

ORIGINAL ARTICLE

Profile of various congenital anomalies in fetus detected during antenatal period among high risk mothers by Ultrasonography at Tertiary Care Centre, South Gujarat

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ABSTRACT

Background: With the improvement of basic maternal and child health care services, the deaths due to malnutrition and infections are on a descending trend and due to westernization and lifestyle modifications, perinatal deaths due to anomalies are in ascending trend. **Aim & Objectives:** The purpose of this study was to estimate the antenatal proportion of congenital anomalies, types, pattern of anomalies and factors associated with anomalies in Tertiary Care Centre, South Gujarat. **Settings and Design:** Cross-sectional study in Department of Fetal Medicine of New Civil Hospital, Surat. **Methods and Material:** Sample size was 435. First 10 consecutive high-risk ANC patients, who were referred to Fetal Medicine department from Obstetrics and Gynecology department per day were selected. Data was collected during September 2021 to February 2022. **Statistical analysis used:** Frequencies, proportions and tests of significance was applied for testing the association of socio-demographic variables, clinical variables etc on presence or absence of congenital anomalies. **Results:** Proportion of different types of congenital anomalies in high risk pregnant mother was 17.2%. There was significant association between presence of congenital anomalies with history of irregular menstruation (p value: 0.001, OR: 5.19, [CI: 1.76-15.27]) and inadequate amniotic fluid (p value: 0.00, OR: 14.5[CI: 7.23-29.06]). Among total detected anomalies in Ultrasonography scanning, most common anomalies were Cardio-vascular system anomalies (36.4%) and Central Nervous System anomalies (24%). **Conclusions:** Congenital anomalies were present in about 17 % of high risk antenatal mothers.

KEYWORDS

Congenital Anomaly, Ultrasonography, High Risk Pregnant Women

INTRODUCTION

World Health Organization reported that birth defects are estimated to affect one in every 33 infants globally (1). The antenatal prevalence

of congenital anomalies was 52.1 per 1000 pregnancies (2). Birth defects affect 6% of all children in India, which translates to a staggering 1.7 million birth defects

annually(3).Congenital anomalies were the second largest cause of neonatal deaths. The congenital anomaly prenatal diagnosis prevalence was 10.98 per 1000 births and the congenital anomaly termination of pregnancy rate was 4.39 per 1000 births(4). As per NFHS 4- data, the proportion of pregnancies with an ultrasound test increased from 24 percent to 61 percent. Ultrasound tests are particularly common in urban areas (5). There are areas in India where a pregnant woman does not get even a single ultrasonography throughout pregnancy. High risk cases and fetal anomalies are only detected when women are delivering. The social support system in many societies is poor, bringing up a child with certain type of congenital deformity is a major burden for the parents and family (6). In cases where primary prevention is not possible, prenatal diagnosis by ultrasonography provides the next best alternative. Present study was conducted with the objectives to estimate the proportion, types and risk factors of congenital anomalies of fetus detected during antenatal period among high risk pregnant women by ultrasonography at Tertiary Care Centre, South Gujarat

Aim & Objective

1. To estimate the proportion of congenital anomalies of fetus detected during antenatal period among high risk pregnant women by ultrasonography at Tertiary Care Centre, South Gujarat.
2. To document types these congenital anomalies of fetus.
3. To document associated risk factors with these congenital anomalies.

MATERIAL & METHODS

It was a cross-sectional study done in Department of Fetal Medicine, New Civil Hospital, Surat. Approximately 100 ANC patients come for routine ANC checkup in tertiary care center daily. Considering working days, approximate 2500 ANC patients come for routine ANC checkup per month. Thus, approximately 30,000 ANC patients come in one year as per hospital record of previous year.

Among 100 patients, every day 20 to 25 high risk ANC patients are referred from Obstetrics and Gynecology department to fetal medicine department for antenatal scan. Among them, 2 to 3 ANC patients/ mothers are detected to have congenital anomaly of their fetus in ultrasonography scan per day as hospital records. Thus, prevalence of congenital anomalies is 12% among referred high risk patients. As per this prevalence, Sample size calculated was 435, with 95% Confident Interval and 80% power, as per Open-epi software calculations. First 10 consecutive high-risk ANC patients who are referred to Fetal Medicine department from Obstetrics and Gynecology department per day were selected for the study.

