European Journal of Clinical Pharmacy

Print ISSN: 2385-409X **Online ISSN**: 3105-0409
Website: https://farmclin.com/

Original Research Article

Frequency of Ultrasonographically Detected Fetal Congenital Anomalies among Pregnant Women with Polyhydramnios

Article History:

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Received: 19-10-2025
Revised: 16-12-2025
Accepted: 20-12-2025
Published: 30-12-2025

Abstract: **Objective:** To determine the frequency of ultrasonographically detected fetal congenital anomalies among pregnant women with polyhydramnios. **Study Design:** Cross sectional study. **Place and Duration of Study:** Conducted from 16 April 2025 to 16 October 2025 at Department of Radiology, Liaquat University of Medical and Health Sciences, Jamshoro. **Methodology:** Total 73 pregnant women with polyhydramnios were included. Polyhydramnios was diagnosed when amniotic fluid index was ≥24 cm or deepest vertical pocket was ≥8 cm on ultrasound. Mean and standard deviation was calculated for quantitative variables. Frequency and percentage was calculated for qualitative variables. Chi square test and Fisher exact test was applied after stratification. P-value <0.05 was taken as statistically significant. **Results:** Mean maternal age was 28.04 ± 6.41 years and mean gestational age was 33.42 ± 3.71 weeks. Consanguinity was present in 35 (47.9%) women and family history of birth defects in 8 (11.0%). Fetal congenital anomalies was detected in 15 (20.50%) cases while 58 (79.50%) had no anomaly. No statistically significant association was found with maternal age (p=0.316), gestational age (p=0.748), parity (p=0.448), duration since marriage (p=0.145), consanguinity (p=0.294) and family history (p=0.348). **Conclusion:** Fetal congenital anomalies was found in one fifth of women with polyhydramnios. Ultrasound helps in early detection and counseling.

Keywords: Amniotic fluid; Congenital abnormalities; Polyhydramnios; Pregnancy complications; Prenatal diagnosis.

INTRODUCTION

Polyhydramnios refers to a condition that occurs during pregnancy and is associated with an abnormal increase in amniotic fluid volume surrounding the fetus [1]. This condition can usually be diagnosed by ultrasound and is associated with an amniotic fluid index greater than 24 cm or a deepest vertical pocket greater than 8 cm [1]. This condition can present at any time during the second or third trimester and can be an acute or chronic condition. The exact cause of polyhydramnios can vary and may be associated with a number of factors. These factors may include maternal diabetes mellitus, multiple gestation, anemic conditions in the fetus, and congenital abnormalities [2]. An abnormal increase in the volume of the uterus associated with polyhydramnios can cause maternal discomfort and can lead to abdominal distension, dyspnea, and preterm labor [3]. This condition also increases the risks associated with premature rupture of membranes, malpresentation, cord prolapse, and postpartum hemorrhage [4]. Considering the risks associated with polyhydramnios and its complications, it is very important to identify the cause of polyhydramnios.

Fetal congenital anomalies include structural and functional abnormalities that develop during fetal development [5]. In pregnancies with polyhydramnios, there is a high occurrence of fetal anomalies that affect swallowing. These include anencephaly, hydrocephaly, esophageal atresia, and

duodenal atresia [6]. These abnormalities impair normal swallowing and absorption of amniotic fluid, thereby causing polyhydramnios. Some chromosomal abnormalities and genetic syndromes can be associated with polyhydramnios. There is a higher occurrence of congenital anomalies in pregnancies with moderate to severe polyhydramnios compared to those with mild polyhydramnios [7].

Ultrasound imaging plays a pivotal role in the evaluation of fetal anomalies among pregnant women who present with polyhydramnios [8]. Ultrasound imaging is a safe, noninvasive, and accessible imaging tool that allows real-time imaging of fetal anatomy as well as the amount of amniotic fluid present [9]. A detailed anomaly scan can help identify significant anomalies in the brain, spine, heart, abdominal region, and limbs of the fetus [10]. In the presence of polyhydramnios, ultrasound imaging of the fetus is strongly suggested to rule out anomalies like gastrointestinal tract obstruction, neural tube defects, and other anomalies [11]. In addition, Doppler ultrasound may be of value in the evaluation of fetal anemia or heart function [12]. With the advent of high-resolution ultrasound imaging, accuracy in imaging fetal anatomy has greatly improved [12].

