

Pattern of Congenital Anomalies of Musculoskeletal System in Newborns: A Hospital Based Study

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Abstract:

Background: The present study aims to study the incidence and pattern of musculoskeletal system anomalies in our region.

Material And Method: It is a cross-sectional study conducted in the Department of Obstetrics and Gynecology, SMGS hospital, GMC Jammu over a period of one year from October 2012 to September 2013. All women diagnosed antenatally with musculoskeletal system anomalies and those women having no antenatal record but after delivery, examination of baby revealed musculoskeletal system anomaly; were included in the study. Maternal and neonatal characteristics were recorded.

Results: 49 cases of musculoskeletal anomalies were diagnosed with an incidence of 3.2 per 1000 births. Club foot was the most common anomaly in musculoskeletal system followed by limb and spine deformities, polydactyly, facial dysmorphism, dolichocephaly, achondroplasia, bony dysplasias, deformed head in decreasing order of their frequency. These anomalies were almost equally present between males and females (p value= 0.88).

Conclusion: Congenital anomalies are emerging as an important perinatal problem contributing to still birth and infant mortality and morbidity. Musculoskeletal anomalies contribute to physical disabilities and their timely diagnosis and treatment can prevent these long term complications.

Keywords: Congenital, anomaly, musculoskeletal, club foot, male, female

I. Introduction

A congenital anomaly is an abnormality of structure, functions or body metabolism that is present at birth (even if not diagnosed until later in life) and results in physical or mental disability or is fatal¹. In the early part of 19th century, the percentage of deaths from congenital anomalies was relatively low. This was because preventive medicine, immunology and antibiotics were not in usage. Now the number of deaths from infections, metabolic and endocrinal disorders has decreased and so birth defects as a cause of perinatal mortality have come to the forefront.² It was also observed that better maternal care and improved standards of living have very little effect on the overall frequency of congenital malformations.^{3,4} In general, we cannot measure the incidence of congenital anomalies due to the prenatal loss of fetuses such as blighted ova, miscarriages and ectopic pregnancies.⁵ Various sources estimate the prevalence of congenital anomalies to be in the range of 1-3% of all live born infants and considerably higher for infants that are still born or spontaneously aborted.^{6,7,8} This rate increases to 5-6% when the ascertainment period is extended to the age of 5-6 years.^{6,8} clinical Manifestations Of Fetal Anomalies Include

- Miscarriages: 80% occur in the first trimester⁹
- Fetal death⁹
- Live birth with malformed neonates¹⁰
- Functional disorder : neurological, endocrine or metabolic^{11,9}

Congenital anomalies regardless of their cause can affect any organ or system of the body, yet some types of congenital anomalies are more common than the others. Most congenital anomalies can be classified under general categories according to their frequency as–musculoskeletal defects, congenital heart defects, digestive system disorder, circulatory system disorder, central nervous system disorder; urinary system disorder and genital organ defects.¹² Congenital anomalies are now etiologically considered as the outcome of intricate interaction between host and environment. This indicates the importance and urgency of epidemiological investigations in this particular field. The present study aims to study the incidence and pattern of musculoskeletal anomalies in northernmost part of our country as these anomalies are a major cause of physical disability and lifelong mental trauma.

II. Material And Method:

The present study was conducted in the Department of Obstetrics and Gynaecology, SMGS Hospital, GMC Jammu over a period of one year. All pregnant women who had ultrasound scans revealing anomalies of musculoskeletal system and those women whose babies after delivery revealed these anomalies, were included in the study. Maternal demographic data including age, parity, and residence were recorded. Neonatal characteristics such as gestational age at the time of delivery, birth weight, apgar score at birth, sex and type of anomaly were recorded. Statistical analysis was done using SPSS software and reported as percentages deemed appropriate for variables.

III. Results

During the study period, 49 cases of musculoskeletal system anomalies were detected out of 15447 births at SMGS Hospital in one year giving an incidence of 3.2 per 1000 births. Out of 49 cases, 24 affected fetuses were males (48.97%), 23 were females (46.94%) and 2 (4.08%) were having ambiguous genitalia. Thus, male to female ratio is 1.04:1 and the p value is 0.88, which is statistically not significant. Table 1 shows the number of cases in males and females

S.NO	CASES	NO	PERCENTAGE
1	MALE	24	48.97
2	FEMALE	23	46.94
3	AMBIGUOUS	2	4.08

Table 1: Number of cases and sex of newborns

Out of 49 cases diagnosed with urinary system anomalies, 13(26.53%) had other associated anomalies including central nervous system, cardiovascular, urinary, gastrointestinal systems and 36 cases(73.46%) had isolated musculoskeletal system anomalies.

