Study of Incidence of congenital musculoskeletal malformations and its relation to maternal age, parity and birth weight of the newborn

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Abstract

Objectives: The present study was undertaken

1. To know the incidence of congenital musculoskeletal malformations in neonates
2. To assess the relation of congenital musculoskeletal malformations with maternal age, parity and birth weight of the newborn
3. To compare the data obtained with other studies

Materials and Method: The data for the study was collected from the department of Obstetrics and Gynecology and Paediatrics, Cheluvamba Hospital, attached to Mysore Medical College and Research Institute, Mysore. The study was done for a period of one year during which period, thirty seven babies with musculoskeletal malformations were diagnosed.

Results: The incidence of congenital musculoskeletal malformation in our study was 2.9/1000 births. 43.3% of cases were observed in mothers belonging to age group of 21-25 years. Malformations were more common in first and second para (45.9% each) and declined with increased parity.43.2% of the babies showed anomalies with birth weight above 2501grams.

Conclusion: The above study revealed the incidence of congenital musculoskeletal malformation and musculoskeletal malformations were common in mothers of younger age group, common in first and second para and significant number of babies were born with birth weight above 2501grams.

Conclusion: The above study revealed the incidence of congenital musculoskeletal malformation. Musculoskeletal malformations were common in newborn babies of mothers of younger age group of 15-20years, common in babies of mothers of first and second para and significant number of malformations were found in babies born with birth weight weighing above 2501grams

Keywords: Congenital, Musculoskeletal, Malformation, Parity, Neonates, Birth weight

Received: 2nd June, 2017 **Accepted:** 8th August, 2017

Introduction

Congenital malformations is a physical defect present at birth, irrespective of whether the defect is caused by a genetic factor or by events existing before or at birth. In a malformation, the development of a structure is arrested, delayed or misdirected early in embryonic life and the effect is permanent.(1)

Any defect or malformation which is present at birth is called congenital malformation. Malformation is caused by disturbances in the development of a structure in early embryonic period and it results in permanent deformity.(1)

For centuries, skeletal deformities have fascinated many civilizations. For example, the Egyptian God path was depicted as a short limbed dwarf and metabolic bone disorders were recognised in early Christian times.(2)

Polydactyly is evident in a hand print described from the Hal Saflieni Hypogeum.(3)

Polydactyly is an anomaly of having more than five digits in hand or foot and has been described in a hand print from Hal Saflieni Hypogeum.(3)

Major structural anomalies occur in 2-3% of live born infants, and 2-3% are recognised in children by age of 5 years for a total of 4-6%.

2-3% of live born infants show major structural anomalies and around 2-3% of congenital anomalies are found in the age group of children of less than five years accounting for a total of 4-6%.(4)

Malformations are caused by environmental and or genetic defects acting independently or in concert. Malformations have their origin during the third to eight weeks of gestation.(4)

The congenital malformations can involve many different systems including musculoskeletal system. In some of the studies conducted to know the incidence of congenital malformations, musculoskeletal malformations have stood first.

Experimental embryology has revealed a wide array of environmental agents capable of affecting normal development. Thalidomide (a sedative and anti-nauseant) drug consumed during pregnancy has caused Phacomelia, Congenital heart disease, anal stenosis and atresia of external auditory meatus in Germany. A congenital malformation need not necessarily be caused by a single etiological factor as some defects are purely

genetically determined whereas others are the result of environmental factors like maternal medication, infection or irradiation. Majority of them are the result of complex interaction of genetic and environmental factors.

Recent advances in the science of prenatal diagnosis allow for the evaluation of an affected embryo or an abnormal cell line prior to gestation within the womb via pre-implantation diagnosis. The technique can be used for any genetic condition which can be detected with a chromosome specific probe.

