Profile of gross congenital malformations among live newborns and its associated risk factors from a tertiary care rural teaching institute

Ashish Marwah
Assiastant Professor, Department of Pediatrics, BPS G.M.C for women, Khanpur Kalan, Sonepat

Research Article

Article Info:
Received on: 14/08/2015
Published on: 30/04/2016

QR Code for mobile

Literati Open Access

ABSTRACT :

Objective: To study the profile of gross congenital malformations among live newborns and its associated risk factors.

Material and methods: All the babies born in this hospital (live births) during January 2014 to December 2014 formed the baseline population and those with congenital malformations were included in the study. Antenatal records were assessed and mothers were interviewed for socio-demographic variables.

Study design: Cross- sectional, descriptive.

Results: There were 6143 live births out of which 109 had gross congenital malformations giving an incidence of 1.7%. Central nervous system was most commonly affected (48%) followed by musculoskeletal (28%) and gastrointestinal system (13%). Incidence of congenital malformations was more among multipara females, preterm and low birth weight babies.

Conclusion: A relative increase in the incidence of congenital malformations once again implies the importance of public health education and regular antenatal screening. We also, strongly recommend mandatory screening of all newborns by pediatrician after birth for timely detection and best possible treatment of these congenital malformations.

Keywords: congenital malformation, newborn, live birth, antenatal, genetic.

INTRODUCTION:

Congenital malformations are defects in morphogenesis during early fetal life. [1] Although, its etiology is unknown in 50% of cases, genetic (30-40 %) and environmental (5-10 %) form the majority of known causes. [2] Malformations may range from minor abnormalities to major structural defects. Minor abnormalities involve non vital organs with little or no functional effects and there is no urgency for their correction whereas major anomalies impair function or are of significant cosmetic value. These may even be life threatening requiring immediate medical/surgical treatment. Congenital malformations are the fourth major cause of neonatal mortality in India after prematurity, neonatal sepsis and birth asphyxia. [3] With gradual reduction in mortality due to other causes in view of improvement in perinatal and neonatal care, there has been an increase in the proportion of deaths due to congenital malformations. Global incidence of congenital malformations is 3-7 %, however, ethnic and geographic variations exist between countries. [4] In India, 2.5 % infants are affected at birth accounting for 8-15 % of perinatal and 13-16 % of neonatal mortality. [5] The incidence rises to 5 % if anomalies detected later in childhood such as heart, kidney, lung and spine are included.[6] Apart from this, congenital anomalies result in approximately 3.2 million birth defect related disabilities every year. [3] Minor anomalies visible to naked eye may sometimes be associated with serious underlying major defects such as heart defects, neural tube defects and renal anomalies which if detected and treated timely can go a long way in reducing morbidity and mortality in later life. Also, parental counseling done at this stage for future prenatal testing would be more valuable as parents are more receptive at this stage. Since, there is a relative paucity of literature on congenital malformations in newborns in our country; we therefore, planned this study to present the incidence and pattern of congenital malformations in newborns at birth and to identify its possible risk factors from a tertiary care teaching hospital of north India.

METHODS:

After approval from institute's ethics committee, a cross sectional descriptive study was conducted in the newborn care unit of a tertiary care teaching hospital. All the babies born in this hospital (live births) during January 2014 to December 2014 formed the baseline population. All the live newborns were examined within 48 hours of birth systematically by a pediatrician to detect any malformations and those babies with any gross malformation were enrolled in the study after taking a written informed consent from the parents. Still births were excluded from

***Corresponding author:**
Dr. Ashish Marwah,
House no. 1457, Sector-9,HUDA, Ambala City (Haryana).
Email:ashmar1980@gmail.com
Contact:09996797731

Conflict of interest: Authors reported none

the study. The distribution of all malformations and other relevant history (by interview of parents and from maternal records) with regard to maternal age, birth weight, parity, consanguineous marriage, drug intake, any chronic disease was recorded in a predesigned performa. Diagnosis of congenital malformation was based on clinical examination of the newborn by the pediatrician. Further evaluation of babies including X-ray, ultrasonography of viscera, skull and 2D-echocardiography (as indicated) were done to rule out any other associated anomalies. Genetic and chromosomal analysis could not be done due to its non-availability in our hospital. All babies with birth defects were advised appropriate treatment/surgery and genetic counseling was given to parents of the affected newborns.

