ASSESSING THE RELATIONSHIP BETWEEN POLYHYDRAMNIOS SEVERITY AND FETAL CONGENITAL MALFORMATIONS

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ABSTRACT

Objective: To determine the frequency of fetal malformations among pregnant women with polyhydramnios.

To determine the association between the severity of polyhydramnios and fetal congenital malformations.

Method: A descriptive cross-sectional study was conducted at Lady Reading Hospital, Peshawar over a period of six months (April-Oct, 2022). A sample size of 380 pregnant women was selected through non-probability consecutive sampling technique including pregnant women undergoing medical history, physical examination and detailed ultrasound screening. Data was collected using a proforma. IBM SPSS 24 was used to analyze the data descriptive statistics were applied and chi-square test was used to determine the association between the severity of polyhydramnios and fetal congenital malformations.

Result: 380 pregnant women were included in the study, with mean age of 28.8 years (SD ± 5.70) and mean gestational age of 34.3 weeks (SD ± 4.89). Majority of the women had primary (41.3%) or secondary (30.0%) education, 19.2% were uneducated, and 9.5% were highly educated. 42.1% were primigravida, and 57.9% were multigravida. Regarding parity, 42.1% were nulliparous, 25.5% primiparous, and 32.4% multiparous. Ultrasound revealed mild polyhydramnios in 19.7%, moderate in 47.6%, and severe in 32.6%. Fetal anomalies were present in 50.8% of cases of polyhydramnios, including gastrointestinal (12.1%), cardiovascular (11.1%), central nervous system (16.6%), musculoskeletal (7.6%), and multi-organ system malformations (3.4%) while 49.2% had no anomalies. A significant association was observed between fetal anomalies and the severity of polyhydramnios ($p < 0.001$). Fetal anomalies were more commonly noted with severe polyhydramnios.

Conclusion: A significant association was found between the severity of polyhydramnios and the presence of fetal anomalies with approximately half of the cases exhibited fetal malformations i.e., GIT, CVS and CNS abnormalities being the most common. The likelihood of fetal malformations increased notably with severity of polyhydramnios.

Keywords: Polyhydramnios, fetal anomalies, ultrasonography, Congenital malformations, congenital defects, pregnancy.

INTRODUCTION

For proper growth and development of the fetus, the presence of adequate amount of amniotic fluid is crucial but excess accumulation of this fluid, called polyhydramnios, is pathological and associated with congenital malformations. Polyhydramnios is characterized by an excessive build-up of amniotic fluid relative to gestational age, typically exceeding 2000 ml.

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In recent research, it is also defined as an amniotic fluid index (AFI) of more than 95th or 97th percentile for the gestational age, while some other studies define polyhydramnios as amniotic fluid having deepest vertical pool of ≥ 8 cm or amniotic fluid index [AFI] ≥ 24 as seen on obstetric ultrasound.(1) Polyhydramnios is classified based on amniotic fluid index (AFI) as mild (25–29.9 cm), moderate (30–34.9 cm), and severe (≥35 cm).(2) The prevalence of polyhydramnios has been reported to vary significantly across different populations and settings. Recent studies indicate that the incidence ranges from 1 to 2 % of all pregnancies worldwide.(3,4) The degree of polyhydramnios often correlates with the likelihood of finding an underlying cause. It is noted that usually mild polyhydramnios is often idiopathic or related to maternal diabetes. Moderate to severe cases are more frequently associated with fetal anomalies, especially gastrointestinal and neurological defects, as well as chromosomal abnormalities or infections.(4) Excessive accumulation of amniotic fluid can be due to both maternal and fetal conditions. Poorly controlled diabetes is