Prenatal diagnosis plays a very important role in the detection of an affected embryo. Abnormal cell line and any genetic disorders can be detected using specific chromosome probes in an embryo before it is implanted in the uterus.[5]

The risk assessment for abnormal foetus is a part of prenatal care and should begin prior to conception and continue in the antepartum period.[5]

The awareness of incidence, prevalence and patterns of malformations can help medical and paramedical personnel to identify cases at risk and plan appropriate and effective intervention in treatment to prevent morbidity and mortality. Hence, the present study was undertaken with a view to assess the incidence of congenital musculoskeletal malformations and to analyse the magnitude of the problem.

Materials and Method

This cross-sectional descriptive study was carried out by collecting the data from the department of Obstetrics and Gynecology, and Paediatrics, Cheluvamba Hospital, attached to Mysore Medical College and Research Institute, Mysore. The study was done for a period of one year from October 2011 to September 2012, during which period 12,753 new born babies were examined. All births after 28 completed weeks were examined for congenital musculoskeletal malformations within 24-48 hours after birth with a written consent from parents/relatives. The details regarding the maternal age, parity, birth weight of the newborn were recorded as per the proforma. After birth babies with musculoskeletal malformations were examined clinically. Photographs and radiographs were taken in necessary situation.

Inclusion criteria

1. All births after 28 completed weeks (live birth and still births) with musculoskeletal malformations were included.
2. Babies who have undergone surgery for the correction of musculoskeletal defects during early neonatal period were included.
3. Babies with other system anomalies along with musculoskeletal malformations were included

Exclusion criteria

1. Babies born before twenty eight completed weeks were excluded from the study.

Statistical method applied: Descriptive statistics, Chi-square test for goodness of fit.

Results

Out of 12,753 births, 37 cases of congenital musculoskeletal malformations were observed with an incidence of 2.9/1000 births, of which 21 cases (56.8%) were male, 16 cases (43.2%) female with male to female ratio of 1.31:1. In these 37 cases, a total of 40 malformations were noted with an incidence of 3.2/1000 births.

Total births - 12,753

No. of cases with congenital musculoskeletal malformation - 37

Total No. of musculoskeletal malformations in 37 cases – 40

(Table 1 shows the incidence of various musculoskeletal malformations)

Maternal age; Out of 37 cases, 16 cases (43.3%) of musculoskeletal malformations were distributed in the maternal age group of 21-25 years, whereas 10 cases (27%) each were distributed in the age group of 15-20 years and 26-30 years respectively. Only one case was born to an elderly primi (2.7%). (Fig. 1)

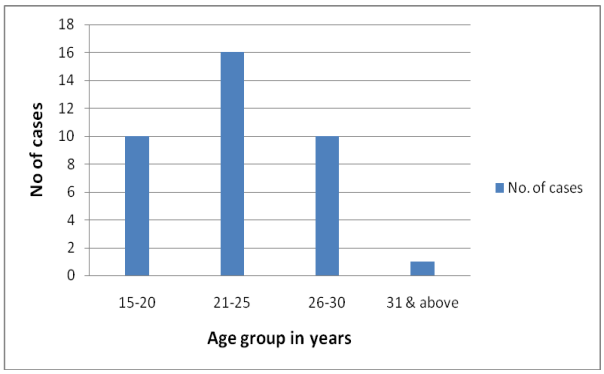

Fig. 1: Distribution of cases according to maternal age

The highest occurrence of cases were distributed in the age group of 21-25 years (43.3%) and the difference was statistically significant (P=0.006).

Parity: Out of 37 cases, we observed 17 cases (45.9%) each belonged to para I and II, 2 cases (5.4%) to para III and 1 case (2.7%) to para IV. The difference in the distribution of cases in different parity was statistically significant. (Chi-square test for goodness of fit, P=0.001) (Table 2).

Birth weight of the babies: Out of 37 cases, significant number of babies (16 babies - 43.2%) were found in the birth weight range of 2,501 gm and above constituting 43.2% (p=0.006). The number of cases increased with increase in the birth weight of babies. Only 3 babies had the birth weight <1000 gm. (Table 3) Babies weighing less than 2.5 kg are considered as low birth weight babies. In our study, most of the babies born

with congenital musculoskeletal malformations had normal birth weight.

