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Original Research Article

## Congenital anomalies in fetus during pregnancy-predisposing factor and management in rural set up

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### ABSTRACT

**Background:** Congenital anomalies are the vital causes for perinatal mortality and morbidity. Therefore, an antenatal diagnosis and foetal therapy have attained importance in the field of human embryology.

**Methods:** This was an observational study conducted in antenatal, labour and postnatal ward at Rural medical college and hospital for 2 years. The aim of study is to review the overall frequency of congenital malformation and to increase public awareness in saving newborns from birth defects.

**Results:** Cause of congenital anomalies was unknown in 82.1% of the cases in our study. Out of 162 congenital anomalies 60.49% were antenatally registered whereas 39.51% were unregistered. Most common system involved in congenital anomaly was Central nervous system (CNS) followed by gastrointestinal anomaly. In central nervous system anencephaly was commonest abnormality. sensitivity of ultrasonography was 74.03% and specificity was 98.86%.

**Conclusions:** Every mother should undergo early anomaly. Fortification of food with iron, folic acid, iodine should be carried out. Anemia should be tackled early in reproductive age group. Maternal perinatal infection can be prevented through vaccination and if identified, diagnosed and treated early, would also help in decreasing congenital anomalies in baby.

**Keywords:** Congenital anomaly, Antenatal diagnosis

### INTRODUCTION

Human evolution from a single cell, 'zygote' to a multi cellular organism, is an intricate and a complex process. The birth of a malformed baby is an unfortunate event for any family and equally for the society too. Influence of teratogens in the form of pathogens, chemicals, environmental pollution and use of drugs by the mothers indiscriminately in their day to day life, have resulted in an increased incidence of congenital abnormalities in the newly born children.<sup>1</sup> Congenital anomalies are the vital

causes for perinatal mortality and morbidity. Therefore, an antenatal diagnosis and foetal therapy have attained importance in the field of human embryology. According to Dolk, "Environmental factors include any non - genetic factor that increases the risk of a birth defect for the exposed individual."<sup>2</sup>

Such factors are nutritional excesses or deficiencies (e.g. folic acid), maternal illnesses or infections, drugs which are taken during pregnancy, chemical exposure in the workplace or home and radiation." Scientific literature is

interested in the association between congenital anomalies and the possible role of chemical contaminants, and fetuses are thought to be a further subgroup of the population who could be vulnerable to the effects of air pollutants.<sup>1,3</sup> Many structural abnormalities in the fetus can be reliably diagnosed by an ultrasound scan, and these can usually be made before 20 weeks. Common examples include hydrocephalus, anencephaly, myelomeningocele, achondroplasia and other dwarfism, spina bifida, exomphalos, Gastroschisis, duodenal atresia and fetal hydrops.

With more recent equipment, conditions such as cleft lips/palate and congenital cardiac abnormalities are more readily diagnosed and at an earlier gestational age. The third one, could be done after the 26<sup>th</sup> week of pregnancy. An ultrasound examination will not, of course detect chromosomal anomalies, inherited disorders of metabolism or gene defects such as sickle cell disease.

Some congenital abnormalities can be curable if they are detected early in the antenatal period (e.g. Cardiac anomalies). In - utero surgical interventions have been made possible by the advancement in the field of medicine. Pregnant women who carry anomalous fetuses can be counseled regarding the foetal anomalies and they can be sent to neonatal paediatricians for an early management or they can be advised to go for termination of pregnancy if the anomalies are of an incurable variety (e.g. anencephaly).

In view of the above, authors were prompted to take up the present study of congenital Anomalies in fetus during pregnancy” which involve all pregnant patient coming to Rural medical college and hospital. Apart from determining the incidence of congenital foetal abnormalities, such a study might bring to light certain factors which could possibly play an aetiological role in the production of foetal anomalies.

The aim of study is to review the overall frequency of congenital malformation and to increase public awareness in saving newborns from birth defects.

**To study**

- 1) Frequency of congenital anomalies
- 2) Most common congenital anomaly
- 3) Etiology of congenital malformation
- 4) Management of pregnancy and their follow up

**METHODS**

The present study was undertaken in a large multidisciplinary referral institute Rural medical college and hospital attached to a medical college. This was observational study and duration of the study was 2 years. The total number of confinements in 2 years were recorded.