With regard to individual malformation, club foot was the most common anomaly in musculoskeletal system followed by limb and spine deformities, polydactyly, facial dysmorphism, dolichocephaly, achondroplasia, bony dysplasias, deformed head in decreasing order of their frequency. Pattern of musculoskeletal anomalies is shown in table 2

Type of anomaly	Congenital anomalies			
	Male No.	Female No.	Ambiguous No.	Total No.(%) (n= 49)
Club foot	10	3	1	14(28.57)
Facial dysmorphism	1	2	0	3(6.12)
Deformed head	0	1	0	1((2.04)
Dolicocephaly	2	1	0	3(6.12)
Polydactyly/ Syndactyly	2	4	0	6(12.24)
Limb deformity	5	4	0	9(18.36)
Spine deformity	1	7	1	9(18.36)
Achondroplasia	1	1	0	2(4.08)
Bony dysplasia	2	0	0	2(4.08)
Total	24(48.97%)	23(46.94%)	2(4.08%)	49 (100)

Table 2: Pattern of musculoskeletal anomalies in newborns

Club foot was present in 14(28.57%) babies. It was present in 10 male babies as compared to 3 female babies (M: F=3.33:1). Limb deformities were present in 9 cases (18.36%), where 5 were male fetuses and 4 were female fetuses (M: F=1.25:1). Spine deformities were also present in 9 babies; 1 male ,7 female and one baby having ambiguous genitalia(M:F=0.14:1).Polydactyly was seen in 6 babies;2 males and 4 females(M: F=0.50:1). Facial dysmorphism was seen in 3 cases, out of which 1 was male and 2 were females (M: F=0.50:1). Dolichocephaly was also seen in 3 babies; 2 males and 1 female (M: F=2:1). Achondroplasia and bony dysplasias were present in 2 babies each. Deformed head was observed in one baby only.

In the present study, majority of anomalies were present in mothers between 26-30 years of age (55.10%), followed by 21-25 years of age (24.49%). With regard to parity, there was higher frequency of these anomalies in newborns of primiparas (42.86%) as compared to other women.

57.14% of anomalies were present in newborns of mothers residing in urban areas as compared to 42.85% of rural group. There was higher preponderance of anomalies of this system in fetuses with non cephalic presentation (60.25%) as compared to those with cephalic presentation (39.75%). Most fetuses with these anomalies were born to mothers having term pregnancy (65.46%), 3.2% to post-term pregnancies and 31.34% to preterm pregnancies.

Family history of congenital anomalies was present in two patients. One patient had previous baby born with anencephaly and other with gut atresia. There was history of oligohydramnios in four patients and polyhydramnios in one patient. 17 mothers did not take folic acid during antenatal period. History of pregnancy induced hypertension was present in one mother. Five newborns were from consanguineous marriages. There was no history of radiation exposure or drug intake.

IV. Discussion

In our study, the incidence of musculoskeletal defects was found to be 3.2 per 1000 births. These anomalies constituted 12.99% of total anomalies reported in the hospital. El-Koumi et al. (2013)¹³, Mohammed et al. (2011)¹⁴ reported musculoskeletal to be the commonest system involved. Agrawal D et al (2014)¹⁵ reported incidence of musculoskeletal anomalies to be 4.4 per 1000 births.

In current study, club foot was the most common anomaly present in 28.57% cases.

Mohammed et al. (2011)¹⁴ observed club foot constituting 28.20% of all musculoskeletal anomalies, followed by polydactyly (10.25%) in a prospective study of 5000 newborns in Egypt.

Taksande et al. (2010)¹⁶ reported that 72.2% of all musculoskeletal anomalies to be constituted by club foot and polydactyly in a study in central India.

In our study, musculoskeletal anomalies were equally observed among males and females, although significant heterogeneity between subtypes was present. Male predominance for club foot was seen as compared to females (10 males v/s 3 females). Miedzybrodzka (2003)¹⁷ also observed similar results.