There is a scarcity of data concerning the prevalence of ultrasonographically detected fetal malformations among pregnant women with polyhydramnios in Jamshoro. Existing studies conducted in other regions of the world may not be a true reflection of the problem among pregnant women with polyhydramnios in Jamshoro, given the diversity in the health condition of pregnant mothers, nutritional status, diabetic condition, and access to antenatal care facilities. Therefore, this study is necessary to quantify the problem in the local setting, to increase awareness of polyhydramnios among pregnant women, to use ultrasonography in diagnosing polyhydramnios, and to counsel pregnant women with polyhydramnios for better management.

METHODOLOGY

This cross-sectional study was carried out in the Department of Radiology, Liaquat University of Medical and Health Sciences, Jamshoro from 16 April 2025 to 16 October 2025. Ethical approval was obtained from institutional ethical review committee with certificate No. LUMHS/REC/-573 dated 06-01-2025 before starting the study. Sample size was calculated by taking prevalence of fetal congenital anomalies in patients with polyhydramnios as 25.33%,13 margin of error 10% and confidence interval 95%, therefore required sample size was 73 patients. All singleton pregnant women aged 18 – 40 years, having gestational age 12 to 40 weeks assessed by date of LMP or ultrasound, presenting for fetal anomaly scan or routine antenatal ultrasound during second or third trimester were included. Patients with polyhydramnios of any parity and those who provided informed consent were enrolled. Pregnant women with multiple gestations, diabetes mellitus and intrauterine fetal death confirmed on history and documented medical record were excluded from study. Polyhydramnios was considered when deepest vertical pool was ≥ 8 cm or amniotic fluid index (AFI) was ≥ 24 cm or AFI above 95th centile for gestational age measured on ultrasound.

Informed consent was obtained from each participant before the study. The participants were asked to provide an in-depth obstetric history, and a general physical examination was done. Ultrasonographic examination was done using an ultrasound machine with a 3.5 MHz convex transducer. The results were noted at the time of examination, and the presence or absence of fetal congenital anomalies was noted by the researcher. Fetal congenital anomalies were defined as structural or chromosomal abnormalities detected by ultrasonography, including Down syndrome, which is associated with trisomy 21; trisomy 18; trisomy 13; and other chromosomal disorders, as well as structural anomalies of the abdominal wall and internal organs. The cases with such abnormalities were classified as positive for fetal congenital anomalies, and those without such abnormalities were classified as negative.

Data was entered and analyzed using SPSS version 21.0. For maternal age, gestational age, duration since marriage, parity and gravidity mean and standard deviation were calculated. Frequency and percentage were calculated for qualitative variables such as consanguinity, history of birth defects in family and fetal congenital anomalies. Effect modifiers were controlled through stratification with respect to maternal age, gestational age, parity, gravidity, duration since marriage, consanguinity, history of birth defects in family and socio-economic status. Chi-square test or Fisher exact test was applied where appropriate. If expected frequency was ≤ 0.05, Fisher exact test was applied. P-value < 0.05 was taken as statistically significant.

RESULTS

The total study sample was 73 pregnant women with polyhydramnios. The mean maternal age was found to be 28.04 ± 6.41 years, while the mean gestational age at the time of examination was 33.42 ± 3.71 weeks. The mean parity among the study participants were recorded as 2.16 ± 1.68, and the mean duration since marriage was 7.66 ± 5.93 years. With regard to consanguinity, 35 women (47.9%) were reported to have consanguineous marriage while 38 (52.1%) were not. A history of birth defects in family was present in 8 (11.0%) of the patients, whereas majority 65 (89.0%) had no such history (Table 1).