**Inclusion criteria**

- All mothers who are diagnosed antenatally on ultrasonography with birth defect and delivered at Rural medical college and hospital
- All mothers who delivered babies with congenital birth defects at Rural medical college and hospital
- Babies admitted in postnatal ward with congenital birth defects
- Abortus showing congenital anomalies.

The following details were noted.

**Maternal details**

Every patient was evaluated closely and in detail. Age of the mother, consanguinity, past obstetric history, history of medical illness specifically viral infections in the form of rash/ fever in first trimester was noted. Details of intake of any drug intake, radiation exposure, family history was specifically noted. Her nutritional status, pallor was noted. Antenatal papers were screened for blood sugars level, blood pressure in antenatal period. Special note of ultrasonography findings for congenital anomalies, oligohydramnios, polyhydramnios was made in every patient. Associated medical and surgical illness was noted.

**Perinatal information**

Assessment as to whether pregnancy was terminated or continued after diagnosis of congenital anomaly was made. Mode of delivery, fetal outcome and management required postdelivery was noted. Foetal outcome in terms of weight, sex and type of congenital anomalies were noted.

Foetuses were followed, and note was made about their survival.

**RESULTS**

A total of 162 cases of various congenital anomalies were studied from November 2013 to May 2015.

**Table 1: Total number of confinements with congenital anomalies detected / missed on USG.**

<b>Total confinements</b>	<b>16170</b>
Total live birth	14814
Total abortions and ectopic pregnancies	980
Total still births	376
Total number of congenital anomalies detected	162
Total number of congenital anomalies detected on USG	114
Total anomalies missed on USG	40
Total number cases with congenital anomalies in which USG was not done	08

**Table 2: Distribution of cases.**

	Frequency	Percent
<b>Consanguinity</b>		
Consanguineous	19	11.73
Non-consanguineous	143	88.27
<b>Age of mother</b>		
≤20 years	4	2.47
20-30 years	148	91.36
>30 years	10	6.17
<b>Parity</b>		
Primi	68	41.98
Multi	94	58.02
<b>Antenatal registration</b>		
Registered	98	60.49
Unregistered	64	39.51
<b>Sex of fetus</b>		
Female	88	54.32
Male	74	45.68
<b>Gestational age</b>		
<20 weeks	81	50.0
20-27.6 weeks	15	9.26
28-36.6 weeks	26	16.05
≥37 weeks	40	24.69
<b>Outcome</b>		
Live	62	38.27
Still birth	19	11.73
Abortion	81	50.0

Total number of confinements during period 2 years were 16170. Of which 14814 were live birth, 980 were abortions and ectopic pregnancies and 376 were still birth. Of the above confinements a total number of pregnancies with congenital anomalies were 162,

detected either prenatally or after birth i.e. an incidence of 10.94/1000 Live Birth. Out of 162 anomalies, 114 congenital anomalies were prenatally diagnosed, whereas 40 anomalies were missed on ultrasonography. 8 cases did not have antenatal USG. Out of the 16170 confinements 9860 mothers had done antenatal USG scan.

**Table 3: Distribution of study group as per maternal high-risk factor.**

Congenital anomaly	Frequency
<b>Elderly mother</b>	
Hydrocephalus	11
Duodenal atresia	3
CTEV	3
Cleft lip and palate	3
<b>Family history</b>	
Achondroplasia	6
Hydrocephalus	8
Skeletal dysplasia	1
Scoliosis	2
<b>Severe anemia</b>	
Anencephaly	3
Hydrocephalus	2
CTEV	1
Hydrops fetalis	1
Hirschsprungs disease	1
<b>Chronic malnutrition</b>	
Neural tube defect	6
Cleft lip and palate	2
Hydrops fetalis	1