Discussion

The incidence of congenital musculoskeletal malformations in our study revealed 2.9/1000 births. In India, the incidence of congenital musculoskeletal malformations varies between 1.5/1000 births in Baroda[6] to 13/1000 births in Jammu.[7] Worldwide the incidence of congenital musculoskeletal malformations varies between 1.6/1000 births in Iraq[8] to 31/1000 births in Nigeria.[9]

The incidence in our study is quite consistent with other studies reported by Hussein AJ[8] (2.5/1000) and Tan KL et al.[10] (3.5/1000). Present study is in agreement with previous studies by Menasinkai SB[11] in the same hospital with an incidence of 2.3/1000 births.

Some authors have reported higher incidence of congenital musculoskeletal malformations: Tayebi N et al.[12] (14.2/1000), Simpkiss and Lowe[13](16.5/1000) and Oyinbo CA[9] et al. (31/1000). In India, Singh A[14](8.05/1000), Bhat BV[15] (9.69/1000), Gupta RK et al.[7] (13/1000) have found higher incidence of congenital musculoskeletal malformation.

A comparative Table 4 and 5 shows incidence rates of congenital musculoskeletal malformations at different regions within India and abroad.

The variability in the incidence of malformations reported may be due to the following reasons:

1. The difference in the real magnitude of population and source of statistics.
2. Experience of the person diagnosing the anomaly.
3. Maturity of the foetus at the time of birth.
4. Whether the source of data includes only live born or still born babies.
5. The spectrum of investigations carried out in confirming the diagnosis.

Table 1: Incidence of various musculoskeletal malformations

Malformation	Number of malformations in 37 babies	Incidence per 1000 births
CTEV	24	1.88
Polydactyly	2	0.15
Kyphoscoliosis	2	0.15
Spinabifida	2	0.15
Oligodactyly	2	0.15
Short fingers and toes	1	0.07
Osteogenesis imperfect	1	0.07
Hypoplastic phalanges	1	0.07
Congenital dislocation of hip	1	0.07
Syndactyly	1	0.07
Rhizomelic dwarf	1	0.07
Gastroschisis	1	0.07
Achondroplasia	1	0.07
Total	40	

Table 2: Distribution of cases according to parity

Parity	No. of cases	Percentage
P1	17	45.9
P2	17	45.9
P3	2	5.4
P4	1	2.7
Total	37	100

Table 3: Birth weight of babies with musculoskeletal malformation

Weight (grams)	No. of cases	Percentage
<1000	3	8.1
1001-1500	4	10.8
1501-2000	6	16.2
2001-2500	8	21.6
2501 and above	16	43.2
Total	37	100

Table 4: Comparison of incidence of congenital musculoskeletal malformations in India

SI. No.	Author	Region	Year	Incidence (per 1000 births)
1.	Gupta S[6]	Baroda	2012	1.5
2.	Swain S et al.[16]	Varanasi	1994	1.7
3.	Menasinkai SB[11]	Mysore	2011	2.3
4.	Present study	Mysore	2012	2.9
5.	Datta V et al.[17]	Maharashtra	2000	3.2
6.	Taksande A et al.[18]	Maharashtra	2010	3.8
7.	Verma M et al.[19]	Ludhiana	1991	6.2
8.	Singh A et al.[14]	Jammu	2009	8.05

9.	Bhat BV et al.[15]	South India	1998	9.69
10.	Gupta RK et al.[7]	Jammu	2003	13

Table 5: Incidence of congenital musculoskeletal malformations in different regions

