10.5005/jp-journals-10006-1440

ORIGINAL ARTICLE

A Study on Incidence of Various Systemic Congenital Malformations at Birth in a Tertiary Care Hospital of North Western Rajasthan

[1]Suniti V Verma, [2]Ram N Sehra

ABSTRACT

Aims: To study the overall incidence, proportion, and type of congenital malformations in newborns and immediate outcome of anomalous neonates.

Materials and methods: The present study was an observational study. It was carried out between January 2012 to July 2013 among patients admitted to the Department of Gynecology and Obstetrics, PBM Hospital, Bikaner.

Results: We found that the maximum number of mothers was in the age group of 21 to 25 years (44.9%). Central nervous system (CNS) was the most common anomaly (53.3%), followed by gastrointestinal (GIT) anomaly (13.2%). All three anomalies of respiratory system were diaphragmatic hernia. There were totally 35.7% mothers who had consanguinity. Accuracy of ultrasonography was 71.4%. Out of 167 deliveries, 85 babies were live birth, 79 were still birth, and 3 abortions, in that 92 (55.1%) cases were males while 71 (42.5%) babies were females, and 4 (2.4%) ambiguous genitalia. Maximum number of babies (40.1%) had their birth weight between1.5 and 2.50 kg, while 30.6% babies had their birth weight < 1.5 kg, and 29.3% had their birth weight > 2.5 kg.

Conclusion: The overall incidence of congenital malformation in our study was 1.23%. The most common anomaly was of CNS (53.3%), followed by GIT (13.2%); multiple system (10.2%); musculoskeletal (9%); urogenital (4.8%); respiratory (4.8%); and central venous system (1.25%). Consanguinity (35.3%) was a common associated factor in this study.

Clinical significance: These patients need good counseling, balanced diet with folic acid supplementation, treatment of precipitating factors, regular follow-up in subsequent pregnancies, early screening for congenital malformations, and early termination if an anomaly is present.

Keywords: Anencephaly, Congenital malformation, Diaphragmatic hernia, Fetal malformation, Multiple malformations.

How to cite this article: Verma SV, Sehra RN. A Study on Incidence of Various Systemic Congenital Malformations at Birth in a Tertiary Care Hospital of North Western Rajasthan. J South Asian Feder Obst Gynae 2016;8(4):309-313.

[1,2]Professor

[1]Department of Gynecology and Obstetrics, SMS Medical College, Jaipur, Rajasthan, India

[2]Department of Pediatrics, SMS Medical College, Jaipur Rajasthan, India

Corresponding Author: Suniti V Verma, Professor, Department of Gynecology and Obstretics, SMS Medical College, Jaipur Rajasthan, India, Phone: +911412754110, e-mail: rnsehra@gmail.com

Source of support: Nil

Conflict of interest: None

Date of received: 10 July 2016

Date of acceptance: 16 August 2016

Date of publication: October 2016

INTRODUCTION

Congenital malformations are morphological defects that occur in the prenatal period as a result of genetic mutations, chromosomal abnormalities, and adverse intrauterine environment. These are present at birth and clinically manifest at any time in life.[1] It is a stimulating problem for research study because of the high frequency of their occurrence and the devastating effect they may have on the individual and his or her family.

Congenital malformation accounts for 8 to 15% of perinatal deaths and 13 to 16% of neonatal deaths in India. Around 40 to 60% of congenital anomalies are of unknown etiology; 20% are attributed to a combination of hereditary and other factors, 7% due to single gene mutation, 6% caused by chromosomal anomaly, and 5% by maternal illness.

Worldwide incidence of congenital malformation is estimated as 3 to 7% but actual incidence varies widely between countries. This study will generate data of congenital malformations which will help national registry in future. It will also help to classify the etiology of malformations in neonates in order to allow proper genetic counseling, early management, and rehabilitation.

MATERIALS AND METHODS

The present study was an observational study. It was carried out during January 2012 to July 2013 among patients admitted to the Department of Gynecology and Obstetrics, PBM Hospital, Bikaner. All congenital anomalous babies born in the department during the study period either detected before birth by ultrasonography (USG) of mother or detected at birth were included in this study.

To cover all the findings of relevant history and examination, a proforma was designed. All anomalous babies were examined in detail by neonatologist. Immediate outcome of the baby, i.e., whether the baby was alive or dead; whether the baby needs immediate neonatal support or

not was recorded. Baby was followed till the period of mother's hospital stay to know whether the baby needs any immediate corrective surgery or not.

