Pattern of congenital anomalies and risk factors in newborn in a city of a developing country: An observational study

Mithlesh Dewangan^{1,*}, S. Manazir Ali², Uzma Firdaus³

¹Senior Resident, ²Professor, ³Assistant Professor, Neonatalogy Section, Dept. of Pediatrics, Aligarh Muslim University, Aligarh, UP

***Corresponding Author:** Email: dewangan.mithlesh@rediffmail.com

Abstract

Congenital anomalies are important causes of childhood death, chronic illness and disability in many countries. Congenital anomalies also have long-term disability. It has significant impacts on individuals, families, health-care systems and societies. Data are limited and various factors which result in congenital anomalies can be prevented. This hospital based cross-sectional observational study which was conducted in a Neonatology Section, Department of Pediatrics, J N Medical College, Aligarh Muslim University, Aligarh, UP from January 2015 to December 2015. The incidence of congenital malformation was 2.5%. Majority of the congenital anomalies was associated with musculoskeletal system (58%) followed by cardiovascular (28%) and genitourinary (18%). Least common is respiratory (2%) and gastrointestinal (7%). The present study concluded that congenital anomalies in newborns were associated with maternal factors like maternal age, consanguinity and history spontaneous abortion. Early diagnosis and management can result in better outcome of these newborns with congenital anomalies.

Keywords: Congenital Anomalies, Risk Factor, Congenital Malformations, Birth Defect, Newborn.

Introduction

Congenital anomalies are important causes of childhood death, chronic illness and disability in many countries. Worldwide 276 000 babies die within neonatal period every year because of congenital anomalies.⁽¹⁾ Congenital anomalies also have long-term disability. It has significant impacts on individuals, families, healthcare systems and societies. Heart defects, neural tube defects and Down syndrome are some of the most common severe congenital anomalies. It is quite difficult to know the exact causes of congenital anomalies. Several factors are contributing, may be genetic, infectious, nutritional or environmental. Some congenital anomalies can be prevented. For example, vaccination, adequate intake of folic acid or iodine through fortification of staple foods or provision of supplements, and adequate antenatal care are keys for prevention. Every year 3rd march World birth defect day is observed.

In India we are more concern with mortality and morbidity due to low birth weight, birth asphyxia and sepsis than congenital anomalies. Government is also putting all efforts in these three prominent causes of neonatal mortality. Although Government of India through Rashtriya Bal Swasth Karyakaram (RBSK) setting District early intervention Centre for prompt recognition of congenital anomalies and treatment, but still we are very far from ground reality. Limited data are available at local level about the prevalence and incidence of congenital anomalies for early intervention. Various maternal factors are involved in congenital anomalies which can be prevented. So, the study of pattern of congenital anomalies at local level provides effective tool for necessary intervention. This present study is done with the aims and objectives to determine

the prevalence of clinically detectable congenital anomalies in newborns, their pattern and risk factors.

Material and Methods

This hospital based cross-sectional observational study which was conducted in a Neonatology Section, Department of Pediatrics, J N Medical College, Aligarh Muslim University, Aligarh, UP from January 2015 to December 2015. All live born babies with major congenital anomalies during the study period were included in the study. Still births were excluded from the study. All the live newborns with any congenital anomalies were included in the study after taking a written informed consent from the parents. The babies were examined soon after birth for the presence of major as well as minor congenital malformations. Baby's gestational age, birth weight, sex and symptoms in postnatal period were noted. After that meticulous general physical and systemic examinations of the babies were done. A complete medical, family, antenatal and personal history was taken. Information such as drugs, vaccines, acute and chronic diseases, vaginal bleeding in the first trimester of pregnancy, maternal residential place, maternal age, parental consanguinity, previous spontaneous abortions were recorded during the period of mother's hospital stay. Data were analyzed using the computer programs and Statistical Packages for Social Sciences (SPSS) version 23. Ethical clearance and approval to conduct this study was obtained from the Institutional ethics committee.

Observation and Results

A total of 5315 live births were occurred during this time period, out of which 139 were having congenital anomalies. The incidence of congenital malformation was 2.5%.