the most common maternal cause of polyhydramnios. Additionally, other maternal risk factors are alloimmunization and infection. Aneuploidy, structural abnormalities, hydrops and central nervous system anomalies are the common fetal causes of polyhydramnios while about 76.8% polyhydramnios remain idiopathic. The frequency of fetal congenital anomalies increases according to the severity of polyhydramnios.[5,6] Term pregnancies with polyhydramnios, especially moderate–severe ones are at a significantly increased rate for adverse pregnancy outcome. [7] Fetal congenital anomalies are the primary cause of severe polyhydramnios. Most frequent among them are the central nervous system anomalies, gastrointestinal tract defects, congenital airway malformations, musculoskeletal anomalies and congenital diaphragmatic hernia (CDH).[8,9] A study on amnioreduction for polyhydramnios showed the overall prevalence of polyhydramnios as 1.2% of all pregnancies out of which CNS anomalies were the most frequent, found in 28% of affected fetuses. Cardiovascular anomalies accounted for 22%, while gastrointestinal malformations were seen in 16%. Additionally, multiple congenital anomalies involving more than one system occurred in 13% of cases. These findings highlight the diverse fetal conditions associated with polyhydramnios.[10] Kornacki et al. reported that 53.3% of severe polyhydramnios cases were associated with fetal anomalies. Gastrointestinal anomalies (5.3%) and chromosomal anomalies (2.1%), particularly Trisomy 18, were the most frequent.[11]

Understanding the local burden and causes of polyhydramnios is crucial for timely diagnosis, management, and prevention of adverse outcomes. The aim of this study is to determine the local magnitude of polyhydramnios, its severity and association of polyhydramnios with fetal anomalies. It highlights the study's focus on understanding the prevalence, severity, and relationship of polyhydramnios with fetal anomalies in a specific local context.

MATERIAL AND METHODS

A descriptive cross-sectional study was conducted in the department of obstetrics and gynaecology, Lady Reading Hospital, Peshawar for the duration of six months (20th April 2022 to 20th oct 2022). Sample size of 380 women was calculated using online sample size calculator- Rao soft with 95% confidence interval and 5% margin of error. Non-probability consecutive sampling technique was used. Patients fulfilling the following criteria were included in the study:

Women diagnosed with polyhydramnios via obstetric ultrasound characterised by deep vertical pool of more than 8cm or AFI of more than 24cm were included in the study. Eligible participants were those who had parity of less than 5, pregnancy beyond 24 weeks of gestation and age between 15-45 years. Pregnant women who had placenta previa or placental abruption were excluded from the study as these placental abnormalities are found to be independently associated with fetal abnormalities as they could confound outcomes of polyhydramnios. Additionally, women with multiple gestation and women with known diabetes mellitus or gestational diabetes were not included as diabetes is also a well-established cause of polyhydramnios.

After approval from the ethical research board of Lady Reading Hospital, Peshawar, women presenting with polyhydramnios who met the inclusion criteria were enrolled in the study after obtaining informed consent. After medical history, physical examination and baseline investigations, the participants underwent a detailed anomaly scan to identify fetal anomalies associated with polyhydramnios. Information such as age, BMI, gravidity, parity, AFI, fetal malformations and period of gestation was recorded on a pre-designed proforma. To minimize confounding variables and bias in the study results, strict adherence to exclusion criteria was maintained. Data was entered and analyzed in IBM SPSS version 20. Mean and SD were calculated for quantitative variables like age, period of gestation and BMI. Frequencies and percentages were calculated for categorical variables i.e., educational status, gravidity, parity, amniotic fluid index and fetal malformations. Fetal malformations were classified as cardiovascular system (CVS), gastrointestinal (GI), central nervous system (CNS), musculoskeletal (MSK), or multi-organ system (MOS) anomalies. The association between the severity of polyhydramnios and the presence of fetal anomalies was evaluated using Chi-square test. A p-value <0.001 was considered statistically significant.

RESULTS

This study included 380 pregnant women diagnosed with polyhydramnios on ultrasound. The mean age of participants was 28.8 years

(SD ± 5.70), and the mean gestational age was 34.3 weeks (SD ± 4.89). Detailed distributions of educational status, obstetric history, polyhydramnios severity, and types of fetal malformations are presented in Table 1. Polyhydramnios severity was classified as mild in 19.7%, moderate in 47.6%, and severe in 32.6% of the cases. Fetal malformations were detected in over half the cases (50.8%). A statistically significant association ($p < 0.001$) was observed between the severity of polyhydramnios and the presence and type of fetal malformations. Mild cases had the highest proportion of fetuses without anomalies, whereas moderate and severe cases showed a higher frequency of malformations, particularly involving the CNS. The association between anomaly types and amniotic fluid index categories is shown in Table 2.