Table 1. Patient Demographics

Demographics	Mean ± SD
Maternal Age (years)	28.04 ± 6.41
Gestational Age (weeks)	33.42 ± 3.71
Parity	2.16 ± 1.68
Duration since marriage (years)	7.66 ± 5.93
Consanguinity	
Yes n (%)	35 (47.9%)
No n (%)	38 (52.1%)
H/O Birth Defects in Family	
Yes n (%)	8 (11.0%)
No n (%)	65 (89.0%)

With regard to the frequency of ultrasonographically detected fetal congenital anomalies among the pregnant women with polyhydramnios, it was observed that 15 (20.50%) of the cases were having fetal congenital anomalies, while the remaining 58 (79.50%) were not showing any anomaly out of the total 73 cases examined (Table 2).

Table 2. Frequency of Ultrasonographically Detected Fetal Congenital Anomalies Among Pregnant Women with Polyhydramnios (N=73)

Fetal Congenital Anomalies	Frequency	% age
Yes	15	20.50 %
No	58	79.50 %
Total	73	100%

Among women aged ≤30 years, fetal congenital anomalies was present in 8 (17.0%) cases, whereas in women aged >30 years, it was found in 7 (26.9%) cases, with the p-value being 0.316. For gestational age, anomalies was detected in 12 (21.8%) of women at ≤36 weeks and in 3 (16.7%) of those at >36 weeks, with p-value of 0.748. Regarding parity, anomalies was seen in 11 (18.3%) of women with parity ≤3 and in 4 (30.8%) of those with parity >3, and the p-value was 0.448. In terms of duration since marriage, anomalies was present in 4 (11.8%) of women married for ≤5 years and in 11 (28.2%) of those married for >5 years, with p-value of 0.145. Among women with consanguineous marriage, anomalies was observed in 9 (25.7%) cases, compared to 6 (15.8%) in those without consanguinity, and the p-value was 0.294. Finally, among women with a positive family history of birth defects, anomalies was detected in 3 (37.5%) cases, while among those with no such history, it was found in 12 (18.5%) cases, with p-value of 0.348 (Table 3).

Table 3: Association of Fetal Congenital Anomalies with Demographic Factors

Demographic Factors	Subgroup	Fetal Congenital Anomalies		p-value
		Yes n(%)	No n(%)	
Age (years)	≤30	8 (17.0%)	39 (83.0%)	0.316**
	>30	7 (26.9%)	19 (73.1%)	
Gestational Age (weeks)	≤36	12 (21.8%)	43 (78.2%)	0.748*
	>36	3 (16.7%)	15 (83.3%)	
Parity	≤3	11 (18.3%)	49 (81.7%)	0.448*
	>3	4 (30.8%)	9 (69.2%)	
Duration since Marriage (years)	≤5	4 (11.8%)	30 (88.2%)	0.145*
	>5	11 (28.2%)	28 (71.8%)	
Consanguinity	Yes	9 (25.7%)	26 (74.3%)	0.294**
	No	6 (15.8%)	32 (84.2%)	
H/O Birth Defects in Family	Yes	3 (37.5%)	5 (62.5%)	0.348*
	No	12 (18.5%)	53 (81.5%)	

*Fischer Exact Test **Chi-square Test

DISCUSSION

In the present study, fetal congenital anomalies were detected among 15 of 73 pregnant women with polyhydramnios, accounting for 20.50%. This is clinically important, indicating that one in five women with polyhydramnios may be at risk of having a fetus with a congenital anomaly. The mechanism by which polyhydramnios is linked to fetal anomalies is through excessive amniotic fluid volume, which occurs due to decreased fetal swallowing, usually because of gastrointestinal, neuromuscular, or craniofacial abnormalities. These abnormalities interfere with the normal mechanism of fluid reabsorption, thereby resulting in increased amniotic fluid volume, making polyhydramnios an important

clinical indicator of possible fetal anomalies. In the present study, 35 women (47.9%) were found to be consanguineous, among whom fetal anomalies were detected in 9 women, accounting for 25.7%, while among the non-consanguineous women, fetal anomalies were detected in 6 women, accounting for 15.8%. The p-value was 0.294. Although this difference was not significant statistically, the higher percentage of anomalies observed in the consanguineous group may have a scientific rationale, as consanguineous marriages are known to result in homozygous gene mutations of autosomal recessive genes, which are well established to cause various structural and metabolic anomalies in the offspring. In addition, women who had a family history of birth defects had a higher percentage of anomalies compared to those who did not have a family history of birth defects, i.e., 3 (37.5%) compared to 12 (18.5%), with a p-value of 0.348. This also supports the fact that birth defects are genetically determined, as it is observed that familial clustering of birth defects may result from genetic mutations that may be present in the family, which may be passed on to the offspring, increasing their susceptibility to birth defects in subsequent pregnancies.