With regard to maternal age, no definite pattern was observed in present study. Higher incidence of congenital anomalies was present among non cephalic presentations (60.25%) as compared to cephalic (39.75%), in current study. It is well recognized that a fetus presenting by breech is more likely to have a congenital malformations than a fetus with cephalic presentation as observations made by Shawky and Sadik (2011)¹⁸. No significant relation of congenital anomalies with location of residence was observed in the present study. In our study, amniotic fluid abnormalities were present in five mothers. **Shawky and Sadik(2011)**¹⁸ recorded increased frequency of congenital malformations in off springs of mothers who had hydraminos.

V. Conclusion

Congenital anomalies are emerging as an important perinatal problem contributing to still birth and infant mortality. They also lead to emotional upset and social stigma to parents, which is beyond the limit of our imagination. Inherited and chromosomal anomalies are associated with loss of physical, mental and/or intellectual abilities. To decrease the incidence of various congenital anomalies and their prevalence in the particular region, it is important that the distribution and prevalence of congenital anomalies are identified in that region and country as a whole. The present study aims to study the pattern and incidence of musculoskeletal anomalies in our region which is the northernmost part of the country having different geographical and racial background. Data is important as these anomalies are mostly a cause of physical disability and can be corrected to a large extent.

References

March of Dimes Resource Center. Birth Defects 1998. Available from: www.modimes.org

- [1]. Gupta S, Gupta P, Soni J. A study on incidence of various systemic congenital malformations and their association with maternal factors. *Nat J Med Res* 2012; 2 (1):19-21
- [2]. Carter CO. Congenital malformations. *WHOChr* 1967; 21: 287
- [3]. Nelson MM, Forfar JG. Congenital malformations at birth. *Dev Med Child Neurology* 1969; 11: 13
- [4]. Andrew EC. Birth defects are preventable. *Int J Med Sci* 2005; 2 (3): 91-92
- [5]. De Santis M, Straface G, Carducci B, et al. Risk of drug-induced congenital defects. *Eur J Obstet Gynecol Reprod Biol* 2004; 117 (1): 10-19
- [6]. Kalter H. Teratology in the 20th century: environmental causes of congenital malformations in humans and how they were established. *Neurotoxicol Teratol* 2003; 25 (2): 131-282
- [7]. Schumacher GH. Teratology in cultural documents and today. *Ann Anat* 2004; 186 (5-6): 539-46
- [8]. Stamatin F. Diagnostical prenatal al malformatiilor congenital. In: Muntaneu I (editor), *Tratat de obstetrica*. Ed Acad 2000: pp. 1323-55
- [9]. Gorduza EV. Screening prenatal al anomalilor congenitale. *Viatamedicala Nr* 2006; 36: 8-9
- [10]. Sadler TW. *Embriologie medicala*. Editia 10th edition. Med Callisto 2007.
- [11]. Birth Defect Prevalence in Canada, 1995. In: Laboratory Centre for Disease Control (CCASS, editor); Health Canada, 1997
- [12]. El-Koumi MA, Al-Banna EA and Lebda I. Pattern of congenital anomalies in newborns: A hospital-based study. *Pediatr Rep* 2013 ; 5 (1):20-23
- [13]. Mohammed YA, Shawky MR, Soliman AS, Ahmed MM. Chromosomal study in newborn infants with congenital anomalies in Assiut University Hospital: Cross-sectional study. *Egyptian J Med Hum Genet* 2011; 12: 79-90
- [14]. Agrawal D, Mohanty BB, Sarangi R, Kumar S, Chinara PK, Mahapatra SK. Study of Incidence and Prevalence of Musculoskeletal Anomalies in A Tertiary Care Hospital of Eastern India. *Journal of Clinical and Diagnostic Research*. 2014;8(5):AC04-AC06.
- [15]. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in central India: A rural medical college hospital-based data. *Indian J Hum Genet* 2010; 16 ss(3): 159-63

- [16]. Miedzybrodzka Z. Congenital Talipes equinovarus (clubfoot): A disorder of the foot but not the hand. *J Anat* 2003; 202 (1): 37-42
[17]. Shawky RM, Sadik DI. Congenital malformations prevalent among Egyptian children and associated risk factors. *Egyptian J Med Hum Genet* 2011; 12: 69-78



FIGURE 1: CLUB FEET



FIGURE 2:FUSED LIMBS