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Frequency of Fetal Anomalies in Pregnancy Complicated by Polyhydromnios

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ABSTRACT

Background: Polyhydramnios is a condition characterized by an excessive accumulation of amniotic fluid, defined as a deepest vertical pool measuring 8 cm or more, or an amniotic fluid index (AFI) equal to or greater than 24 cm, which corresponds to values above the 95th centile for gestational age. The AFI is determined by calculating the deepest vertical pool in each of the four quadrants of the pregnant patient's abdomen. Based on AFI values, polyhydramnios is classified into three categories: mild, with an AFI of 24–29.9 cm; moderate, with an AFI of 30–34.9 cm; and severe, with an AFI of 35 cm or more. **Incidence** 0.2%–3.9% of pregnancies and can arise from diverse maternal or fetal conditions. Idiopathic polyhydramnios, where no identifiable cause exists, poses significant diagnostic and management challenges, as it may be associated with adverse fetal outcomes and congenital anomalies. **Objective:** This study aimed to determine the frequency and types of fetal anomalies in pregnancies complicated by polyhydramnios to aid in clinical risk assessment and management. **Methods:** A cross-sectional study was conducted at Moula Bakhsh Hospital, Sargodha, from January 15 to June 15, 2025. A total of 149 pregnant women beyond 28 weeks' gestation with polyhydramnios were enrolled through consecutive sampling. Participants underwent detailed history, examination, and targeted ultrasonography to detect fetal anomalies. Data were analyzed using SPSS 25, with Chi-square tests for statistical associations. **Results:** Fetal anomalies were detected in 41 (27.5%) cases. Central nervous system anomalies (31.7%) were most common, followed by genitourinary (21.9%), and musculoskeletal (19.5%), gastrointestinal (12.2%) anomalies. Significant associations were found between fetal anomalies and history of congenital anomalies ($p = 0.016$) and polyhydramnios in previous pregnancies ($p = 0.041$), while maternal age and BMI showed no significant relationship. **Conclusion:** Polyhydramnios is associated with a high prevalence of fetal anomalies, emphasizing the need for thorough prenatal assessment. Systematic ultrasound evaluation and consideration of maternal history can enhance early detection and management of these high-risk pregnancies.

INTRODUCTION

The phenomenon of polyhydramnios is a fairly common complication of pregnancy (the incidence in reported studies varies between 0.2-3.9 percent in incidence) (Vanda, Bazrafkan et al. 2022, Pagan, Magann et al. 2023). It can be because of excess fluid production or lowered absorption. It can be caused by many maternal conditions contributing to the layering of inappropriate amount of amniotic fluid (Jha, Raghu et al. 2023, Hwang, Jenkins et al. 2024). In many patients no cause is discoverable. This is referred to as idiopathic or unexplained polyhydramnios. No particular cause attributable to their placenta, maternal, or fetal origins is detected during this condition (Kechagias, Triantafyllidis et al. 2024). The diagnosis of polyhydramnios may be upsetting to parents and hard on clinicians because although most patients have an idiopathic disorder, polyhydramnios can relate to major fetal deformity or maternal pathology (Lim, Lustestica et al. 2024, Wu and Chen 2024).

Polyhydramnios has been linked with the elevated risk of many perinatal morbidity and mortalities that include preterm birth, aneuploidy, cesarean section, fetal anomalies, and perinatal and postnatal mortality (Gurrel, Ayhan et al. 2024, Atalay, Yilmaz et al. 2025). Polyhydramnios during pregnancy can be a diagnostic and treatment challenge to obstetricians (Vanda, Bazrafkan et al. 2022, Bibi, Qayyum et al. 2023). A polyhydramnios was considered as a factor of higher risks of complications of the pregnancy by many clinicians and proposed a significant exploration of such pregnancies (Walter, Calite et al. 2022, Soni, Teefey et al. 2023).