RESULTS:

During the study period, there were 6143 live births including 245 still births. Among them, 109 had gross congenital malformations resulting in an incidence of 1.7 %. Out of these, 94 babies had single and 15 babies had multiple congenital malformations. Table 1 summarizes the pattern of congenital malformations in the newborns. The most common system involved was central nervous system (48 %) followed by musculoskeletal (28 %) and gastrointestinal (13 %). As cardiovascular anomalies usually present late after birth, only 8% babies were found affected with cardiovascular anomalies. The maternal and fetal factors associated with congenital anomalies at birth are as shown in Table 2. Most of the mothers were in the age group of 20-35 years age group and incidence of congenital malformations was 1.49 % in < 20 years, 1.77 % in 20-35 years and 2.12 % in > 35 years. Risk of congenital anomalies showed an increasing trend with increasing parity. Cases of congenital anomalies were found in 2.27% of multi-para females and 1.63 % of primipara females. Consanguinity was observed in 12 cases out of which 2 were affected. Among fetal factors, males (2.0 %) were found to be affected more than females (1.43 %). Preterm (3.86 %) had a higher incidence of malformations as compared to term newborns (0.47 %).

DISCUSSION:

With improved control of infections and nutritional deficiency diseases, congenital malformations are becoming an important cause of neonatal and infant mortality worldwide.[3] The detection of birth defects in infants has increased during the antenatal and neonatal period due to advanced diagnostic techniques especially ultrasound.

Congenital malformations account for 6.6% of neonatal deaths in the rural as well as urban slum communities as reported by a national collaborative community based study by ICMR. [7] The incidence of congenital malformations in our study was found to be 1.7 per 1000 live births which is consistent with studies conducted in various parts of our country.[1, 8-14] The reported incidence in various studies is around 2% except the study conducted by Marwah et al who have reported a higher incidence of malformations (4.44%) in Punjab.[15] This could be due to increasing number of referrals coming to the hospital, inclusion of minor anomalies, racial, geographic and ethnic factors. Most common system involved in our study was central nervous system (48%) followed by musculoskeletal (28%) and gastrointestinal (13%) which is similar to the observations of other studies.[1,8,10,12,15,16] However, Sarkar et al reported that musculoskeletal anomalies were the commonest.[9] Cardiac anomalies were reported to be most common by some studies.[11,14,17] This could be due to routine echocardiography in babies of high risk mothers leading to early detection of cardiac anomalies which otherwise present late. In our study, males (2%) were found to be more commonly affected as compared to females (1.43%) which is similar to the observations of Sarkar et al and Taksande et al while Sachdeva et al found higher incidence of malformations in females as compared to males. [8, 9, 12] Swain and Marwah et al found no significant difference among males and females. [10, 11]

Table 1: Classification of congenital anomalies in newborns (n=109)

System	No%
Central Nervous System (48%):	
Anencephaly	3(2.75)
Encephalocele	5(4.58)
Spina bifida	11(10.09)
Meningo-myelocele	28(25.68)
Microcephaly	2(1.83)
Hydrocephaly	3(2.75)
Musculoskeletal (28%):	
CTEV	15(13.76)
Short limbs(Amelia, Meromelia)	2(1.83)
Polydactyly	1(0.91)
Sternocleidomastoid Tumor	3(2.75)
Calcaneovalgus	3(2.75)
Absence of depressor anguli oris	2(1.83)
CDH	4(3.66)
Gastrointestinal System (13%):	
Cleft lip and palate	5(4.58)
Congenital tooth	3(2.75)
Imperforate anus	2(1.83)
Exomphalos	1(0.91)
Tracheoesophageal fistula	2(1.83)
Mal-rotation of gut	1(0.91)
Congenital diaphragmatic hernia	1(0.91)
Laryngomalacia	2(1.83)
Urogenital system:	
Hypospadias	3(2.75)
Hydronephrosis	3(2.75)
Posterior urethral valve	1(0.91)
Micro-penis	1(0.91)
Congenital hydrocele	1(0.91)
Ear and neck:	
Ear anomalies	2(1.83)
Chromosomal anomalies:	
Down's syndrome	5(4.58)
Pierre Robin syndrome	2(1.83)
Cardiovascular system:	
Patent ductus arteriosus	4(3.66)
Cyanotic CHD	3(2.75)
Single umbilical artery	2(1.83)
Skin:	
Pre-auricular skin tag	3(2.75)

Maternal factors

Incidence of congenital malformations was found to be

higher among mothers aged more than 35 years in our study (2.12 %) but the difference was not statistically significant which is similar to the observations of other studies in India.[8-12,15]Among multipara females, incidence of congenital malformations was found to be high (2.27%) as compared to primipara females (1.63 %) but the difference was statistically insignificant. The findings are similar to the findings of other studies. [8-12, 15]

Table 2: Risk factors (Maternal and Fetal).