Gestational age	Term	93	66.9%
	Preterm	45	32.4%
	Post term	1	0.7%
Birth Weight	>2.5 Kg	72	51.5%
	2.5-1.5 Kg	53	38.5%
	1.5-1 Kg	11	8%
	<1 kg	3	2%
Gender	Male	79	57%
	Female	60	43%
Mode of delivery	Vaginal	75	54%
	LSCS	64	46%
Gravidity	Primipara	58	41%
	Multipara	81	59%
Maternal Age	>30 years	66	47%
	<30 years	73	53%
Religion	Hindu	53	38%
	Muslim	86	62%

Fable	1:	Baseline	Characteristi	c of	study	group
					•/	-

There were more term babies than preterm babies. Almost half of the babies were more than 2.5 kg. Male to female ratio was 4:3. 54% babies were delivered through normal vaginal route and 46% were delivered through Cesarean Section. Most common blood group was B +ve (34%).

 Table 2: Showing maternal risk factors in percentages

Maternal Risk factors	No. of	Percentage
	cases	
Consanguinity	22	15.8%
Maternal Diabetes	5	3.5%
Spontaneous Abortions	23	16.5%
Drugs	4	3%
Polyhydraminos	4	3%
Oligohydraminos	7	5%
Infections	10	7%
Others /unknown	64	46%

Among the maternal risk factors studied consanguinity (15.7%) and spontaneous abortion (16.3%) was the most common seen. Most of the congenital anomalies were associated with unknown risk factor.



Majority of the congenital anomalies was associated with musculoskeletal system (58%) followed by cardiovascular (28%) and genitourinary (18%). Least common is respiratory (2%) and gastrointestinal (7%).

Discussion

In our study incidence of congenital anomalies was 2.5% in live births. Other studies like Thaddane et al, $^{(2)}$ Datta et al,⁽³⁾ Swain et al,⁽⁴⁾ Taksande et al,⁽⁵⁾ Anand et al⁽⁶⁾ and Karla et al⁽⁷⁾ found incidence of congenital anomalies were1.23%, 1.24%, 1.2%, 1.91%, 2% and 1.98% respectively which was lower than our study. Some studies like Kumbhar et $al^{(8)}$ found 2.37%, Deshpande et $al^{(9)}$ 2.2%, Jain et $al^{(10)}$ found 2.69%, Francine R et al⁽¹¹⁾ showed 2.4% and El Koumi et al⁽¹²⁾ 2.5% which was comparable to our study. Jehangir et al⁽¹³⁾ observed the prevalence of 2.95%, Desai et al⁽¹⁴⁾ and Saifullah et al⁽¹⁵⁾ showed slightly higher incidence (3.6%) and Pabbati et al⁽¹⁶⁾ 4.08% which was more than our study. The actual incidence and occurrence of congenital anomalies depends on many factors i.e., it is multifactorial in nature. Various factors such as ethnicity, religion, population distribution, type of study, autopsy rate, and availability of diagnostic facility. In the present, low incidence as compared to some of the studies is due to inclusion of congenital anomalies of live birth.

In the present study majority of the congenital anomalies was associated with musculoskeletal system (58%) followed by cardiovascular (28%) and genitourinary (18%). Least common is respiratory (2%) and gastrointestinal (7%). Prashar et al,⁽¹⁷⁾ Pabbati et al⁽¹⁶⁾ and Singh et al⁽¹⁸⁾ also found high incidence of musculoskeletal system anomalies. Desai et al⁽¹⁴⁾ in their study observed that the most common system involved was musculoskeletal system (31.65%), followed by gastrointestinal (17.2%) and cardiac anomalies (16.46%). Deshpande et $al^{(9)}$ the most common system involved was musculoskeletal system (33.2%), followed by gastro-intestinal tract (GIT) (15%), CNS (11.2%), genitourinary (10.5%), cardiovascular system (9.1%), skin (8.7%) etc. Ndibazza J et al⁽¹⁹⁾ in their study revealed that the most commonly affected systems were the musculoskeletal (42.7 per 1000 births) and skin (16.1