ANC patients who are referred from Obstetrics and Gynecology department to Fetal Medicine department for antenatal scan were included in study. Data was collected during September 2021 to February 2022. Written consent was taken from participant before inclusion in study. Outcome parameter was proportion of patients in which congenital anomalies had been detected by ultrasonography, proportion of types, pattern of different types of congenital anomalies (Cardiac anomaly, CNS anomaly, GI anomaly), Chi-Square test, Odd ratio for association between risk factors and anomalies. Study was approved by Scientific Review Committee and Ethical Committee of institute

RESULTS

Total sample size was 435. Proportion of different types of congenital anomalies in high risk pregnant mother was 17.2% (Figure 1).

Figure1: proportion of congenital anomalies among high risk ANC mothers

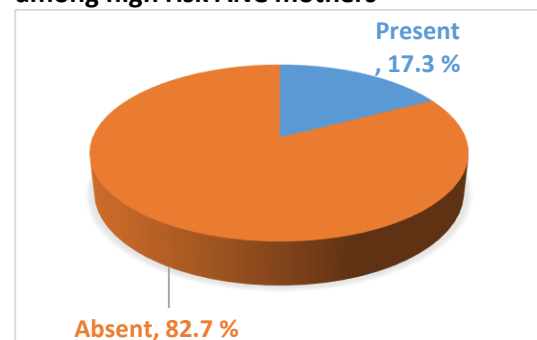


Table 1: Relation between congenital anomalies and various risk factors (n: 435)

Sr No.	Variables	Category	Congenital anomalies		Chi-square value	p value	Odds ratio	Confidence level (95% confidence limit)
			Present	Absent				
1	Age of mother	More than 35 yrs	2	28	2.52	0.1	3.07	0.83-19.53
		Less than 35 yrs	73	332				
2	Religion of mother	Hindu	58	310	3.67	0.055	1.81	0.95-3.33
		Muslim	17	50				
3	Occupation of mother	Household	283	66	3.45	0.06	0.5	0.22-1.05
		Working	77	9				
4	History of consanguineous marriage	Yes	2	6	0.01	0.7	1.61	0.31-8.16
		No	73	354				
5	Mode of conception	Artificial	1	11	0.19	0.3	0.42	0.05-3.37
		Natural	74	349				
6	Week of Amenorrhoea	1st Trimester	1	10	2.57	0.2	NA	NA
		Second Trimester	63	272				
		Third Trimester	11	78				
		Irregular	7	7				
7	Menstruation history	Regular	68	353	10.87	0.001	5.19	1.76-15.27
		Irregular	7	7				
8	History of co-morbidity to mother	Present	20	55	0.2	1.5	1.43	0.80-2.53
		Absent	73	287				
9	Amniotic fluid	Adequate	46	345	81.25	0	14.5	7.23-29.06
		Inadequate	29	15				

Out of total 435 participant, 405 (93.1%) were below 35 years and 30 (6.9%) were more than 35 (high risk age group) years. Mean age of mother was 27 years with SD: 5.32 years and ranged from 18 to 39 years. Among mothers aged less than 35 years, 73 (18%) had children with congenital anomalies compared to 2 (6.7%) among more than 35 years mothers. There is no significant association between maternal age group and presence of congenital anomalies in this study. (Chi-square=3.1, p value: 0.07, OR- 3.07)

In this study, 368 (84.6%) pregnant women followed Hindu religion and 67 (15.4%) followed Muslim religion. Proportion of congenital anomalies was higher (25.4%) in children of women with Muslim religion (25.4%) compared to Hindu religion (15.8%). There was non-significant association between

religion and congenital anomalies. (Chi-square=3.67, p value: 0.055, OR-1.81).

