.05) (1.00/1000) Births</b>	<b>8(21.05%)1.00/1000 Births</b>	<b>3 (7.90%) 0.25/1000 Births</b>

**MSK-** musculoskeletal system; **MISC-** miscellaneous syndromes; **MULTIPLE-** multiple system

Talipes equinovarus deformity and polydactyly were the commonest musculoskeletal system malformation observed in the present study (incidence of each malformation being 0.375/1000 births), followed by one case of rudimentary forearm and achondroplasia (0.125/1000 births).

Sadler TW [1] states that achondroplasia, the most common form of dwarfism, occurs in one per 26,000 live births. It primarily affects the long bones, large skull (megaloccephaly) with small midface, short fingers, and accentuated spinal curvature. The achondroplasia case in the present study had deformed long bones, large skull and short fingers as shown in Figure 6.

In the year 1966, in a WHO sponsored global study, the incidence of talipes equinovarus was found to vary from 0.42/1000 in Kolkata to 10.85/1000 in Panama city [21].

**Multiple System involvement:** Among the multisystem group the musculoskeletal system involvement was seen in seven cases (0.88/1000 births).

One case (0.125/1000) of omphalocele major with imperforate anus (Figure 8), one case (0.125/1000 births) of hand contracture along with CTEV (Figure 9) was found, as shown in Table 5.

Sugunabai NS et al [23] observed an incidence of involvement of multiple systems in 5.7/1000 births, which was quite high when compared with the present study. Whereas Choudhury A et al [24] have noted incidence of 0.14/1000, which is comparably less than the present study (Table 5).

In the present study, central nervous system malformations were mostly associated with musculoskeletal system anomalies.

**Miscellaneous group:** In the present study, three cases 3/38 (7.90%) of miscellaneous syndromes were noted with the incidence rate of 0.37/1000 births. These were Prune belly syndrome, Frontonasal hypoplasia and Fetal hydrops. Prune belly syndrome was observed in a male fetus where there were defects of abdominal wall. Sadler TW1 described that this defect results due to partial or complete absence of abdominal musculature.

A female preterm baby of fetal hydrops (with an incidence rate of 0.125/1000 births) (Figure 10) and a case of live term female baby with fronto nasal hypoplasia (incidence rate of 0.125/1000 births) (Figure 7) was also found in the present study shown in Table 6.

Most of the observations are comparable with the similar studies undertaken in the other parts of the country. However, some of the observations differed from findings of other authors, which was probably due to different study design or due to differences in geographical or environmental factors and difference in time period.

## CONCLUSION

In conclusion, the incidence of congenital malformations in the present study was comparatively low than in other parts of India and abroad.

The present study has helped us to know the pattern and frequency of musculoskeletal system malformations and other anomalies associated with them, which will help us to plan future strategies for prevention, early diagnosis & timely management of these cases.

## AUTHOR CONTRIBUTIONS

**H C Sarangsa-** Collection of data, preparation of manuscript,

**K K Biswas-** Preparation of the manuscript, data collection and concept of the study.

**B Rabha-** Collection of data and Interpretation

**S Pal-** Manuscript Review and interpretation

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