In cases of polyhydramnios in pregnancy, a focused

ultrasonographic study should be carried out to check the existence of any abnormality in the fetus (Wang, Wu et al. 2022, Liu, Hu et al. 2024). Risk levels of underlying fetal abnormality comprising trisomy 13 or 18 are high in fetal growth restriction (FGR) (Cai, Lin et al. 2022, Beke and Simonyi 2025). The pregnant women having excessive polyhydramnios are advice to be delivered in tertiary centers where there is obstetric and pediatric assistance at the time of delivery due to high likelihood of the development of fetal anomalies (Ghaffar, Channa et al. 2024, Mangla and Anne 2024). Hence, the research explored predictors that might be used to forestall chromosomal abnormalities in a pregnant fat pregnant woman with polyhydramnios.

Qadir S et al identified the prevalence of ultrasonography visible fetal defects in polyhydramnios pregnancies. The age of the participants in the research was between 20 and 40 years with a mean of 28.87+/-4.16 years. Most of the patients 92(61.33) were age 20 to 30 years. The average gravidity was 3.01 +/- 0.99. The average time since getting married was 5.50 +/- 1.22. Frequency of ultrasound detectable fetal anomalies during pregnancy complicated by polyhydramnios was 38 (25.33%) in this study.

The present study is focused on establishing the fetal outcomes in a pregnant patient with polyhydramnios in order to give a data to the clinicians so that they can develop effective tactics to deal with the condition, and the consequences of the condition on the fetus, which can be easily dealt with provided early detection and regular follow ups could be conducted.

MATERIAL AND METHODS

This cross sectional research was carried out at the Department of Gynecology and Obstetrics, Moula Bakhsh Hospital, Sargodha between Jan 15,2025 and June 15, 2025 after institutional ethical review committee permitted the synopsis of the study. The sample profile was pregnant women who had come to the hospital with polyhydramnios after taking 28 weeks of gestation. First, participant selection was based on a non-probability consecutive sampling method. In calculating the sample size, the WHO sample size calculator was used where the significance level was set at 5 percent, approximated probability of fetal abnormalities of 25.33 percent, and margin of error at 7 percent resulting in a final sample size of 149 patients. Fetal anomalies in polyhydramnios are reportedly 0.2-3.9 %.

Inclusion criteria included women aged between 20- 40 years with singleton pregnancy diagnosed with polyhydramnios after 28 weeks of gestation. The exclusion criteria were women with pre-existing diabetic mellitus, gestational diabetes and Rhesus incompatibility. Eligible participants were assessed using a structured proforma purposefully designed to assess the study participants after consent. Maternal age, parity, history of previous congenital anomalies, polyhydramnios in past pregnancies, family history of twins or malformed babies were examined and discussed in detail. A full clinical exam was conducted, followed by general physical/abdominal examination; in case of laboring women, per vaginal examination was done too.

Confirmation and the severity of polyhydramnios was examined through use of ultrasound to determine the amniotic fluid index (AFI). Sonographically identified fetal abnormalities were categorized into six organ system organ-based, following the identification of structural and functional anomalies that occurred in the developing fetus by ultrasound irrespective of functional and structural type; -central nervous system (CNS), face and neck, cardiac, genitourinary, gastrointestinal, and musculoskeletal. The participants were tracked to delivery, and occurrence of fetal anomaly was reported according to the study operational definitions. The structured proforma was used to collect data (Annexure-I).

The statistical analysis was conducted with the help of SPSS 25. Means and standard deviations were used to describe quantitative variables including maternal age, weight, height, BMI and gestational age. Categorical variables, such as the presence of fetal anomalies, history of congenital abnormalities, previous polyhydramnios, as well as family history of twins or abnormal babies, were given in frequencies and percentages. Chi-square test was used to evaluate associations of fetal anomalies with categorical data. To adjust the possible effect modifiers of stratification, e.g., maternal age, BMI, gestational age, and other obstetric history, were controlled. The Chi-square was re-used (after post-stratification) to assess statistical significance, where a p-value of <0.05 was to be regarded as significant.