Out of total participants, 349 (80.2%) pregnant women were doing house hold duty and 86 (19.8%) were working. Among pregnant women, who were doing house hold duty, 66 (18.9%) had child with congenital anomalies compared to 9 (10.5%) in working mother. There is a non- significant association between maternal occupation and present of congenital anomalies in child in the present study. (Chi-square=3.45, p value: 0.06, OR- 0.5)

Out of total women, 8 (1.8%) women had history of consanguineous marriage and all of them belonged to Muslim religion. Among pregnant women following Muslim religion, 8 (12%) had history of consanguineous marriage with distant relative. Among Muslim religion

(67), 2 (25%) women had child with congenital anomalies with history of consanguineous marriage compared to 15 (25.4%) women without history of consanguineous marriage. Thus, similar proportion of children had congenital anomalies in Muslim religious with and without consanguineous marriage in present study.

Among participants, 12 (2.8%) women had history of artificial mode of conception. 1 (8.3%) child had congenital anomaly in women with history of artificial mode of conception compared to 74 (17.5%) children who had congenital anomalies in women with history of natural mode of conception. There was no significant association between mode of conception and congenital anomaly in present study (Chi-square: 0.3, p value: 0.4, OR: 0.42). Majority of the congenital anomalies were detected in second trimester (77%). Some of the congenital anomalies were also detected at 3rd trimester (20.5%). Anomalies detected at first trimester was just 2.5%, as USG for detection of anomalies is advised during second trimester of pregnancy by the gynecologists.

Out of total study participants, 14 (3.2%) had history of irregular menstruation. Presence of congenital anomalies among the child with history of irregular menstruation was 50% compared to normal menstruation (16.2%). There is significant association between congenital anomalies and history of irregular menstruation in present study (Chi-square=10.87, p value: 0.001, OR- 5.19). Thus, odds of

developing fetal congenital anomalies among women with irregular menstruation was 5.19 times higher than women with regular menstruation cycle.

Out of total study participants, 93 (21.4%) had history of co-morbidity like Diabetes, Hypertension, Thyroid and intake of medication history. Of the women with history of co-morbidity, 20 (26.7%) had fetus with congenital anomalies compared to women without history of co-morbidity (16.1%). There is no significant association between women having presence of co-morbidity and presence of congenital anomalies in child (Chi-square: 0.2, p value: 1.5, OR: 1.43).

Out of total, USG detected inadequate amniotic fluid among 44 (10.1%) women. Women with inadequate amniotic fluid in USG finding had 65.9% fetus with congenital anomalies compared to women with adequate amniotic fluid (11.8%). This difference is significant. Thus, there was significant association between amount of amniotic fluid and presence of congenital anomalies as per this study (Chi-square=81.2, pvalue: <0.00, OR: 14.5). Odds of developing fetal congenital anomalies among women with inadequate amniotic fluid was 14.5 times higher than women with adequate amniotic fluid. Among total detected anomalies in USG scanning, most common anomalies were Cardio-vascular system anomaly (36.4%) and Central Nervous System anomalies (24%)(Figure 2).