SI. No.	Author	Region	Year	Incidence (per 1000 births)
1.	Hameed NF[20]	Baghdad, Iraq	2007	1.6
2.	Hussein AJ[8]	Diwaniyah, Iraq	2009	2.5
3.	Present study	Mysore	2012	2.9
4.	Tan KL et al.[10]	Singapore	1996	3.5
5.	Dolk H et al.[21]	Europe	2010	3.8
6.	Fida NM[22]	Western Saudi-Arabia	2007	4.1
7.	Muga RO[1]	Kenya	2009	5.2
8.	Temtamy et al.[23]	Egypt	1998	6.4
9.	Ali A et al.[24]	Ahwaz, Iran	2008	7.9
10.	Karbasi SA et al.[25]	Yazad, Iran	2009	8.3
11.	Tayebi N et al.[11]	Yazad, Iran	2010	14.2
12.	Simpkiss and Lowe[13]	Uganda	1968	16.5
13.	Oyinbo CA et al.[9]	Nigeria	2009	31

Table 6: Shows incidence of various musculoskeletal malformations in different studies

(Fig. in the table indicate incidence per 1000)

Author	Syndactyly	Achondroplasia	Osteogenesis Imperfect	Polydactyly	CDH	Spina bifida	Rhizomelia
Present study	0.07	0.07	0.07	0.15	0.07	0.15	0.07
Muga RO[1]	0.4	0.81	0.1	10.2	-	-	-
Simpkiss M[13]	-	-	-	13.5	-	-	-
Van meerdervoort HFP[32]	-	-	-	8.8	1.5	0.7	-
Verma M et al.[19]	0.9	-	0.1	0.9	-	-	-
Swain S et al.[16]	-	-	0.3	-	0.3	-	-
Bakare TIB et al.[31]	1.6	-	-	14.2	-	1.6	-
Gupta RK et al.[7]	-	-	-	2.5	2.5	2	-
Tayebi N et al.[12]	5.02	-	-	5.1	2.5	-	-
Ahuka OL et al.[27]	0.3	-	-	-	-	0.6	-
Ali Aet al.[24]	0.4	-		0.9	3.2	-	-
Singh A et al.[14]	-	-	-	2	-	2.4	-
Taksande A et al.[18]	1.3	-	0.21	1.3	-	-	-
Karbasi SA et al.[25]	1.8	-	-	4.3	1.8	-	0.6

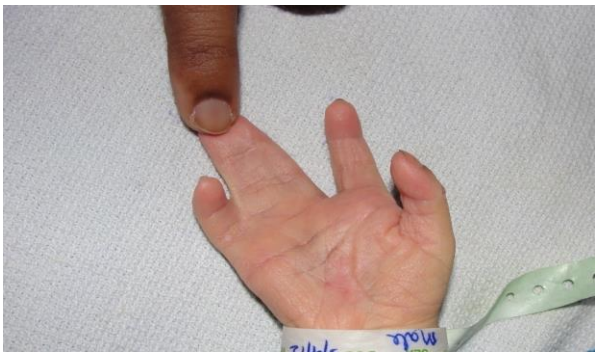

Fig. 2: Syndactyly of right hand showing complete syndactyly of 3rd& 4th digit, partial syndactyly of 4th& 5th digit

Fig. 3: Shows Polydactyly (six toes) in right foot and Oligodactyly(four toes) in left foot with bilateral clubfoot

Incidence of various musculoskeletal malformations: Altogether there were 40 malformations from 37 cases. The most common musculoskeletal malformation was Congenital talipes equino varus (CTEV) accounting for 24 cases (1.9/1000 births). The next common malformations were polydactyly, spina bifida, kyphoscoliosis, oligodactyly 2 cases each (0.15/ 1000 births). Other malformations were Achondroplasia, Rhizomelic dwarf, Short fingers and toes, Osteogenesis imperfecta, Hypoplastic phalanges, Congenital dislocation of hip, 1 case each (0.07/1000births).