per 1000 births). Sarkar S et al⁽²⁰⁾ found that the predominant system involved was Musculo-skeletal system (33.2%) followed by gastrointestinal (GI) system (15%) and central nervous system (CNS) (11.2%). El Koumi et al⁽¹²⁾ found musculoskeletal system was the most commonly affected (23%), followed by the central nervous system (CNS) (20.3%), gastrointestinal system (GIT) (16.2%), genitourinary system (13.5%), craniofacial (10.8%), cardiovascular system (CVS) (9.5%), and chromosomal anomalies (6.8%). Jain et al⁽¹⁰⁾ found that malformations of circulatory system was highest i.e. 29.6% followed by nervous and musculoskeletal system. Kumar et al⁽²¹⁾ reported that cardiovascular system were commonest (37%), followed by musculoskeletal (30%), gastrointestinal system (23%), central nervous system (13%) and genitourinary system (6.6%) and Takshande et al⁽⁵⁾ found that Cardiovascular malformations were most common in live births, followed by musculoskeletal malformations. The CNS defects were most commonly seen in still born.

In our study there were 48.5% low birth weight. Low birth weight was associated with increased risk of congenital malformations. It is due to congenital anomaly which retards the growth of a developing fetus. Some studies such as Thaddanne et al,⁽²⁾ Taksande et al,⁽⁵⁾ Karla et al,⁽⁷⁾ Desai et al⁽¹⁴⁾ and Saifullah et al.⁽¹⁵⁾

In our study more than 50% of women were below 30 years. Lower maternal age of ≤ 25 years and were at an increased risk of anomalies during pregnancy^(22,23) and this was reported in previous studies also such as Swain et al,⁽⁴⁾ Taksande et al,⁽⁵⁾ Desai et al⁽¹⁴⁾ and Sagunabai et al⁽²⁷⁾ have found statistically significant association of increased maternal age and congenital anomalies. Other studies such as Datta et al,⁽³⁾ Khanna et al⁽²⁶⁾ and Karla et al⁽⁷⁾ have found no statistically significant association between congenital malformation and maternal age.

In our study 15.8% had consanguineous marriage. Agarwal⁽²⁸⁾ and Desai et al⁽¹⁴⁾ found significant correlation between congenital malformation and consanguinity.^(23,24,25) In our study 16.5% congenital malformation had previous history of abortion. Similar findings were also obtained in the study of Saifullah et al.⁽¹⁵⁾ The mechanism responsible for such association is not known. The incidence of congenital anomalies was more in baby born to consanguineous couples because they express the homozygous genes inherited from their common ancestors.

Conclusion

The present study concluded that congenital anomalies in newborns were associated with maternal factors like maternal age, consanguinity and history spontaneous abortion. Early antenatal detection and genetic counseling can prevent many congenital anomalies. Regular antenatal checkup, Iron and folic acid supplementation and avoidance of teratogenic agents are key for prevent congenital anomalies. Early diagnosis and management should be done for better the outcome. Further management for the disability and rehabilitation of these babies should be done so that they can leave normal life.