Figure 2: Different types of congenital anomaly detected in USG scan

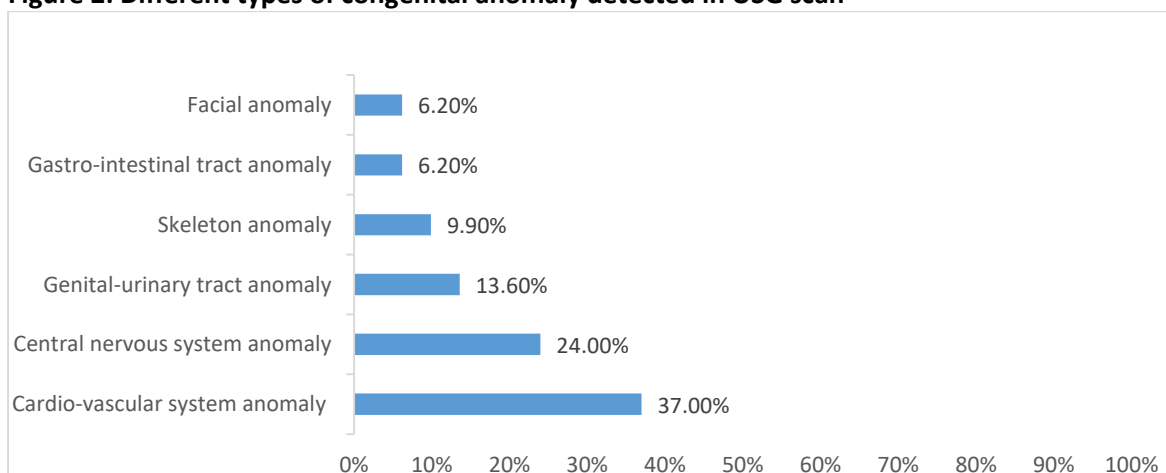


Table 2: Types of different anomalies detected in different system (n=75):

CVS anomalies: 28 cases	CNS anomalies: 18 cases	GUT anomalies: 10 cases	Skeleton anomalies: 8 cases	Facial anomalies : 5 cases	GIT anomalies: 4 cases	RS anomalies: 2 cases
VSD: 16 cases	Anencephaly : 5 cases	Renal agenesis: 3	Talipes: 2	Absent nasal bone: 2	Omphalocele : 2	Kartagener syndrome
TOF, TA: 2 cases	Spina bifida: 4 cases	VUR	Club foot: 2	Cleft palate: 2	Meckel's diverticulum:	Congenital Diaphragmati c Hernia
Pericardial effusion: 2 cases	Agenesis of corpus luteum: 3 cases	Ectopic kidney	Tibia/femur shortening	Dysmorph ic face	Anorectal malformatio n	
Single umbilical artery	Choroid plexus cyst: 2 cases	Dysplastic kidney	Tetraphaco melia			
Single echogenic focus in left ventricle:	Encephaloce le: 2 cases	Bicornuate uterus	Sacral dysgenesis			

Table 2 describes different types of congenital anomaly among different systems detected by USG scanning of high risk ANC mother. Among Cardio-vascular system, most common anomaly were VSD, TOF, TA and Pericardial effusion. Among Central Nervous system, most common anomaly were Anencephaly and Spina bifida. Renal agenesis is also present in 3 (4%) fetus with congenital anomaly.

DISCUSSION

Study was conducted in Tertiary Care Centre of South Gujarat, in which more than 100 ANC patients come daily for routine checkup. As per data record, 20 to 25% high risk mother are normally referred to Department of Fetal Medicine for USG evaluation. Total 435 women from Fetal Medicine department were included in present study. Out of them, proportion of different types of congenital anomaly among fetus was 17.2% and mean age of mother at diagnosis was 27 years. In a study done by Tella Sunitha et al in South India, it was found that 11% pregnant women were carrying fetuses with congenital anomaly among High risk mother (6). Akinmoladun et al study found that a total of 418 high risk pregnant women underwent detailed fetal anomaly scan and congenital anomalies were detected in 13(3.1%) of them. The prevalence of high risk pregnant women is high in study conducted at South Western Nigeria but the

incidence of congenital anomaly among them is similar to that reported in the general population(7). This difference due to both studies conducted within short time frame with small sample size and different geographical location. Multicentric large representative study is needed for more specifications.

As per Bahauddin Sallout et al in their study in Saudi Arabia, the antenatal prevalence of congenital anomalies was 52.1 per 1000 pregnancies among general ANC mother(2). Shreshtha et al study conducted at Tertiary Care Centre found that the prevalence of congenital malformation was 2.6% of total live births(8). Proportion of congenital anomaly was high among high risk mother than general ANC mother.

