

Detection of Congenital Structural Anomalies by Mid Trimester Ultrasonography

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ABSTRACT

Introduction: Every pregnant female desires to have a healthy child. The detection and prevention of congenital anomalies are main targets of proper antenatal care. Ultrasonography is the best modality for screening and diagnosis of congenital anomalies prenatally as it is non-invasive, quick and safest technique. It is of paramount importance to further decide the management which ranges from termination of pregnancy to elective delivery. The most important period for detection of fetal anomalies by ultrasonography is in midtrimester. When major anomalies are detected, timely termination of pregnancy have significantly decreased the morbidity to the child and the family.

Materials and Methods: A prospective observational study was conducted in which 4130 patients were screened for congenital structural abnormalities by ultrasonography in the Department of radiodiagnosis, Gajra Raja Medical College and JAH group, over a period of 1.5 years. Proper documented informed consent, referral paper, identity card and proper detailed history with risk factors were taken. Patients were evaluated by Mindray DC-30 and Esaote Mylab 7 using 3.5 -5 MHz curve probe.

Results: A total 4130 women were screened out of which 69 positive cases were found. These positive cases were diagnosed with 81 anomalies with prevalence found to be 1.6%. Screening by ultrasonography is efficacious as on follow up, 55% of cases underwent termination with USG sensitivity and specificity being 95.7% and 99.95% respectively.

Conclusion: Mid trimester ultrasonography gives advantage of relatively higher detection rate of anomalies and a useful tool for decision making in further management of the patient.

Key words: Ultrasonography, Mid Trimester, Congenital structural abnormalities.

1. INTRODUCTION

Congenital anomalies can be defined as structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth or later in life.^[1] There are various screening techniques, noninvasive technique and invasive technique for prenatal diagnosis of congenital anomalies.^[2] Detection of structural anomalies by the obstetric ultrasound is the best modality for diagnosis of congenital anomalies prenatally as it is non-invasive, quick, cost effective and safest technique. The

most important period for detection of fetal anomalies by ultrasonography is in mid trimester as most of the anomalies can be detected in this period. Technical advances in ultrasound have helped in easy detection of structural anomalies since few decades.^[3] When major anomalies are detected, timely termination of pregnancy have saved the cost and tragedy of losing viable fetus.^[2]

Antenatal diagnosis of significant fetal anomalies offers a variety of options for the pregnant women ranging from termination of pregnancy to elective delivery at a center, equipped to perform highly specialized neonatal surgical procedure.^[4]

2. MATERIAL AND METHODS

A prospective observational and non interventional study was performed. All the antenatal patients in mid trimester who were referred to the Department of Radiodiagnosis, G.R.M.C. Gwalior from Department of Obstetrics and Gynaecology were screened for structural congenital abnormalities on ultrasonography over a period of one and half years, between November 2019 and June 2021.

After taking proper documented informed consent, referral paper, identity card and risk factors, ultrasonography scan was performed. Collected information and data was evaluated, then outcome of pregnancy and necessary statistical analysis was performed. All antenatal patients in mid trimester referred by department of obstetrics and gynaecology to department of radiology in Chambal region were included while congenital heart diseases anomalies were excluded in this study.

Antenatal ultrasonography examination of 4130 pregnant women was done using with Mindray DC-30 and E-SaoteMylab 7 using 3.5 -5 MHz curve probe used with ultrasonology gel application.

3. RESULTS

Table 1: Summary of the gestational age –wise distribution of anomalies

S. No.	Spectrum of Anomalies	No. of Cases	<20 Weeks	>20 Weeks
1	CNS	42	39	03
2	Urogenital System	15	12	03
3	Musculo-Skeletal System	04	03	01
4	Anterior Abdominal Wall Defect	06	05	01
5	Thoracic	05	05	---
6	Facial & Neck	04	03	01
7	Hydrops Foetalis	01	01	---
8	GI System	04	04	---
	Total	81	72	09

Table 2: Central nervous system anomalies incidence

Group	S. No.	CNS Anomalies	No. of Cases	≤ 20 Weeks	>20 Weeks	%
1) NTD	1	Acrania/Anencephaly	15	15	----	35.71%
	2	Spina Bifida	06	6	----	14.28 %
	3	Encephalocele	05	5	---	11.90 %
2) Ventriculo-megaly	1	Ventriculomegaly Isolated or With Associated Malformation	11	9	2	26.19 %