RESULTS

We found that the maximum number of mothers was in the age group of 21 to 25 years (44.9%), and 31.7% cases were registered while 16.2% were booked and 47.9% unbooked. Patients from rural area were 68.3%, and 31.7% were from urban area. There were 43.7% patients who had their gestational age > 36 weeks, 33.5% cases had gestational age 31 to 36 weeks, and 22.8% cases below 30 weeks. In addition, 73.1% patients were either illiterate or had primary education, 12.6% were educated up to middle level, and 9.6% up to secondary level; 39% patients were > 3rd gravida, 27.5% belonged to 2nd gravid, and 33.5% patients were 1st gravida.

The most common anomalies were of CNS (53.3%), followed by GIT (13.2%), multiple system (10.2%), musculoskeletal (9%), miscellaneous (6.6%), urogenital (4.8%), respiratory (4.8%), and central venous system (CVS) (1.25%) (Table 1). Out of 89 CNS cases, 49 had anencephaly, 34 had hydrocephalus, 11 had meningocele, and 1 each had meningomyelocele and corpus callosum agenesis. Out of 22 GIT cases, 16 had cleft lip and cleft palate, 4 had omphalocele, and 2 had gastroschisis. There were 8 urogenital cases consisting of ambiguous genitalia (2), B/L hydronephrosis (1), hypospadias (2), polycystic kidney disease (2), and post-urethral valve (1). Out of total 15 musculoskeletal cases, congenital talipes equinovarus (CTEV) (9), achondroplasia (1), polydactyly (2), sacrococcygeal teratoma (1), skeletal dysplasia (2). All the three anomalies of respiratory system were diaphragmatic hernia (Table 2).

There were totally 35.3% mothers who had consanguinity. While 16.8% women had taken folic acid, 23.4% mothers had family history of malformed babies. Tobacco abuse was present in 64% of mothers. Fever was present in 79.6% of women, while jaundice was present in 1.2%. On USG scan, 62 had polyhydramnios, 3 had oligohydramnios, and 31 had adequate liquor. Out of 133 total USG scanned patients, 95 cases were diagnosed by USG and remaining 38 cases were not diagnosed by USG. Accuracy of USG was 71.4%. There were total 82.6% women who had spontaneous onset of labor, while 17.4% women had induced labor. Out of 167 cases, 141 mothers had spontaneous vaginal delivery, while 23 mothers underwent lower segment cesarean section.

Table 1: Distribution of cases according to system involved

System	*No.*	*%*
Central nervous system	89	53.3
Gastro intestinal tract	22	13.2
Multiple	17	10.2
Musculoskeletal	15	9.0
Miscellaneous	11	6.6
Urogenital	8	4.8
Respiratory	3	1.8
Cardio vascular system	2	1.2
Total	167	100

Table 2: Pattern of distribution of anomalies according to various systems

System	*Anomalies*	*No.*	*%*
Central nervous system	Anencephaly	42	47.2
	Corpus callosum agenesis	1	1.1
	Hydrocephalus	34	38.2
	Meningoencephalocele	1	1.1
	Meningocele	11	12.3
Cardio vascular system	Congenital heart disease	2	100.0
Gastro intestinal tract	Cleft lip and cleft palate	16	72.7
	Gastroschisis	2	9.1
	Omphalocele	4	18.2
Respiratory	Diaphragmatic hernia	3	100
Urogenital	Ambiguous genitalia	2	25.0
	B/L Hydronephrosis	1	12.5
	Hypospadias	2	25.0
	Polycystic kidney disease	2	25.0
	Post urethral valve	1	12.5
Musculoskeletal	Congenital talipus equinovarus	9	60.0
	Achondroplasia	1	6.7
	Polydactyly	2	13.3
	Sacrococcygeal teratoma	1	6.7
	Skeletal dysplasia	2	13.3
Miscellaneous	Down syndrome	2	18.2
	Nonimmune hydrops fetalis	7	63.6
	Marfans syndrome	1	9.1
	Pierre Robbins syndrome	1	9.1

Eighty-five (50.9%) babies were live birth, 79 (47.3%) were still birth, and 3 (1.8%) abortions. Fifteen (17.6%) live born babies died immediately after birth and 70 (82.4%) got admitted in NICU. Thirty-five (50%) babies discharged from hospital; 34.3% babies expired while 6 (8.6%) babies operated and 5 (7.1%) were referred to higher centers. Out of total 167 deliveries, 92 (55.1%) cases were males while 71 (42.5%) babies were females and 4 (2.4%) were ambiguous genitalia. Maximum number of babies, i.e., 67 (40.1%) babies had their birth weight between 1.5 and 2.5 kg, while 51 (30.6%) babies had their birth weight < 1.5 kg and 49 (29.3%) had their birth weight > 2.5 kg.