In present study, religion, occupation like working or housewife, consanguineous marriage, irregular menstruation and amount of amniotic fluid like adequate and inadequate were significant associated risk factors for presence of congenital anomaly among fetus in USG scanning. Among them, irregular menstruation (OR: 5.19) and amount of amniotic fluid like adequate and inadequate (OR:14.5) were significantly related to congenital anomaly among fetus in USG scanning. In study done by Tella Sunitha et al, maternal age (<25 years, OR = 1.42, p = 0.002), paternal age (<30 years, OR = 1.51, p < 0.001),

consanguinity (OR = 1.39, $p = 0.012$) and primi gravida (OR = 3.40, $p < 0.001$) were identified as risk factors for high risk pregnant women with fetal Congenital anomaly. Marwah *et al* study found that high age and parity, parental consanguinity, bad obstetrical history, inadvertent drug ingestion enhanced risk are risk factors for congenital anomaly at birth(9). Abnormal intrauterine development occurs because of the interference of normal development due to many factors like genetic, environmental, Nutritional deficiencies /insufficiencies (e.g. folate), Maternal age, Maternal illnesses (e.g. diabetes, hypothyroidism), Infectious diseases (e.g. rubella, syphilis), Alcohol abuse, Obesity, Tobacco use. This leads to abnormal cytogenesis, histogenesis and morphogenesis with which the neonate born with a defect known as a congenital anomaly (CA). Present study is observational study. In this study no interventions were done like laboratory investigations eg estimating folic acid level in blood, test for rubella, syphilis, sugar level. Thus, many other interventional risk factors were not studied. In present study, 12 (2.8%) women had history of artificial mode of conception. There was no significant association between mode of conception (natural and artificial) and congenital anomaly. Majority of patients were from lower socio-economic condition. Thus, majority of women had natural mode of conception. Very less number of women with artificial mode of conception were included in present study. In present study, most common anomaly were Cardio-vascular system anomaly (36.4%) and Central Nervous System anomaly (26%). In, Akinmoladun *et al* study, the highest number of anomalies were detected in the genitourinary system while the least was in the central nervous system(7). Tella Sunitha *et al* study found that the major CAs observed were Central Nervous System (CNS) followed by renal anomalies(6). Bhalerao *et al* study found the predominant system involved in Congenital anomaly was the musculoskeletal system (36.90%) followed by the central nervous system (CNS) (25%) and the gastrointestinal (GI) system (16.6%). Talipes (17.1%) was the most common anomaly in the

musculoskeletal group followed by cleft lip and cleft palate in the GI system. It was seen that majority of congenital anomalies were associated with low birth weight (LBW), prematurity, multiparity, and consanguinity(10). Different study conducted with different geographical location have similar types of congenital anomaly with various proportion.

CONCLUSION

Proportion of different types of congenital anomaly in high risk pregnant mother was 17.2% at Tertiary Care Center. There was significant association between presence of congenital anomaly with history of irregular menstruation and inadequate amniotic fluid. Among Muslim religion, there was no significant association between consanguineous marriage and congenital anomalies. Women with history of co-morbidity, 20 (26.7%) had child with congenital anomaly compare to women without history of co-morbidity (16.1%). Among total detected anomaly in USG scanning, most common anomaly was Cardio-vascular system anomaly (36.4%) and Central Nervous System anomaly (26%).

RECOMMENDATION

There is need of increased prenatal screening, awareness and education regarding risk factors and genetic counselling among high risk pregnant women. There is need of more expertise and infrastructure, particularly in the field of ultrasound scanning. This will ensure accurate and early detection of congenital anomalies.

LIMITATION OF THE STUDY

Present study was observational study. In this study no interventions were done like laboratory investigations eg estimating folic acid level in blood, test for rubella, syphilis, sugar level. Thus, many other interventional risk factors were not studied.

Limitation in time period of study. Thus sample size is small

AUTHORS CONTRIBUTION

All authors have contributed equally.

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Nil

CONFLICT OF INTEREST

There are no conflicts of interest.

DECLARATION OF GENERATIVE AI AND AI ASSISTED TECHNOLOGIES IN THE WRITING PROCESS

The authors haven't used any generative AI/ AI assisted technologies in the writing process.

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