ONE YEAR PROSPECTIVE STUDY OF CASES OF PREGNANCY WITH CONGENITAL MALFORMATIONS OF FETUS

Jyotsna HarlalkaKamra¹, Mandeep Kaur², Preeti Jain³, Suman Rana⁴.

1. Assistant Professor, Department of Obstetrics and Gynaecology , Maharaja Agresen Medical College, Agroha, Hisar.
2. Medical officer, Department of Obstetrics and Gynaecology, PCMS , service number 7028.
3. Senior Resident, Department of Obstetrics and Gynaecology , Maharaja Agresen Medical College, Agroha, Hisar.
4. Assistant Professor, Department of Paediatrics , Maharaja Agresen Medical College, Agroha, Hisar.

CORRESPONDING AUTHOR

Dr. JyotsnaHarlalkaKamra,
H.No. 2420, sector 14,
Behind Sreeram Public School,Hisar.
Email- jyotsnakamra@gmail.com

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ABSTRACT: OBJECTIVE: To study the incidence of congenital malformation, to diagnose congenital malformations by USG in antenatal period as early as possible and to find out the cause and associated factors affecting congenital malformation.**MATERIALS AND METHODS:** The present study was conducted for one year in the department of obstetrics and Gynaecology, Rajindra Hospital Patiala. All cases whether detected antenatally or at the time of birth were taken. The diagnosis was made by USG in antenatal cases and at the time of delivery in those cases who have no previous USG reports or antenatal checkups. **RESULTS:** There were total 900 deliveries, out of which 831 were live born and 69 were still born. Congenital malformations were present in 62 cases out of which 23 were live born, 29 were still births and there were 10 cases of abortion. The overall incidence of congenital malformations in the present study was 68.89/1000 births (6.889%). Out of total 62 cases of congenital malformations, CNS malformations were found to be the most common (66.13%), followed by musculoskeletal system (24.19%). In the present study USG was found to be a good method of screening of congenital malformations with high specificity (100%) and sensitivity (73.21%). The PPV was 100% and NPV was 98.01%. The percentage of false positive results was 0% but that of false negative was 26.78%. **CONCLUSION:** Ignorance, illiteracy and unawareness about the availability of USG, lack of adequate antenatal checkups is the main cause of late diagnosis of congenital malformations. Best effort should be put in to make the diagnosis of congenital malformations early so that referral can be made at the earliest for early intervention and planned delivery at an appropriate center.

KEYWORDS: congenital malformations, USG, antenatal, central nervous system

INTRODUCTION: One of the most traumatic experiences not only for the pregnant females, but also for the obstetrician is to deliver a congenitally malformed fetus. It is estimated that each year, more than 4 million children are born with serious birth defects that cause death or lifelong disability for

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the fetus or baby and hardships for the entire family. According to WHO, 1992 incidence of congenital malformations is 17-30/1000 live births and the congenital heart defects are the most common congenital malformations

Currently, the most widely used technique for fetal evaluation is 2 dimensional real-time ultrasound. More recent advances involve the use of 3 dimensional ultrasound. Detection of birth defects is a major reason for which screening antenatal ultrasound is advocated (Carvalho MH et al, 2002). Ultrasound scan is currently considered to be a safe, non-invasive, accurate and cost-effective investigation of the fetus.

MATERIAL AND METHODS: The present study was conducted in the OPD and labor room of the department of obstetrics and Gynaecology, Rajindra Hospital Patiala. This was one year study (September 2006 to august 2007) and all cases whether detected antenatally or at the time of birth were taken. A detailed history was taken. Relevant information regarding maternal age, gestational age, sex, community, birth weight, birth order and consanguinity was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. The diagnosis was made by USG in antenatal cases and at the time of delivery in those cases who have no previous USG reports or antenatal checkups. Various parameters like age, parity, booked/unbooked, rural/urban, socioeconomic status, sex of baby, type of congenital malformations, USG done/not done were taken. Following observations were made from the study:

OBSERVATIONS: As evident from table 1, there were total of 900 deliveries out of which there were 831 live births and 69 were stillbirths. Total number of babies with congenital malformations were 62 out of which there were 23 live born babies, 29 still born babies and 10 were aborted (<20wks). Incidence of congenital malformations in the present study is 68.89/1000 births (6.889%)

TABLE 1: GENERAL SURVEY OF CONGENITAL MALFORMATIONS

Groups	No. of Cases
Total deliveries	900
Total number of babies	900
Total live born babies	831
Live born babies with cong. Malf.	23
Total still births	69
Still births with cong. Malf.	29
No. of abortions of cong. Mal. Fetuses <20wk	10
Total no. of male babies	526
Male babies with cong. Malf.	30
Total female babies	372

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Female babies with cong. Malf.	30
Fetuses with concealed sex	2
Total fetuses with cong. Malf.	62
No. of booked cases	404
No. of unbooked cases	496
Incidence of babies with cong. Malf.	68.89/1000 (6.889%)

Table 2: COMPARISON OF CONGENITAL MALFORMATIONS IN LIVE/STILL BIRTHS

Birth status	Malformed fetuses	Normal babies	Total Babies	Incidence of Malf./ 1000
Live birth	23	808	831	27.06 (2.76%)
Still birth	29	40	69	420.2 (42.02%)
Total	52	848	900	

TABLE 2 clearly indicates that the incidence of congenital malformations is more in still born babies as compared to live born babies and the difference is statistically significant ($p < 0.05$).