DISCUSSION

We studied the congenital malformation in 13,614 consecutive births including still birth at PBM hospital, Bikaner. Out of these, 167 babies had one or the other congenital malformations and the overall incidence

of congenital malformation in our study was 1.23% (12.3/1000 live births). Incidence in other study ranged between 1.2 and 9.5%.[2-7] Among 431 still born babies, 79 had congenital malformed babies (18.3%) as compared to live born babies (0.64%). Age-wise distribution in our study shows that most of the women belonged to 21 to 25 years of age group (44.9%). We had 22 cases in age group 35 and above. Another study[8] also reported that 21 to 25 years age group had maximum number of malformed babies. In a study reported by Singh et al, 76.93% cases were within 20 to 30 years age group.

Majority of patients were unbooked (47.9%) while 31.7% cases visited our ANC OPD only once. Only 16.2% of cases were booked or made their visit regularly to our ANC OPD. Majority of patients in our study were from rural areas (68.3%), and only 31.7% patients belonged to urban areas. In developing country like India, there is a lack of knowledge and illiteracy in rural population, due to which they fail to avail good antenatal care that can help early diagnosis and termination of fetuses incompatible with life.

In our study, majority of mothers with congenital anomalous fetuses belong to gestational age > 36 weeks (43.7%). This is because antenatal visit in majority of mothers was irregular. A study done by Fatema et al[9] also revealed that majority of patients belong to gestational age between 34 and 36 weeks. In our study, majority of patients were either illiterate or had primary education (73.1%). Due to illiteracy, women did not take care of themselves during pregnancy and never visited ANC OPD.

Our study shows that majority of mothers belonged to gravida 3 or more (39%), while 27.5% were in 2nd gravid, and 33.5% belonged to 1st gravida. A study[10] also revealed an increase in frequency in primis and in 4th gravida mothers. Another study[4] also reported a higher incidence of malformation in babies born to mothers of gravidity 4 or more.

The most common anomalies were of CNS (53.3%), followed by GIT (13.2%); multiple system (10.2%); musculoskeletal (9%); urogenital (4.8%); respiratory (1.8%); and CVS (1.25%). Out of total 89 CNS cases, 49 had anencephaly, 34 had hydrocephalus, 11 had meningocele, and 1 each had meningomyelocele and corpus callosum agenesis. Another study[11] reported 6.3/1000 incidence of neural tube defects (NTDs) in Manipal. Anencephaly (40%), spina bifida (46%), and encephalocele (6%) are among the common ones. Thus it seems from our study and those of others that CNS anomalies constitute bulk of all the congenital anomalies, with majority of them incompatible with life. Majority of babies with CNS anomalies had either still birth or neonatal death and six babies got operated and discharged. Apart from folic acid supplementation, early diagnosis of NTDs and advising early termination of affected pregnancies with lethal anomalies will help to lower existing occurrence rate of congenital anomalies at birth.[12]

Out of total 22 (13.2%) GIT cases, 16 had cleft lip and cleft palate, 4 had omphalocele, and 2 had gastroschisis. Another two studies[12,13] reported 11.76 and 20.83% of GI tract anomalies respectively. Babies with cleft lip and cleft palate were discharged and referred to pediatric surgeon, while 4 babies with omphalocele were referred to higher centers, and 2 cases with gastroschisis were still born.

In our study, there were 15 cases of musculoskeletal anomalies, which include CTEV (n = 9), achondroplasia (1), polydactyly (2), sacrococcygeal teratoma (1), and skeletal dysplasia (2). But these anomalies were mostly minor and nonfatal and most of them were diagnosed after birth. Most of the babies were referred to orthopedician or pediatric surgeon. Babies with sacrococcygeal teratoma and skeletal dysplasia did not survive. A study[5] reported an incidence of musculoskeletal malformation in 9.69/1000 births as the commonest one in their study. Another study[14] reported 5.9% incidence of skeletal system anomalies, and similar figures were reported by others also.