The incidence of CTEV is estimated to be 1-2/1000 births.[26] In our study, the incidence of CTEV was 1.9 per 1000 births which is comparable with Ahuka OL[27] (1.1/1000), Samina Shamim[28] (2.32/1000) Tootoonchi[29] (2.6/1000) and Gupta RK et al.[7] (2.5/1000).The incidence of CTEV is higher in studies by Aigoro NF[30] (7.9/1000) and Hussein AJ[8] (9.68/1000). Some authors have documented lower incidence of CTEV: Simpkiss M[13] et al. (0.48/1000).

Most of the studies have documented polydactyly as the commonest congenital musculoskeletal anomaly. Our findings of polydactyly (0.15/1000) is close to study by Verma M et al.[19](0.9/1000). Muga RO,[1] Simpkiss M et al.[13] and Bakare TIB[31] have reported higher incidence of polydactyly 10.2/1000, 13.5/1000 and 14.2/1000 respectively. (Table 6)

The incidence of syndactyly in our studies is 0.07/1000 which is lower than other studies. Tayebi N et al.[12] found higher incidence of syndactyly (5.02/1000). Muga RO[1] reported Achondroplasia (0.81/1000 births) in 6 male babies. We found 1 case of Achondroplasia in male baby with an incidence of 0.07/1000 births. The incidence of Congenital Dislocation of Hip is low (0.07/1000) when compared to other studies by Ali A et al.[24] (3.2/1000), Gupta RK et al.[7] (2.5/1000) and Tayebi N et al.[12] (2.5/1000).

The incidence of osteogenesis imperfecta, Spina bifida and Rhizomelia is comparatively low.

Maternal age: In our study, highest frequency of malformations were found in the age group 21-25 years (16 cases-43.3%) and the difference was significant statistically (p=0.006). Gupta RK et al.[7] observed more babies with malformations distributed in the age group of 21-25 years (1.26%) and in 26-30 years (1.64%) but the difference was not significant statistically. Hameed NF[20] found musculoskeletaI malformations distributed in the maternal mean age of 28.69.

Study by Choudhury AR et al.[33] showed maximum number of CTEV cases distributed in the maternal age group of 25-30 years. Simpkiss M[13] reported distribution of polydactyly in the mean maternal age of 23.63.

Parity: In the present study, the incidence of malformations declined with increasing parity. Majority of the cases were seen in primi para and second para (17 cases each-45.9%) followed by third (2 cases-5.4%) and para four (1 case-2.7%). It shows that there is a strong association between first and second order of birth and occurrence of malformations.

But Gupta RK et al.[7] observed more cases of musculoskeletal malformations in para IV (7.14%) and para V (8.33%). Simpkiss M et al.[13] found highest number of polydactyly cases in the mean birth order of 2.

Weight of the baby: Birth weight is an index of the quality of prenatal care and a predictor of both survival and health of the baby. The present study revealed a higher incidence of congenital musculoskeletal malformation in babies weighing more than 2.501grams.

Some of the studies have documented the association of malformations in low birth weight babies. Muga RO[1] and Bahadur RA[34] found increased occurrence of musculoskeletal malformations in low birth weight babies. Gupta RK[7] et al. found an incidence of 2.38% in babies weighing <2.5 kg but the difference among two groups was not significant statistically.

Conclusion

The above study revealed the incidence of congenital musculoskeletal malformation and is comparatively low to other parts of India and abroad. Musculoskeletalmal formations were common in mothers of younger age group, common in first and second para and significant number of babies had normal birth weight. The significance of congenital musculoskeletal malformations lies not only in their contribution to mortality but also in causing disability and handicaps. One of the major steps in reducing the incidence of anomalies and proper management would be early detection. Medical termination of pregnancy for lethal type of congenital musculoskeletal anomaly will partially reduce the incidence of congenital malformations. For this proper antenatal care and a high degree of awareness is essential. Provision and development of services for early prenatal diagnosis by ultrasonography and amniotic fluid studies is important. Prenatal ultrasonography at about 14-16 weeks of pregnancy can detect most of malformations which can later be followed by adequate treatment at the earliest.

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