References

- 1. WHO. Congenital anomalies. Fact sheet. updated Sep 2016.Available from http://www.who.int/mediacentre/factsheets/fs370/en.
- Thaddanee R, Patel HS, Thakor N. A study on incidence of congenital anomalies in newborns and their association with maternal factors: a prospective study. International Journal of Contemporary Pediatrics. 2016;3(2):579-82.
- 3. Datta V, Chaturvedi P. Congenital malformations in rural Maharastra, Indian Paediatrics. 2000;37:998-1001.
- Swain S, Agarwal A, Bhatia BD. Congenital malformations at birth. Indian paediatrics. 1994;31:1187-91.
- 5. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformation at birth in central India, Indian journal of human genetics. 2010;16:159-63.
- Anand JS, Javadekar BB, Belani M. Congenital malformations in 2000 consecutive births. Indian Pediatr. 1988;25:845-51.
- 7. Kalra A, Kalra K, Sharma V. Congenital malformations, Indian Paediatrics. 1984;21:945-9.
- 8. Kumbhar S, Lele PR. The incidence of congenital malformations at birth and mid-trimester abortions and possible etiological factors in Pune camp, Maharashtra, India. International Journal of Research in Medical Sciences. 2016;4(6):1910-4.
- Deshpande N, Borle M, Agarkhedkar S. Congenital Malformations: Patterns and Prevalence at Birth. Webmed Central Paediatrics 2015;6(10):WMC004988.
- Jain SR, Naik JD, Dhakne BR, Prabhu PM, Kamble SV, Mathurkar MP. Pattern of congenital malformations in newborn: a hospital-based study. Int J Res Med Sci. June 22, 2016];4(2):524-528.
- Francine R, Pascale S, Aline1a H. Congenital anomalies: prevalence and risk factors. Universal Journal of Public Health. 2014;2(2):58-63.
- 12. El Koumi MA, Al Banna EA, Lebda I. Pattern of congenital anomalies in newborn: a hospital-based study. Pediatric reports. 2013 Feb 5;5(1):5.
- Jehangir W, Ali F, Jahangir T, Masood MS. Prevalence of gross congenital malformations at birth in the neonates in a tertiary care hospital. In APMC 2009 (Vol. 3, No. 1, pp. 47-50).
- Desai N, Desai A. Congenital anomalies, a prospective study at Bombay hospital, Bombay hospital journal. 2006;48:442-5.
- Saifullah S, Chandra RK, Pathak IC. Congenital malformation in newborn. Indian Paediatrics. 1967;4:251-60.
- Pabbati J, Subramanian P, Sudharshan Raj C, Sadhana N, Rao R. Study on incidence of congenital anomalies in a rural teaching hospital, Telangana, India. International Journal of Contemporary Pediatrics. 2016;3(3):887-90.
- Prashar N, Kachroo N, Gupta S, Bhagat BR, Gupta A, Sharma G, Sharma P. Sex differences in the congenital anomalies: a hospital based cross sectional study. System. 2016 May 12;74(39.57):39-57.
- Singh M. Hospital based data on perinatal and neonatal mortality in India, Indian paediatrics. 1986;23:579-84.
- Ndibazza J, Lule S, Nampijja M, Mpairwe H, Oduru G, Kiggundu M, Akello M, Muhangi L, Elliott AM. A description of congenital anomalies among infants in

International Journal of Medical Pediatrics and Oncology, October-December, 2016:2(4):152-155

Entebbe, Uganda. Birth Defects Research Part A: Clinical and Molecular Teratology. 2011 Sep 1;91(9):857-61.

- Sarkar S, Patra C, Dasgupta MK, Nayek K, Karmakar PR. Prevalence of congenital anomalies in neonates and associated risk factors in a tertiary care hospital in eastern India. Journal of clinical neonatology. 2013 Jul;2(3):131.
- Kumar J, Gordillo R, Kaskel FJ, Druschel CM, Woroniecki RP. Increased prevalence of renal and urinary tract anomalies in children with congenital hypothyroidism. The Journal of pediatrics. 2009 Feb 28;154(2):263-6.
- 22. Materna-Kiryluk A, Wiśniewska K, Badura-Stronka M, Mejnartowicz J, Więckowska B, Balcar-Boroń A, Czerwionka-Szaflarska M, Gajewska E, Godula-Stuglik U, Krawczyński M, Limon J. Parental age as a risk factor for isolated congenital malformations in a Polish population. Paediatric and perinatal epidemiology. 2009 Jan 1;23(1):29-40.
- Gul F, Jabeen M, Khan AS. Frequency of congenital malformations and associated risk factors at liaqat memorial hospital, kohat. Khyber Medical University Journal. 2012 Jul 1;4(3).
- Rittler M, Liascovich R, López-Camelo J, Castilla EE. Parental consanguinity in specific types of congenital anomalies. American journal of medical genetics. 2001 Jul 22;102(1):36-43.
- Bromiker R, Glam-Baruch M, Gofin R, Hammerman C, Amitai Y. Association of parental consanguinity with congenital malformations among Arab newborns in Jerusalem. Clinical genetics. 2004 Jul 1;66(1):63-6.
- Khanna MP, Prasad LS. Congenital malformations in the newborn. Indian journal of paediatrics. 1967;230:63-71.
- 27. Sagunabai NS, Mascarena M, Syamalan K. An etiological study of congenital malformation in the new born, Indian pediatrics.1982;19:1003-7.
- Agarwal SS, Singh US, Singh PS, Singh SS, Das VI, Sharma AN, Mehra PR, Malik GK, Misra PK. Prevalence & spectrum of congenital malformations in a prospective study at a teaching hospital. The Indian journal of medical research. 1991 Dec;94:413-9.