**Table 4: Distribution of congenital anomaly as per system involved.**

Anomaly found	Diagnosed on USG	Missed on USG	Total	Anomaly found	Diagnosed on USG	Missed on USG	Total
<b>Central Nervous system</b>				<b>Gastrointestinal system</b>			
Anencephaly	25	0	25	Imperforate anus	0	4	4
Hydrocephalus	18	1	19	Ileal atresia	4	2	6
Neural tube defect	6	1	7	Congenital pyloric stenosis	3	2	5
Choroid plexus cyst	5	0	5	Hirschsprungs disease	4	0	4
Corpus callosal agenesis	1	3	4	Intestinal obstruction	3	0	3
Arnold Chiary malformation	2	0	2	Omphalocele	6	0	6
Holoprosencephaly	3	0	3	Gastroschisis	6	0	6
NTD with Arnold Chiary malformation	1	0	1	Duodenal atresia	2	0	2
<b>Total</b>	61	5	66	Midgut malrotation	1	1	2
<b>Skeletal system</b>				<b>Renal system</b>			
Cleft lip and cleft palate	1	16	17	Esophageal atresia	1	0	1
Congenital dislocation of Hip	7	0	7	<b>Total</b>	30	9	39
CTEV	5	0	5	Dilatation of pelvicalyceal system	7	0	7
Achondroplasia	4	0	4	Renal cyst	4	0	4
Calcaneovalgus deformity	2	0	2	Single kidney	4	0	4
Congenital vertical Talus	0	1	1	PUJ obstruction	6	0	6
Others	3	7	10	<b>Total</b>	21	0	21
<b>Total</b>	22	24	46				

**Table 5: Postnatal Management required.**

Postnatal management required	Frequency	Percentage
Active management	21	12.96
Conservative management	40	24.69
No active management	10	6.17
No management (dead)	91	56.17
Total	162	100

Out of 162 congenital anomalies, 21 anomalies required active management in the form of surgery or medical management. 40 cases were managed conservatively 10 cases were observed and followed up subsequently, did not require active management.

## DISCUSSION

Preeclampsia leads to increased perinatal morbidity and Birth defects include abnormalities in the new born baby's structure, function or body metabolism which usually lead to physical and mental disabilities and can even be fatal sometimes. Congenital anomalies are important causes of still births and infant mortality, and are contributors to childhood morbidity.

Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.<sup>4</sup>

In India, birth defect prevalence varies from 61-69/1000 live birth which is higher as compared to developed countries.<sup>5</sup>

In study conducted by Sarkar et al prevalence of congenital anomaly was 22.16/1000 live births.<sup>6</sup> In study conducted by Amar Taksande incidence of congenital anomaly was 19.1/1000 live births. In present study 162 congenital anomalies were reported in span of 2 years. Incidence rate of congenital anomaly was 10.94/ 1000 live births. Antenatally diagnosed cases were 114 (74.03%)

Consanguinity was observed in 19 cases (11.73%) of the total 162 (Table 2)

Consanguineous marriages being more common in Indian culture, increases the risk of congenital malformation. Hence premarital counseling is required.

Majority of mothers of babies with congenital anomaly belonged to age group 20-30 years (91.33%) (Table 2). In a study conducted by Sarkar et al 55.7% belonged to age group of 21-30 years.<sup>6</sup>

Majority of cases were observed in multiparous mothers (57.9%), as compared to Primipara (42.1%) (Table 2). In a study conducted by Taksande et al<sup>7</sup> Incidence of congenital anomalies was found to be more common in

multipara, Sarkar et al also found the incidence of congenital anomalies more common in multipara as compared to primipara.<sup>6</sup> Multiparous mothers should be screened vigilantly with antenatal detailed congenital anomaly scan, as more number of congenital anomaly are found in multiparous woman.

Out of 162 congenital anomalies 60.49% were antenatally registered whereas 39.51% were unregistered, (Table 2). Since most of the woman seek antenatal care, more congenital anomalies were observed in registered pregnancies. Patient who are antenatally registered in mid second trimester diagnostic modality like ultrasonography may be delayed. Awareness about antenatal registration needs to be stressed for proper antenatal care and proper development of foetus, and to identify congenital anomalies at the earliest. Delivery at tertiary care center is advisable as some of the congenital abnormality require immediate medical or surgical management. Congenital anomalies were found to be more common in females than males (Table 2). Most of the babies with congenital anomalies were delivered at full term, 06 were delivered by caesarean section. Most of them were hydrocephalus having cephalo-pelvic disproportion. 81 cases were terminated before 20th week of gestation for life threatening congenital anomaly, (Table 2). 25 of them had anencephaly, (Table 4).