TABLE 3: DISTRIBUTION OF MALFORMATIONS IN DIFFERENT SYSTEMS

System involved	No. of cases having cong. Malf.	%age	Incidence per 1000 births
CNS	41	66.13	45.55
Musculoskeletal group	15	24.19	16.66
Orofacial group	7	11.29	7.77
CVS	6	09.67	6.66
Gastrointestinal	5	08.06	5.55
Genitourinary	4	06.45	4.44
Miscellaneous	9	14.51	10

Out of total 62 cases of congenital malformations, CNS malformations were found to be the most common (45.55/1000), followed by musculoskeletal system (16.66/1000), miscellaneous (10/1000), orofacial group (7.77/1000), CVS (6.66/1000), GIT system (5.55/1000) and genitourinary system (4.44/1000).

In the present study, incidence of congenital malformations was found to be more in unbooked cases than in those with booked cases (7.66% vs 5.94%) and was more in rural population (7.15%) as compared to urban population (6.50%) the difference is statistically significant ($p < 0.05$).

The highest proportion of malformed babies, 41.94% were born to parents of lower middle class. 58.06% malformed babies were belonged to combined poor & lower middle class. This indicates that congenital malformations are more in lower social class groups and rural population and it is suggested that malnutrition of pregnant mother, poor hygiene and living status could have contributed to a higher incidence in these classes.

The incidence of congenital malformations was more in female babies as compared to male babies with a difference (8.77% vs 5.70%) which is very much significant ($p < 0.05$) statistically.

The rate of fetal malformations was found to be highest in 25-29 year age group (13.04%) followed by age group >35 years (6.12%). The rate of congenital malformation was observed to be lowest in age group <19 year. It was observed that the incidence of congenital malformations was more in multigravida as compared to primigravida and maximum in second gravida (10%) followed by gravida four (8.79%). The incidence was lowest in primigravida.

TABLE 4: HIGH RISK CASES OF PREGNANCY AND CONGENITAL MALFORMATIONS

High risk factor	No. of cases	
	No.	%age
Oligohydramnios	9	14.52
Polyhydramnios	8	12.90
Age (>35 years)	6	9.68
Prev. history of cong. Malformation	5	8.06
Prev. H/o 2 or more abortions	2	3.22%
H/o epilepsy	1	1.61%
Jaundice	1	1.61%
Diabetes Mellitus	1	1.61%

The table no 5 reveals an association between various obstetrical conditions and the occurrence of congenital malformations.

Table 5: CONGENITAL MALFORMATIONS AND ULTRASONOGRAPHY

USG	Control Group	Study group	Total
Done	738	56	794
Non done	100	6	106
Total	838	62	900

In the present study, USG was done in 794 cases, out of which 738 cases were without any congenital malformations and 56 were with malformations. USG was not done in 106 cases out of which 100 were without congenital malformations and 6 cases were with congenital malformations.

TABLE 6: USG IN DIAGNOSIS OF CONGENITAL MALFORMATIONS

USG Result	Cong. Anomaly	No cong. Anomaly	Total
Cong. Anomaly	41 (True positive)	0 (false positive)	41
No cong. Anomaly	15 (False negative)	738 (True negative)	753
Total	56	738	794

The overall sensitivity of USG in diagnosing congenital malformations was 73.21% and specificity was 100%. Positive predictive value of USG was 100% and negative predictive value was 98.01% whereas percentage of false negative was 26.78% and that of false positive was 0%.

TABLE 7: OVERALL SENSITIVITY AND SPECIFICITY OF USG IN DIAGNOSING CONGENITAL MALFORMATIONS IN UTERO

System	Sensitivity	Specificity	PPV	NPV	%of false -ve	%of false +ve
CNS	97.22	100	100	99.86	2.78	0
Musculoskeletal	46.15	100	100	99.11	53.85	0
Orofacial	33.33	100	100	99.49	66.66	0
GIT	100	100	100	100	0	0
GUT	100	100	100	100	0	0
CVS	40	100	100	99.62	60	0

In the present study the sensitivity of USG in diagnosing congenital malformations was found to be highest for GIT (100%) and GUT (100%) system followed by CNS (97.22%) system, the sensitivity was lowest for orofacial system (33.33%). The specificity was observed to be 100% for all systems. The PPV of USG in diagnosis of congenital malformations was again found to be 100% for all systems. The NPV was appreciably high for all systems (99-100%). The percentage of false negative cases was observed highest in orofacial group followed by CVS group and musculoskeletal system. It was lowest in GIT and GUT (0%) system. The percentage of false positive cases was 0 in all group.

DISCUSSION: As shown in table 8, the incidence of congenital malformations in various studies is low (1.6 to 3.7%) as compared to the present study which is comparatively high (6.888%). Incidence is also high in still births in present study in comparison to other studies.