The frequency of fetal congenital anomalies detected ultrasonographically in the present study was 15 (20.50%) among pregnant women with polyhydramnios. This finding is somewhat comparable to the results reported by Tariq et al. 14 who found fetal anomalies in 26 (31.7%) cases, and by Ismat et al. 15 who reported congenital malformations in 22 (14.7%) cases, and by Ayub et al. 16 who detected anomalies in 14 (10%) fetuses. The variation in these frequencies may be explained by differences in study sample size, gestational age at time of ultrasound examination, severity of polyhydramnios included, and the level of sonographic expertise available at different centers. Higher frequencies was reported by Zia et al. 17 who found fetal anomalies in 50.8% cases, and by Mughal et al. 18 who detected anomalies in 30% patients, and by Anwar et al. 19 who reported anomalies in 41 (27.5%) cases. These higher rates may possibly be because these studies included more severe cases of polyhydramnios or had larger sample sizes, as severity of polyhydramnios is well known to directly correlates with higher likelihood of underlying fetal structural defects due to greater impairment of fetal swallowing mechanism. Regarding maternal age, the present study found no statistically significant association between maternal age and fetal congenital anomalies (p=0.316), which is in agreement with findings of Anwar et al. 19 and Ayub et al. 16 who also observed no significant association between maternal age and fetal anomalies. This similarity suggests that maternal age alone may not be an independent predictor of congenital anomalies in the presence of polyhydramnios, although advanced age is biologically associated with chromosomal errors, the polyhydramnios itself may be a stronger indicator of anomaly regardless of age group. The present study observed higher frequency of anomalies among women with consanguineous marriage, where 9 (25.7%) consanguineous cases had anomalies compared to 6 (15.8%) in non-consanguineous group, though this was statistically not significant (p=0.294). This finding has partial support from Anwar et al. 19 who reported significant association between previous history of congenital anomalies and fetal defects (p=0.016), which indirectly points toward genetic and familial contribution. Similarly, in the present study, women with positive family history of birth defects showed higher anomaly rate 3 (37.5%) as compared to those without such history 12 (18.5%), with p=0.348. The lack of statistical significance in present study may be attributed to relatively smaller sample size which reduces the statistical power to detect true associations that may exists in larger populations. The genetic explanation remains valid however, as consanguinity increases homozygosity for autosomal recessive mutations which are responsible for many structural fetal anomalies.

Limitations

However, there are certain limitations of the present study, which need to be kept in mind while interpreting the findings. Firstly, this is a hospital-based study, where only one hospital has been considered. It is possible that the findings of this study may not be generalized to the entire population of Pakistan. Secondly, there were only 73 women in the sample, and this could be a small sample, thereby reducing the ability to detect significant associations between fetal abnormalities and demographic variables. Furthermore, there has been no assessment of the severity of polyhydramnios, and this could have provided a better understanding of the relationship between polyhydramnios and fetal abnormalities. Lastly, there has been no postnatal assessment of abnormalities detected by ultrasonography, and this could have provided a better validation of ultrasonography findings.

CONCLUSION

The present study has shown that fetal congenital abnormalities are not an uncommon occurrence in pregnant women with polyhydramnios. Ultrasonography has been found to be a useful investigation in detecting such abnormalities in pregnant women. However, from the trends observed in consanguineous couples and in individuals with a positive family history of birth defects, a genetic influence in the etiology of birth defects cannot be ruled out.

Disclaimer:

Nothing to declare.

Acknowledgment:
The author thankful to the doctors and staff of the department. They kept proper records and managed patient data in organized way, which was very important for completing this research work.

Conflict of Interest:
The author declare that no competing interest is present regarding this research.

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