RESULTS

A total of 149 expectant women with polyhydramnios that met the inclusion criteria were recruited in a study. In this group, the occurrence of fetal anomalies was 0.2 to 3.9 percent. Average age of women was 30.4 years + 5.2 years and a gestational age at the time of presentation was 32.6 weeks + 2.3 weeks. Most of the participants (61.1%) involved multigravida. The average BMI was 27.3 kg/m 2 with standard deviation 3.6 kg/m 2. Table 1 shows the demographic, obstetrical history and examination data of the research sample.

Table 1

Baseline Characteristics of Study Participants (n = 149)

Variable	Mean ± SD / n (%)
Maternal Age (years)	30.4 ± 5.2
Gestational Age at Presentation (weeks)	32.6 ± 2.3
BMI (kg/m^2)	27.3 ± 3.6
Gravidity	
• Primigravida	58 (38.9%)
• Multigravida	91 (61.1%)
History of Congenital Anomalies	17 (11.4%)
Polyhydramnios in Previous Pregnancies	19 (12.8%)
Family History of Twins	14 (9.4%)
Family History of Abnormal Babies	12 (8.1%)

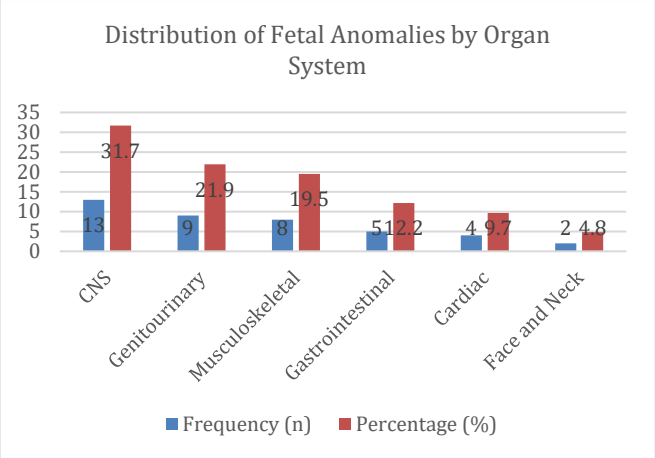
Ultrasound examination revealed fetal anomalies in 41 (27.5%) of the cases. The most commonly detected anomalies were of the gastrointestinal system (20–30%), followed by central nervous system (CNS) (31.7%), genitourinary (21.9%), and musculoskeletal (19.5%) anomalies (Table 2).

Table 2

Distribution of Fetal Anomalies by Organ System (n = 41)

Anomaly Type	Frequency (n)	Percentage (%)
CNS	13	31.7
Genitourinary	9	21.9
Musculoskeletal	8	19.5
Gastrointestinal	5	12.2
Cardiac	4	9.7
Face and Neck	2	4.8

Figure 1



A significant association was observed between fetal anomalies and history of congenital anomalies (p = 0.016), as well as polyhydramnios in previous pregnancies (p = 0.041). Maternal age, BMI, and gestational age were not found to be significantly associated with fetal anomalies. Table 3 presents the stratified analysis of fetal anomalies against selected risk factors.

Table 3

Association Between Fetal Anomalies and Selected Risk Factors

Risk Factor	Fetal Anomalies Present (n = 41)	Fetal Anomalies Absent (n = 108)	p-value
History of Congenital Anomalies	10 (24.4%)	7 (6.5%)	0.016*
Polyhydramnios in Previous Pregnancies	9 (22.0%)	10 (9.3%)	0.041*
Family History of Abnormal Babies	5 (12.2%)	7 (6.5%)	0.317
Family History of Twins	3 (7.3%)	11 (10.2%)	0.575
Maternal Age ≥ 35 years	12 (29.3%)	30 (27.8%)	0.861
BMI ≥ 30	10 (24.4%)	23 (21.3%)	0.688