We had 8 (4.85%) cases of urogenital anomalies consisting of ambiguous genitalia (2), B/L hydronephrosis (1), hypospadias (2), polycystic kidney disease (2), and posterior urethral valve (1). In our study, hydronephrosis was diagnosed before delivery while cases of ambiguous genitalia, hypospadias, polycystic kidney disease, and posterior urethral valve could not be detected by USG and were diagnosed at birth. Both cases of ambiguous genitalia, 1 case of bilateral hydronephrosis, and both cases of polycystic kidney disease did not survive. In Glasgow study[15] over a period of 6 years, 66 cases of obstructive uropathy, 27 cases of unilateral, and 39 cases of bilateral obstruction were detected. There were 4 cases of vesicoureteric and 18 cases of urethral obstruction. Hydronephrosis was a common sonographic diagnostic feature.

In our study, we had 3 cases of diaphragmatic hernia. Only 1 case was diagnosed before birth and rest of all were diagnosed after birth. One case was referred to higher center; rest 2 cases did not survive. In our study, we had 17 cases (10.2%) of multiple system malformation. Most of the major anomalies were associated with minor anomalies of another system. In a study[2], the authors found multiple anomalies in 37.6% of anomalies, whereas another study[4] reported multiple anomalies in 18.8% babies.

In our study, we analyzed 167 cases, some were prospective and some retrospective to find out the cause or predisposing factor in those cases. Consanguinity was present in 59 cases (35.3%) and was associated with several anomalies like hydrocephalus, spina bifida,

omphalocele, cleft lip and palate. Various studies[10,16] also had consanguinity in their study as one of the associated factors of congenital malformation. In our study, we had 133 cases (79.6%) who had history of febrile illness in their 1st trimester and 2 had history of jaundice. Various studies[2,3,10] also reported an increased incidence of congenital malformation associated maternal febrile illness in 1st trimester in their studies. Women who have delivered a malformed baby are more prone to deliver an abnormal child again. In our study, we had 20 such cases (12.6%) who had one or more malformed babies in the past and again delivered babies with congenital malformations. It is accepted that the risk factor for the 2nd child if the 1st is anomalous is 1 in 20; after the birth of 2 malformed children, the risk of the 3rd child to be malformed is 1 in 10.

On USG scan 62 (37.1%) had polyhydramnios, 3 (1.8%) had oligohydramnios, and 31 had adequate liquor. In our study, 62 cases who presented as polyhydramnios had anencephaly, hydrocephalus, meningocele, while 3 cases of oligohydramnios had polycystic kidney disease, bilateral hydronephrosis. Out of 133 total USG scanned patients, 95 cases were diagnosed by USG and remaining 38 cases were not diagnosed by USG. Accuracy of USG was 71.4%. In a large study, on ultrasound diagnosis for congenital malformation which screened 1,737 women in 1st and 2nd trimester, 27 fetal anomalies were diagnosed (1.55%). Authors advocate routine sonography preferably around 20 weeks of gestation to detect congenital malformations.

In our study, 85 babies were live birth, 79 were still birth, and 3 abortions. Fifteen (17.6%) live born babies died immediately after birth and 70 (82.4%) got admitted in NICU. Thirty-five (50%) babies discharged from hospital; 34.3% babies expired while 6 (8.6%) babies operated, and 5 (7.1%) were referred to higher center. These babies who went home had minor anomalies. Another study[11] had 11 abortions, 24 intrauterine death, 8 neonatal death, and 7 survivals. Out of total 167 deliveries, 92 (55.1%) cases were males, while 71 (42.5%) babies were female and 4 (2.4%) ambiguous genitalia. Various studies have reported higher incidence of malformations in males as compared to females. Maximum babies (40.1%) had their birth weight between 1.5 and 2.5 kg, while 30.6% babies had their birth weight <1.5 kg and 29.3% had their birth weight >2.5 kg. Hence, most congenitally anomalous babies belonged to the low birth weight group.