Sarkar et al had observed more number of cases in babies born prematurely.<sup>6</sup>

Most of the babies with congenital anomalies had normal birth weight. This observation is in contrast to the result obtained by Sarkar et al.<sup>6</sup> Present study result correlates well with the finding made by Amar taksande<sup>7</sup> et al. 38.27% of the cases with congenital anomaly had live birth, (Table 2).<sup>7</sup> Taksande et al had also observed 94.2% babies of congenital anomalies being live at birth.<sup>7</sup>

Most commonly implicated high-risk factor was increased maternal age. Mothers aged 30 years and above were 10 in number. Sugunabai et al reported a higher incidence of malformations in the babies born to mothers aged over 35 years, whereas Datta et al documented statistically insignificant association of increased maternal age and congenital anomalies.<sup>8,9</sup> Family history was found to be significant high-risk factor in 6 cases of short stature. Carrier frequencies for various genetic disorders like thalassemia, sickle cell anemia, and metabolic disorder are high among Indians. Severe anemia and chronic malnutrition were found to be significant high-risk factor in neural tube defects, (Table 3). Maternal deficiencies of iodine and folic acid and other macro and micro nutrient found to be associated with birth defects. Sensitivity of ultrasonography in diagnosis of congenital abnormality was 74.03%.

Most common system involved in congenital anomaly was Central nervous system (CNS) followed by gastrointestinal anomaly. In study conducted by Indian

council of medical research (ICMR) most common anomaly was cardiovascular anomalies followed by musculoskeletal system (Table 4).<sup>10</sup> Neural tube defects was the commonest anomaly found in our study, (Table 4) implying need for improving nutritional status of woman in reproductive age group and increased use of periconceptional use of folate. In central nervous system anencephaly was commonest abnormality, all of them were diagnosed prenatally by USG. Other anomalies like Arnold chiary malformation, hydrocephalus were also diagnosed prenatally on USG, (Table 4). Cleft lip and palate were the commonest skeletal abnormality followed by congenital dislocation of hip and CTEV, (Table 4).

Epispadiasis was commonest abnormality in genitourinary system, followed by cryptorchidism. None of them were picked up on USG, (table no 4). Dilatation of pelvicalyceal system was commonest renal anomaly, all of them were diagnosed on USG. Imperforate anus and ileal atresia were commonest gastro-intestinal anomaly, required operative intervention.

Congenital diaphragmatic hernia and choanal atresia was commonest respiratory anomaly.

## CONCLUSION

This observational study was carried out in a rural medical college over a period of 23 months.

As cause of congenital anomalies was unknown in 82.1% of the cases in our study, every mother should undergo early anomaly scan (early second trimester) and delivery at tertiary care centre is advisable as some of the congenital anomalies require immediate surgical or medical management.

Although 60.49% were antenatally registered most of them were registered after 20<sup>th</sup> week i.e. after legal gestational age of termination of pregnancy. Hence early registration as soon as pregnancy is confirmed should be encouraged so as to make an early diagnosis and selection of plan of management. Thereby reducing fetal morbidity and improving neonatal survival.

In the present study, sensitivity of ultrasonography was 74.03% and specificity was 98.86%, as per Table 2. Ideally basic ultrasound examination should be done in all the patients. More detailed ultrasonographic study (Targeted USG) should be done in high risk group .

Fetal echocardiography is directed towards selected pregnancies carrying a higher than normal risk of fetal cardiac anomalies

Consanguinous marriages are still common in Indian culture, as per Table 3, increasing the risk for congenital malformations. Proper premarital counselling and carrier detection for common inherited disorders. The knowledge and awareness regarding the availability of

various other diagnostic modalities including serum markers, amniocentesis, chorion villus sampling, cordocentesis should be given to the patient. Most of the congenital anomalies are preventable by proper premarital counselling, periconceptional care in the form of improving maternal nutrition, folic acid supplementation, avoidance of teratogenic agents. Most of the anomalies can be picked up by fetal anomaly scan early mid trimester and life-threatening anomalies can be terminated. Attempts to prevent congenital anomalies should be initiated in preconceptional period and continued through the antenatal period upto birth of baby. High risk patients should be counselled and prepared in pre and periconceptional period.

Maternal anemia and malnutrition pose a serious threat to both mother and fetus, hence fortification of food with iron, folic acid, iodine should be carried out. Anemia should be tackled early in reproductive age group.

Maternal perinatal infection can be prevented through vaccination and if identified, diagnosed and treated early, would also help in decreasing congenital anomalies in baby.

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