Table 1: Demographic, obstetric and clinical characteristics of pregnant women with polyhydramnios (n=380)

V		F	P
Education status	Uneducated	73	19.2
	Completed some primary education	157	41.3
	Completed some secondary education	114	30.0
	Highly educated	36	9.5
Gravidity	Primigravida	160	42.1
	Multigravida	220	57.9
Parity	Para 0	160	42.1
	Primiparous	97	25.5
	Multiparous	123	32.4
AFI on ultrasound	Mild polyhydramnios	75	19.7
	Moderate polyhydramnios	181	47.6
	Severe polyhydramnios	124	32.6
Fetal malformations	None	187	49.2
	GI malformations	46	12.1
	CVS malformations	42	11.1
	CNS malformations	63	16.6
	MSK malformations	29	7.6
	MOS malformations	13	3.4

V	M	SD	R	Min	Max
Age of participants	28.8	5.70	27.0	18.0	45.0
Period of gestation	34.3	4.89	38.0	30.0	41.0
Body mass index	26.8	2.73	13.1	18.9	32.0

V; variables: F; frequency: P; percentages: AFI; amniotic fluid index: GI; gastrointestinal: CVS; cardiovascular: CNS: central nervous system; MSK: musculoskeletal; MOS: multi-organ system malformations.

Table 2: Descriptive statistics for scale variables:

V	M	SD	R	Min	Max
Age of participants	28.8	5.70	27.0	18.0	45.0
Period of gestation	34.3	4.89	38.0	30.0	41.0
Body mass index	26.8	2.73	13.1	18.9	32.0

V; variables: M; mean: SD; standard deviation: R; range: Min; minimum: Max; maximum.

Table 3: Association of fetal anomalies with polyhydramnios (n=380)

fetal malformations	AFI on ultrasound			p-value
	Mild polyhydramnios	Moderate polyhydramnios	Severe polyhydramnios	
None	67 (89.3%)	104 (57.5%)	16 (12.9%)	**0.000**
GI	3 (4.0%)	15 (3.8%)	28 (22.6%)	
CVS	0 (0.0%)	17 (9.4%)	25 (20.2%)	
CNS	5 (6.7%)	28 (15.5%)	30 (24.2%)	
MSK	0 (0.0%)	14 (7.7%)	15 (12.1%)	
MOS	0 (0.0%)	3 (1.7%)	10 (8.1%)	

GI: gastrointestinal; CVS: cardiovascular; CNS: central nervous system; MSK: musculoskeletal; MOS: multi-organ system malformations.
Chi ^{2} test. P-value <0.001 taken as significant.

DISCUSSION

Our study included 380 pregnant women diagnosed with polyhydramnios and congenital anomalies were identified in 50.8% of the cases. A statistically significant association was found between anomaly frequency and the severity of polyhydramnios. The most commonly observed anomalies were central nervous system (16.6%), gastrointestinal (12.1%), cardiovascular (11.1%), and musculoskeletal (7.6%) malformations. Additionally multi-organ system (3.4%) anomalies were exclusively found in cases of moderate and severe polyhydramnios. Overall, the incidence of congenital anomalies increased proportionally, particularly in moderate to severe polyhydramnios.

These findings were consistent with the findings of a study by Dr. Girdhar Gopal Nagar et al. who reported a congenital anomaly rate of 16.84% among 196 pregnancies with polyhydramnios, with the highest frequency observed in severe cases (51.5%), followed by moderate cases (45.5%). The most common anomalies included anencephaly, hydrops fetalis, and gastrointestinal and skeletal defects, showing similar results to our study. The higher anomaly rate of 50.8% as evident in our study could be attributed to higher prevalence of moderate (47.6%) to severe (32.6%) polyhydramnios in our cohort as well as systemic challenges in prenatal anomaly detection including socioeconomic barriers and infrastructural limitations. Furthermore, many women in our cohort presented in the late third trimester (34.3 weeks), limiting opportunities for early detection and fetal intervention. This pattern aligns with findings by Nagar et al. who similarly reported that most of their cases presented at 37–40 weeks, with severe polyhydramnios being more likely to harbor congenital anomalies, particularly CNS and GI defects.[1] Similarly, the findings by Laoreti et al. also highlight a strong association between the severity of polyhydramnios and the presence of fetal anomalies, particularly neurological and multi-organ system defects. Their study reported that the majority of anomalies were identified in cases of severe polyhydramnios, aligning with our observation of increased anomaly rates with rising AFI levels. This similarity reinforces the clinical implication that the degree of polyhydramnios can serve as a predictor for underlying structural abnormalities.[10] In our study, CNS anomalies dominated (24.2% of severe