3) Midline Abnormality	1	Dandy-Walker Malformation	02	1	1	4.76%
	2	Holoprosencephaly	03	3	-----	7.14%
Total Cases			42	39	3	100 %

Table 3: Urological System Anomalies Incidence

S. No	Urological Anomalies	No. of Cases	≤20 Weeks	>20 Weeks	%
1	Urinary Tract Dilation (Hydronephrosis)	8	6	02	53.3 %
2	Renal Cystic Disease	5	4	01	33.3 %
3	Renal Agenesis	1	1	---	6.6 %
4	Bladder Outlet Obstruction (BOO)	1	1	---	6.6 %
	Total Cases	15	12	3	100 %

Table 4: System-wise case pattern of pregnancy outcome

S. No.	System Involvement	No. of Cases with Positive Findings	Termination	Still Births	Live Births
1	CNS	34	30	2	2
2	Urogenital System	13	1	2	10
3	Musculo-Skeletal System	03	0	0	3
4	Anterior Abdominal Wall Defect	06	3	0	3
5	Thoracic	05	0	1	4
6	Facial & Neck	04	3	0	1
7	Hydrops Foetalis	01	1	0	0
8	GI System	03	0	0	3
	Total Cases	69	38	5	26

Table 5: Sensitivity and specificity calculation of ultrasound scanning

Ultrasonography Report	Anomaly present	Anomaly absent
Positive	67 True Positive (TP)	02 False Positive (FP)
Negative	03 False Negative (FN)	4058 True Negative (TN)