CONCLUSION

The overall incidence of congenital malformation in our study was 1.23%. Incidence of congenital malformations was much higher in still born babies (18.3%) as compared to live born babies (0.6%). Commonest anomaly were of CNS (53.3%), followed by GIT (13.2%); multiple system (10.2%); musculoskeletal (9%); urogenital (4.8%); respiratory (4.8%); and CVS (1.25%). Consanguinity (35.3%) was a common associated factor in this study. Congenital malformations were mostly found in mothers who had not taken antenatal care, folic acid in their pregnancy. Common age group was 21 to 25 years and gravida 3 or more were more prone to have malformed babies. Diagnostic accuracy of USG scan was 71.4%. Eighty-five babies were live birth, 79 were still birth, and 3 abortions. Fifteen (17.6%) live born babies died immediately after birth and 70 (82.4%) got admitted in NICU. Thirty-five (50%) babies discharged from hospital; 34.3% babies expired while 6 (8.6%) babies operated and 5 (7.1%) were referred to higher centers. Among them, 92 (55.1%) cases were males, 71 (42.5%) babies were females, and 4 (2.4%) were ambiguous genitalia. Maximum babies (40.1%) had their birth weight between 1.5 and 2.5 kg, while 30.6% babies had their birth weight <1.5 kg, and 29.3% had their birth weight >2.5 kg. Hence, most congenitally anomalous babies belonged to the low birth weight group.

CLINICAL SIGNIFICANCE

These patients need good counseling, balanced diet with folic acid supplementation, treatment of precipitating factors, regular follow-up in subsequent pregnancies, early screening for congenital malformations, and early termination if an anomaly is present. This study was an effort to find out an actual picture of congenital anomalous babies in this tertiary care center. However, this study should only be regarded as preliminary. From this study, some clue may be derived regarding frequency and distribution pattern of congenital malformations in North Western Rajasthan.

REFERENCES

1. Sadler TW. Congenital malformations. In: Langmans' Medical embryology. 8th ed. Philadelphia (PA): Lippincott Williams & Wilkins; 2000. p. 95.
2. Mishra PC, Baveja R. Congenital malformations in the newborn – a prospective study. Indian Pediatr 1989;26(1): 32-35.
3. Verma M, Chhatwal J, Singh D. Congenital malformations – a retrospective study of 10,000 cases. Indian J Pediatr 1991 Mar-Apr;58(2):245-252.
4. Swain S, Agarwal A, Bhatia BD. Congenital malformations at birth. Indian Pediatr 1994 Oct;31(10):1187-1191.
5. Bhat BV, Babu L. Congenital malformations at birth – a prospective study from South India. Indian J Pediatr 1998 Nov-Dec;65(6):873-881.
6. Grover N. Congenital malformations in Shimla. Indian J Pediatr 2000 Apr;67(4):249-251.

7. Patel ZM, Adhia RA. Birth defects surveillance study. Indian J Pediatr 2005 Jun;72(6):489-491.
8. Choudhary AR, Mukherjee M, Sharma A, Talukdar G, Ghos PK. Study of 1,26,266 consecutive births for major congenital defects. Indian J Pediatr 1989 Jul-Aug;56(4):493-499.
9. Fatema K, Begum F, Akter N, Zaman SMM. Major congenital malformations among the newborns in BSMMU hospital. Bangladesh Med J 2011;40(1):7-12.
10. Chatuvedi P, Banerjee KS. An epidemiological study of congenital malformations in newborn. Indian J Pediatr 1993 Sep-Oct;60(5):645-653.
11. Lavanya R, Pratapkumar M. Prevalence and outcome of NTDs in a South Indian Hospital. J Obstet Gynaecol India 1996 Dec;46(6):797-802.
12. Sarkar B, Bhaduria S, Gupta, Chandra M. Ultrasound evaluation of pregnancies at risk for congenital anomalies. J Obstet Gynecol India 1991;40(1):40.
13. Rajan R. Ultrasound diagnosis of fetal anomalies. J Obstet Gynecol India 1989;39(4):461-466.
14. Lucas M, Leven KJ, Williams ML, Whalley R. Early pregnancy glycosylated hemoglobin, severity of diabetes and fetal malformations. Am J Obstet Gynecol 1989 Aug;161(2):426-431.
15. Rani R, Cameron A, Munro F. Fetal obstructive uropathies and prenatal diagnostic evaluation and outcome. J Obstet Gynecol India 1996 Jun;46(3):318-323.
16. Kulkarni ML, Kurian M. Consanguinity and its effect on fetal growth and development: A South Indian study. J Med Genet 1990 Jun;27(6):348-352.