polyhydramnios cases) mirroring trends in prior researches. Pillai et al. reported an even higher anomaly rate of 37.7% in polyhydramnios cases, with CNS defects remaining the most frequent category. They also observed high perinatal mortality (33.96%), largely due to congenital malformations. Like our study, late-term presentation (limiting diagnostic and intervention window) and low socioeconomic status (delaying access to advanced prenatal care) contributed to poor outcomes. These overlapping findings underscores the critical the need for early diagnosis and comprehensive fetal evaluation in polyhydramnios cases.[12] Our results regarding CNS anomaly predominance also aligns with Wassan et al although notable quantitative difference. Wassan et al. found that 24.05% of women with polyhydramnios had congenitally malformed babies, with CNS anomalies—particularly hydrocephalus (10.13%) and anencephaly (7.59%)—being most common. In contrast, our study observed a higher anomaly rate of 50.8%, with a similar pattern of CNS predominance. Wassan et al. also reported high rates of preterm births (63.3%) and low Apgar scores (16.5%), outcomes that were consistent with the adverse perinatal risks identified in our findings, though our anomaly rate was notably higher.[13] Similar trends were observed in the Omani study by Tashfeen and Hamdi, which reported an 8.2% anomaly rate among 477 cases, with CNS, GI, and CVS anomalies most prevalent., though the overall rate was lower compared to the anomaly rate of our results (50.8%). Similar to our study where we observed a statistically significant association ($p < 0.001$) between the severity of polyhydramnios and the presence of fetal malformations, Tashfeen and Hamdi also observed a greater burden of anomalies in more severe polyhydramnios and highlighted diabetes and advanced maternal age as significant associated factors.[14] The 2021 UK-based study by Kyriacou et al. found fetal anomalies in 30% of polyhydramnios cases. Interestingly, they reported no significant difference in raw AFI between normal and abnormal cases, challenging the diagnostic reliability of AFI alone. Instead, AFI normalized to fetal weight (AFI/kg) showed stronger association with pathology, especially when polyhydramnios was detected earlier in gestation (20–24 weeks), where the anomaly rate reached 83%. Although, our study did not use AFI/kg, but we did observe a strong correlation between absolute AFI and anomaly frequency, especially in third-trimester diagnoses where more severe cases were detected.[15] Kornacki et al. found a lower overall anomaly rate (21.3%) compared to our study (50.8%) and identified gastrointestinal anomalies as the most common. In contrast, our findings showed central nervous system anomalies predominating in moderate and severe polyhydramnios. Both studies, however, confirmed a strong correlation between anomaly frequency and increasing AFI severity.[11]

Collectively, the evidence from multiple studies underscores that polyhydramnios—especially of moderate to severe degree—serves as a critical indicator of potential congenital anomalies. Despite variations in study methodologies, diagnostic tools, and populations, there is strong consensus on the necessity of early and detailed fetal evaluation. This ideally includes serial ultrasounds, echocardiography, and, where available, amniocentesis or genetic testing. In resource-limited settings, however, the amniotic fluid index (AFI) remains a practical and accessible screening tool. An elevated AFI should trigger a comprehensive fetal assessment and vigilant perinatal monitoring to optimize the outcomes

CONCLUSION

Fetal congenital malformations were associated with polyhydramnios and were common in cases of pregnancy affected by severe polyhydramnios. Our study showed that the fetal malformations like central nervous system, Gastrointestinal, cardiovascular, musculoskeletal and muti-organ system malformations were common among infants born to mothers with severe polyhydramnios.

DECLARATIONS

Authors contributions: Mehreen Zia: Contributed to the conception and design of the study, supervised the research process, critically revised the manuscript for important intellectual content, and approved the final version for publication. She accepts full responsibility for the integrity of the work. Hina Aslam: Assisted in data collection, contributed to data analysis and interpretation, helped draft the manuscript, and approved the final version of the article. Hadiqa: Involved in literature review, data entry, and interpretation; contributed to drafting and revising the manuscript; and approved the final version. Aiman Zia: Assisted in study design,

coordinated data acquisition, contributed to manuscript editing, and approved the final version of the manuscript. All authors agree to be accountable for all aspects of the work, ensuring that any issues related to the accuracy or integrity of any part of the work are appropriately addressed.

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