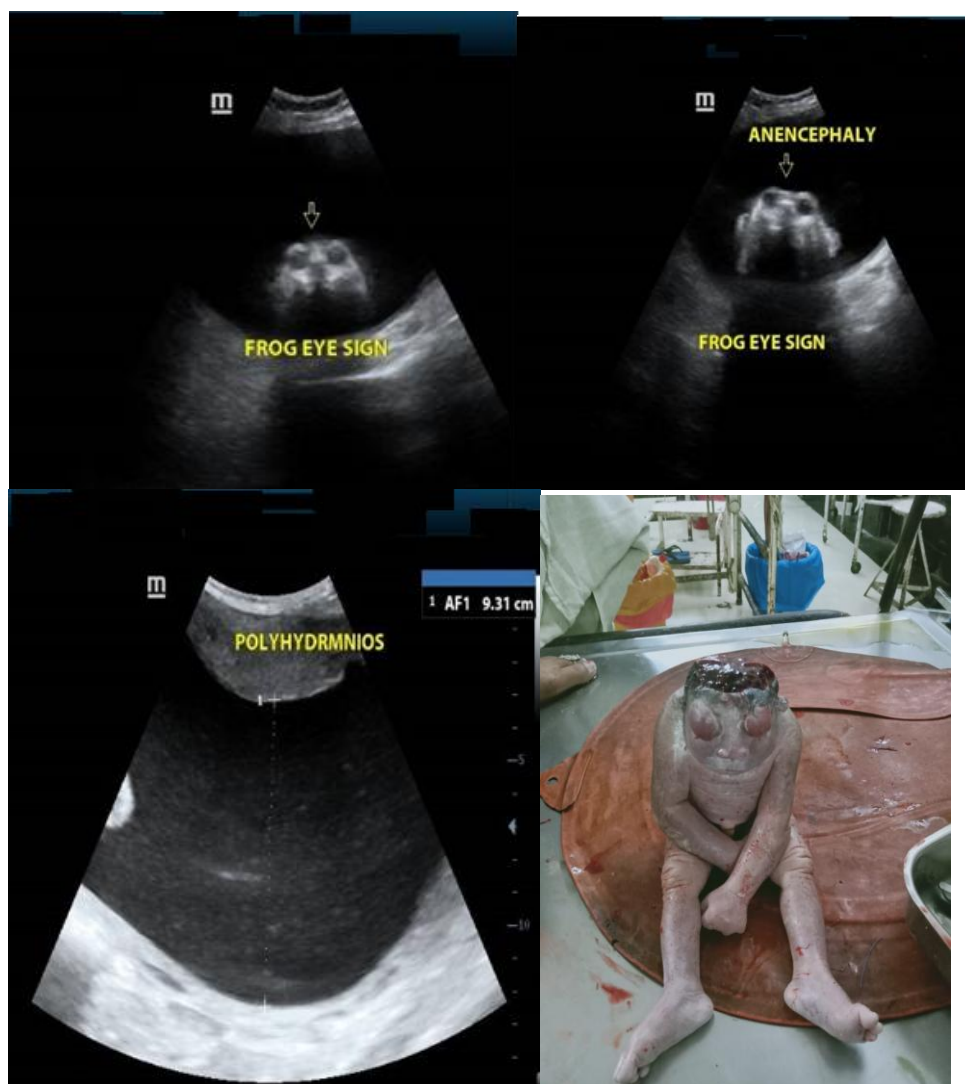


Figure 1: Anencephaly associated with polyhydramnios

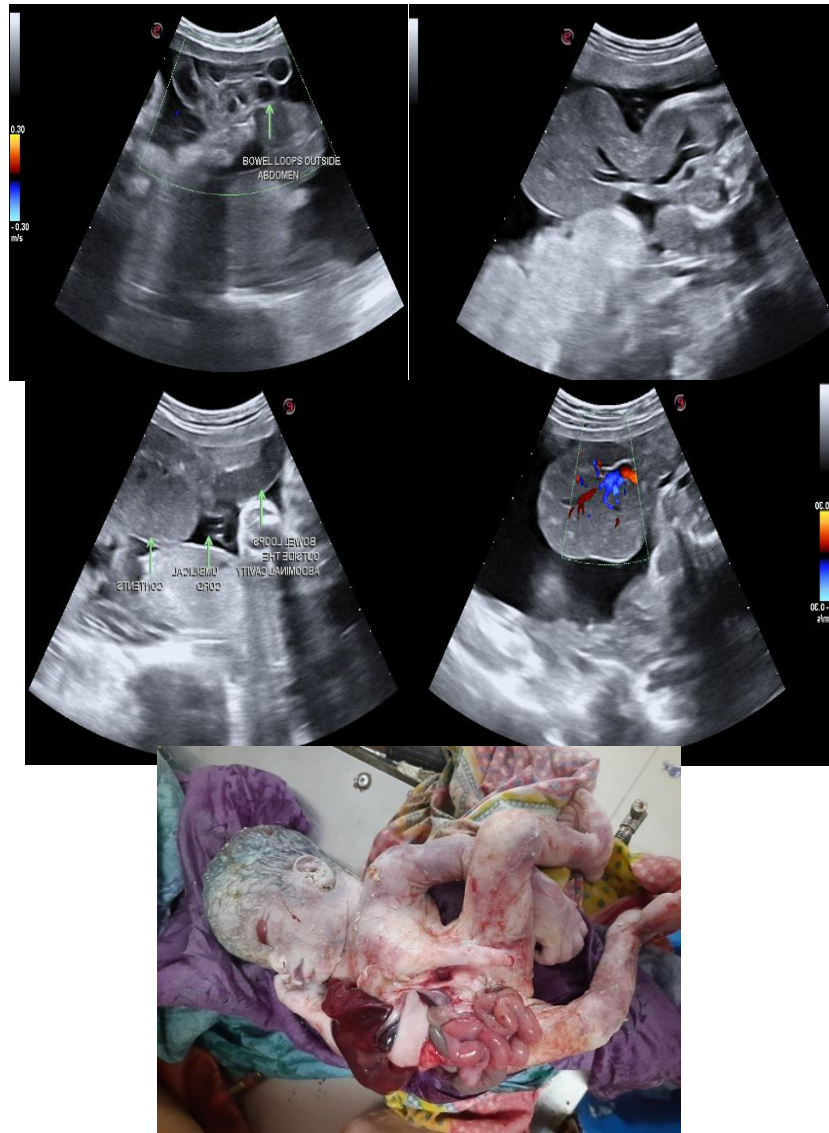


Figure 2: Gastroschisis

A total of 4130 pregnant females were screened by ultrasonography out of which 69 positive cases were found which were diagnosed with 81 anomalies, so this concluded that one fetus can have multiple anomalies. The incidence of congenital anomalies in Chambal region was found to be 1.6%. Maximum number of anomalies were found in the age group of 21-25 years and women above 30 years of age had relatively higher incidence which concludes that the maternal age is an important parameter in prevalence of congenital anomalies. It was observed that primigravida women had more number of anomalies than multigravida women. Consanguineous marriage played a significant risk factor contributing to anomaly incidence. Amongst the risk factors, absent folic acid prophylaxis, elderly pregnant women and women with history of previous child with abnormality played significant risk factor in development of congenital anomalies, out of which absent folic acid prophylaxis was the most common which is preventable. Our results showed that detection rate of anomalies were more (88.89%) prior to 20 weeks of gestation than that after 20 weeks of gestation (11.11%). [Table 1]