*Statistically significant (p < 0.05)

DISCUSSION

The aim of this descriptive cross-sectional study was to assess the prevalence and the pattern of fetal abnormality in polyhydramnios affected pregnancies. Of the 149 pregnant women, fetal anomalies were diagnosed in 27.5% of pregnancies which is within the range of 20-35% that is stated in other research studies regarding prevalence of anomalies in comparable clinical populations. The large number of detected anomalies illustrates the strong correlation between polyhydramnios and underlying structural or functional abnormalities in the fetus, which is why complete fetal evaluation is recommended in all of the matters.

The most commonly affected organ system in this study was the central nervous system (31.7%), followed by the genitourinary (21.9%) and musculoskeletal (19.5%) systems, gastrointestinal (12.2%). These findings align with earlier reports indicating that CNS anomalies, such as anencephaly and hydrocephalus, are frequently associated with excessive amniotic fluid due to impaired fetal swallowing or neurological dysfunction. The notable presence of genitourinary anomalies, such as obstructive uropathies and renal agenesis, may also contribute to polyhydramnios via decreased fetal urine output regulation. Musculoskeletal anomalies, while less frequently discussed in the context of polyhydramnios, may impair fetal swallowing or movement, contributing indirectly to fluid accumulation.

Importantly, a significant association was found between fetal anomalies and both a prior history of congenital anomalies and a history of polyhydramnios in previous pregnancies. These associations suggest that certain maternal or familial predispositions may increase the likelihood of recurrence in subsequent gestations. However, maternal age and BMI were not significantly associated with fetal anomalies in this cohort, indicating that polyhydramnios and associated anomalies may occur independently of these demographic variables.

Ultrasound played a pivotal role in both the diagnosis and classification of fetal anomalies in this study. The use of targeted imaging allowed for early identification and monitoring, supporting the inclusion of detailed anomaly scanning as a routine component of antenatal care in cases of idiopathic polyhydramnios. Moreover, the findings reinforce the clinical value of classifying anomalies by system, which not only facilitates perinatal planning but also enables counseling regarding prognosis and potential postnatal interventions.

The study's limitations include its single-center design and non-probability sampling technique, which may restrict generalizability. Additionally, genetic investigations and postnatal confirmation of anomalies were not within the scope of this study, which could lead to underestimation or misclassification of certain conditions. Despite these limitations, the results provide valuable insights into the burden of fetal anomalies in polyhydramnios and emphasize the importance of detailed sonographic evaluation and clinical vigilance.

In conclusion, the presence of fetal anomalies in over one-fourth of pregnancies complicated by polyhydramnios highlights the need for systematic prenatal evaluation. Central nervous system, genitourinary, and musculoskeletal anomalies remain the predominant findings. Recognizing the associated risk factors, such as previous history of anomalies or polyhydramnios, can guide clinicians in risk stratification and management. Further multicenter studies, including genetic and neonatal outcome data, are warranted to better understand the etiology and improve outcomes in these high-risk pregnancies.

CONCLUSION

This study found a 27.5% incidence of fetal anomalies in pregnancies with polyhydramnios, highlighting the condition's significance as a clinical red flag. The central nervous system was most commonly affected, followed by genitourinary and musculoskeletal systems. Significant

correlations with prior congenital anomalies and previous polyhydramnios suggest that detailed obstetric histories are crucial for risk assessment. Despite no significant association with maternal age or BMI, the findings underscore the importance of targeted fetal anomaly scans in all cases of polyhydramnios. Early identification of structural abnormalities allows for informed clinical decision-making, perinatal planning, and timely interventions. Given the potential implications for fetal morbidity and mortality, management in a tertiary care setting with access to specialized obstetric and neonatal care is recommended. Further large-scale, multicenter studies incorporating genetic evaluation are warranted to enhance diagnostic accuracy and improve outcomes.

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