Risk factors	Total	Malformed (%)	p value
Maternal:			
Age:			
a.< 20 years	1139	17 (1.49)	
b.20-35 years	4108	73 (1.77)	
c. > 35 years	896	19 (2.12)	0.5679
Parity:			
< 4	4783	78 (1.63)	
> 4	1360	31 (2.27)	0.1168
Consanguinity:	12	2 (16.67)	0.371
Fetal:			
Sex:			
a. Male	3642	73(2.00)	
b. Female	2501	36(1.43)	0.1055
Birth weight:			
a. <2.5kg	1963	43(2.19)	
b. ≥2.5kg	4180	66(1.57)	0.0966
Gestation:			
a.<37wks	2356	91(3.86)	
b.>37wks	3787	18(0.47)	<0.0001

Fetal factors

Incidence of malformations was found to be higher in low birth weight babies (<2.5kg) as compared to babies with weight appropriate for age but the difference was not statistically significant. This is consistent with the findings of other studies.[8-12] This could be due to non inclusion of still births in the study. Higher incidence of congenital malformations was observed among preterm babies (3.86%) as compared to term babies (0.47%) and the difference was statistically significant (P < 0.05). This is consistent with the studies from other parts of the country.[1,8,9,12,15] Babies with history of consanguinity had higher incidence of malformations as compared to those with no such history but the difference was not statistically significant which is similar to the findings of Patel et al, Sarkar et al, Marwah et al and Taksande et al while Parmar et al found no association of consanguinity with congenital malformations.[1,9,11,15,18]

Our study had limitations of being an exclusive hospital based study accounting for mostly referred patients. Larger population based studies are required to determine the actual prevalence of these disorders in the population. Also, certain complex disorders could not be diagnosed for the lack of sophisticated genetic testing/ karyotyping at our centre.

The study highlights the incidence and pattern of congenital malformations and their associated risk factors from a tertiary care referral centre in north India. The study stresses the importance of thorough clinical examination of newborn at birth to detect congenital malformations early offering the baby best survival opportunity and decreasing morbidity in later life. We also strongly recommend public health education and compulsory premarital and antenatal counseling for the prevention and timely detection of these congenital malformations.

REFERENCES

1. Patel ZM, Adhia RA. Birth defects surveillance study. Indian J Pediatr 2005; 72:489-91.
2. Rajangam S, Devi R. Consanguinity and chromosomal abnormality in mental retardation and multiple congenital anomalies. J Anat Soc India 2007; 56:30-3.
3. Shamnas M, Arya PS, Viji A Thottumkal and MG Deepak. Congenital anomalies: A major public health issue in India. IJPCBS 2013; 3 (3):577-85.
4. Park K. Congenital malformations. In K Park (ed.) in Park's Textbook of Preventive and Social Medicine. 15th edition. 2005. Pp.379-80.
5. Merchant SM. Indian Council of Medical Research: Annual Report. Bombay: Genetic Research Centre; 1989.
6. Shah K and Pensi CA. Study of incidence of congenital anomalies in newborn. Gujarat Medical Journal 2013; 68 (2):97-9.
7. A national collaborative study of identification of high risk families, mothers and outcome of their offspring with particular reference to the problem of maternal nutrition, low birth weight, perinatal and infant morbidity and mortality in rural and urban Slum communities. Indian Council of Medical Research, New Delhi, 1990, pp 119-26.
8. Sachdeva S, Nanda S, Bhalla K, Sachdeva R. Gross congenital malformation at birth in a government hospital. Indian J Public Health 2014; 58: 54-6.
9. Sarkar S, Patra C, Dasguptamk, Nayek K, Karmakar PR. Prevalence of congenital anomalies in neonates and associated risk factors in a tertiary care hospital in eastern India. J Clin Neonatol 2013; 2(3): 131-4.
10. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. Indian Pediatr 1994; 31:1187-91.
11. Taksande A, Vilhekar K, Chaturvedi P and Jain M. Congenital malformations at birth in Central India: A rural medical college hospital based data. Indian J Hum Genet 2010; 16 (3):159-63.
12. Grover N. Congenital malformations in Shimla. Indian J Pediatr 2000; 67 (4):249-51.
13. Chaturvedi P and Banerjee KS. An epidemiological study of congenital malformations in newborn. Indian J Pediatr 1993; 60:645-53.
14. Kumar V, Singh A and Marwaha RK. An epidemiological study of congenital malformations in rural children. Indian Pediatr 1994; 31 (8): 909-14.
15. Marwah S, Sharma S, Kaur H, Gupta M, Goraya SPS. Surveillance of congenital malformations and their possible risk factors in a teaching hospital in Punjab. Int J Reprod Contacept Obstet Gynecol 2014; 3 (1):162-7.
16. Gupta S, Gupta P and Soni JS. A study on incidence of various systemic congenital malformations and their association with maternal factors. NJMR 2012; 2:19-21.
17. Annual report 2002-03. Indian Council of Medical Research, New Delhi. Reproductive health. p.91.
18. Parmar DA, S.P. Rathod D, Patel DSV and S.M. Patel D. A study of congenital anomalies in newborn. NJIRM 2010; 1 (1):13-7.

Cite this article as:
Ashish Marwah. Profile of gross congenital malformations among live newborns and its associated risk factors from a tertiary care rural teaching institute . Asian Journal of Biomedical and Pharmaceutical Sciences, 6(55), 2016, 17-19.