TABLE 8: INCIDENCE OF CONGENITAL MALFORMATIONS

Study	Incidence (%)	Incidence in stillbirths	Incidence in livebirths
Aggarwal SS et al, 1991	1.6	16.4%	1.6%
Bhat BV et al, 1998	3.7	15.7%	3.2%
Patel ZM et al, 2005	1.6	15.8%	
Present study	6.889	42.02%	2.76%

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This wide variation in the incidence of malformations between different studies is due to various factors such as population sampled, geographical and racial difference, selection of cases-liveborn, stillbirth or both and age of the baby when diagnosis is made. (Some authors did follow up of the cases upto various time period postnatally). It may also be due to the fact that this is a referral institute and more cases with congenital malformations were referred from periphery.

Our study is comparable to study done by Jain VK et al (1993) who observed significant difference between rural and urban population.

According to report of WHO, the Expert Committee 1970, the incidence of congenital malformations were more in lower socioeconomic class. Similar results were obtained by various studies as well as in the present study. This could be because of poor nutrition, poor hygiene, illiteracy, poor living status, no folic acid intake during 1st trimester pregnancy and no antenatal checkups including timely USG during pregnancy.

In the present study, the total incidence of congenital malformations was more in females (8.77%) than males (5.70%). Grover N et al (2000) and Chaturvedi P et al (1993), reported no sex predilection whereas Bhat BV et al (1998), reported an increased incidence in males. Our study differs from these studies because maximum number of cases was of CNS malformations in which females outnumbered males.

In the present study, maximum incidence of congenital malformations was observed in age group 25 – 29 yr. This is similar to observations made by Kulshrestha et al (1983). However Grover et al (2000), and Khanum S et al (2004) found higher incidence in age group >35 yr which is 2nd most common group in this study.

In the present study, incidence is maximum in multipara. This is similar to studies done by Grover N et al (2000), Khanum S et al (2004), all of them found higher incidence in multipara

Overall recurrence rate of CNS malformations in present pregnancy is 8.06% and recurrence of NTD's is 4.83%. This is comparable to studies done by Verma et al (1976), Hansmann M et al (1982), who found recurrence of NTD's between 3to 5%.

Table 9: SPECIFICITY AND SENSITIVITY OF USG IN DIAGNOSING CONGENITAL MALFORMATIONS

Study	Specificity	Sensitivity
Lys et al, 1989	>99%	18%
Rosendhal et al, 1989	>99%	63%
Todros T et al, 2001	99%	56%
Balakumar K, 2004	98.6%	72.2%
Present study	100%	73.21%

The specificity of USG in diagnosing congenital malformations was high in all studies as shown in above table. Similarly the same result was obtained in our study. In the present study, overall sensitivity of USG in diagnosis of congenital malformations was 73.21 % which is comparable to Balakumar K (2004) who found 72.2% sensitivity. Rosendhal et al (1989), Todros T et al (2001) also have comparable results whereas lower rate was observed by Lys et al (1989). The variation in results in various studies is due to that, identification of birth defects is related to the training of the physician performing or interpreting the examination and directly related to the experience of the individual performing the evaluation.

Table10: SENSITIVITY OF USG IN DIAGNOSING VARIOUS MALFORMATIONS

Study	CNS	Musculo-skeletal	CVS	Git	Gut
Vial et al, 2001	82%	42%	23%	56%	71%
TodrosT et al, 2001	88%	37%	28%	54%	88%
Balakumar K, 2004	39.21%	11.79%			18.09%
Present study	97.22%	46.15%	40%	100%	100%

Above table shows various studies and there comparison of USG sensitivity in diagnosing malformations of various system

CONCLUSION: Congenital malformations pose a threat for survival of a newborn in the early neonatal period. Prevention of congenital malformation can be achieved by,

1. Proper marriage counseling for avoiding early and consanguineous marriages.
2. Periconceptional folate consumption should be advised during the marriage counseling itself so that parents understand the need for the same when they are planning for an issue.
3. During pregnancy especially during first trimester mother should be advised to avoid drugs, irradiation, chemicals and infections which act as teratogenic insult.
4. Emphasis should be laid on for a regular antenatal checkup and need for atleast one antenatal ultrasound scan at around 18-20 weeks to rule out gross congenital malformations.
5. An attempt should be made to screen for high risk mothers first and then subject them to undergo investigations like high resolution ultrasound, amniocentesis and chorionic villus sampling wherever required.
6. In view of continued high incidence of congenital heart disease, fetal echocardiography should be done at around 16-20 weeks in high risk cases.
7. Best effort should be put in to make the diagnosis of congenital malformations early so that referral can be made at the earliest for early intervention and planned delivery at an appropriate center.
8. Whenever genetic etiology is suspected, the parents should be referred to genetic counselling after confirming the diagnosis by appropriate methods like karyotyping.

A rational and multidisciplinary approach with good communication between various specialities (obstetrician, ultasonographer, Paediatrician, Genetist and pathologist) and parents is essential.

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Fig. 1 Anencephaly

Fig. 2 Bilateral CTEV