Most common system affected by anomaly was central nervous system out of which neural tube defects were the most common group and anencephaly [Figure 1] followed by ventriculomegaly were the most common ones amongst these [Table 2]. There was an

association of CNS anomalies, anterior abdominal wall defect and duodenal atresia with polyhydramnios and renal anomalies with oligohydramnios.

Urological system anomalies[Table 3] accounted for second most frequent group of anomalies out of which, urinary tract dilatation anomalies were found to be more frequent (53.3%) followed by renal cystic disease(33.3%).

Congenital talipes equinovarus(CTEV)had more prevalence than micromelia and rhizomelia amongst musculoskeletal system anomalies.

It showed prevalence of Gastroschisis[Figure 2] more than the omphalocele in anterior abdominal wall defects detected.

Congenital Diaphragmatic Hernia (CDH)and Congenital Pulmonary Airway Malformation (CPAM) were most frequently diagnosed thorax anomalies.

Duodenal Atresia was frequent diagnosed gastro-Intestinal Tract Anomaly.

Screening by ultrasonography was beneficial in this study as on follow up, 55% of cases underwent termination due to early detection[Table 4]. Sensitivity and specificity calculation of ultrasound scanning was found to be 95.7% and 99.95% respectively[Table 5].

4. DISCUSSION

Prevalence of congenital anomalies in Chambal region was determined. USG was used as a screening tool for anomalies as it is quick, non-invasive and safest technique. The detection rate for the anomalies is higher in mid trimester as compared to first trimester so, mid trimester screening was performed in this study. Weiszl B et al. found that the detection rate of fetal anomalies at 11- 14 weeks is 44% in comparison to 74% by the midpregnancy scan.^[5] Special benefits of routine ultrasound examination at mid-trimester has been the detection of congenital anomalies which was confirmed by the Helsinki trial.^[6]

The incidence of congenital anomalies in Chambal region was found to be 1.6% which closely correlates with a study done by Van Dorsten JP et al.20 in 2031 pregnant ladies where the overall prevalence of congenital anomaly in screening group was 1.3% (n =21) in screening group.^[7]

Central nervous system anomalies(51.85%) contributed to maximum number of anomalies amongst which neural tube defects(61.9%) were the most common group and anencephaly followed by ventriculomegaly were the most common ones in this group. Similar results were found in the study conducted by babu and pasula.^[8]

Antenatal diagnosis of significant fetal anomalies is beneficial as it decreases the morbidity of the child and family and also decreases the burden on the society. Eik-Nes et al. study concluded that screening for congenital anomalies decreased unnecessary inductions and reduced perinatal morbidity and mortality.^[9]

Once anomalies are detected, various options are available for pregnant mother ranging from termination of pregnancy to elective delivery.

The most common risk factor for development of NTD was found to be absence of folic acid prophylaxis (21.7 %) during antenatal period. So, NTD is one the preventable cause which can be prevented by folic acid prophylaxis. The utility of periconceptional folic acid in prevention of NTDs has been documented long ago.^[10,11] Implementation of food fortification with folic acid has shown a reduction in the incidence of NTDs.^[12,13]

Prenatal diagnosis provides various management options ranging from pregnancy termination or elective delivery.

69 positive cases were diagnosed with 81 anomalies which indicates that one fetus can have multiple anomalies which correlated with study done by **injal G. Patel, Chintu C et al.** that showed multiple malformations can be found in a single fetus.^[14]

5. CONCLUSION

A total of 4130 pregnant females were screened by ultrasonography of which 69 positive cases were found with 81 anomalies. The incidence of congenital anomalies in Chambal region was found to be 1.6%. The most common risk factor was absent folic acid prophylaxis which is preventable. Most common system affected was central nervous system out of which neural tube defects were the most common group and anencephaly was the most common ones amongst these.

On follow up, 55% of cases underwent termination, therefore, we conclude by our study that screening by mid trimester ultrasound, most of the anomalies can be detected prenatally which is a useful tool in decision making in further management for the patient and decreasing the burden on the society.

6. REFERENCES

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