

ULTRASOUND DETECTION OF MATERNAL RISK FACTOR AND CONGENITAL ANOMALY IN TERTIARY CARE HOSPITALS LAHORE

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Abstract

Background: Congenital anomalies constitute a serious contributor to fetal and neonatal morbidity and mortality, especially in low- and middle- income settings. Among the possible contributors are maternal risk factors, for example maternal age, pre- Pregnancy BMI, maternal diabetes, maternal intake of folic acid, other clinical comorbidities. Prenatal ultrasound scan represents a useful noninvasive tool in screening and early diagnosis of congenital structural anomalies, however the local evidence for maternal risk factors as predictors for antenatal detection and performance of ultrasound scan are few.

Objective: To evaluate the association of maternal risk factors on the occurrence of congenital anomalies and to assess the accuracy of ultrasonography in detecting these anomalies in a tertiary care hospital in Lahore.

Methods: Descriptive Cross-Sectional Study conducted on 196 singleton pregnancies, women aged from 18-45 years, at Fuji Foundation & Ghurki Hospitals Lahore. Data collected using questionnaires & clinical records. Pearson's chi square tests were used to estimate Associations while Ultrasound Diagnostic performance using Sensitivity, Specificity, Predictive values & accuracy.

Results: This research involved 196 expectant mothers, out of which 32 (16.3%) of the unborn children had congenital anomalies, of which central nervous system deformities were most frequent (34.4%). Other deformities found included heart (15.6%), renal (12.5%), abdominal (12.5%), and musculoskeletal (6.3%) anomalies. Important risk factors concerning the deformities included maternal age, high body mass index (BMI), marriage within the same family, previous deformities in the patient's family, lack of folic acid intake, specific gestational age interval, and abnormal amniotic fluid (p -value < 0.001). Maternal diseases related to the defects included diabetes, infections, kidney diseases, and obesity. The results of the scans appeared to be very accurate: 97% in regards to finding the defect, 100% in regards to confirming there is no defect and in general the scans had 99.5% accuracy.

Conclusion: The relationship between the health concerns of mothers and the incidence of congenital defects in newborns was studied where congenital defects in the brain or spine were noted to occur in 16.3% of babies. The researchers recommend the use of prenatal ultrasound and self-care by the

Introduction

Congenital anomalies refer to abnormalities that are either functional or structural that occur in any of the stages of intrauterine development, and can be identified during any stage of gestation. These abnormalities cause significant problems in terms of perinatal mortality, morbidity, and disability in countries that are not well off in terms of economic development. While research shows significant differences in the incidences reported, the reason behind discrepancies is believed to be variations in the availability of case-ascertainment tools and techniques.^{1,2}

Ultrasonography before labor is considered the most important imaging technique used for assessing any abnormalities in the fetus. However, the efficacy of this technique differs depending on the type of anomaly, the period of pregnancy, ultrasound procedures, and the level of proficiency of the person conducting the ultrasound.³ According to the findings of a systematic review and a meta-analysis that was carried out in 2024, despite having a high specificity of detection of various congenital anomalies, it was noted that the sensitivity of this procedure is variable for different types of anomalies.⁴

Maternal diabetes, obesity, hypertension, poor folate consumption, disease, smoking, and blood relations were investigated as potential causes of congenital defects.⁵⁻⁸ Recent studies affirm the significance of periconceptional folic acid consumption while the epidemiological studies show correlations of maternal and obstetric characteristics with congenital defects.⁹⁻¹¹ In Pakistan, recent studies outlined the congenital defect patterns and the maternal risk factors, but local research linking maternal factors with ultrasound results and diagnostic effectiveness is scarce.¹²

Therefore, this research aimed to explore the association between some maternal and pregnancy-associated factors and congenital defects identified prenatally, as well as to determine the diagnostic correctness of prenatal ultrasound in pregnant women seeking medical assistance in clinics in Lahore.

Methodology

Study Design

This study utilized a descriptive cross-sectional study design.

Clinical Settings

The research was carried out at Radiology & obs and gynea department of Fuji Foundation ,Gurki Hospitals, Lahore

Sample Size:

The study included a total of 196 pregnant women.

Sample Size Calculation.

$Z = 1.96$ (95% confidence Level), p = Expected sensitivity/specificity, E = Margin of error (5% or 0.05)

$p = 0.85$ (85% expected sensitivity or specificity)

Formula:

$$N = \frac{Z^2 \cdot p(1-p)}{E^2}$$

$p = 0.85$ (85% expected sensitivity or specificity)

$$N = \frac{1.96^2 \cdot 0.85 \cdot (0.15)}{0.05^2}$$

Duration of Study:

4 months

Sampling Technique:

A purposive sampling technique was used in this study. Participants who meet the inclusion criteria and are relevant to the objectives of the research are selected. This technique was considered appropriate because it allows the researcher to collect data specifically from pregnant women undergoing ultrasound examination for fetal assessment.

Selection Criteria

The sample selection was depend upon inclusion and exclusion criteria

Inclusion Criteria:

- Women aged 18-45 years.
- Singleton pregnancies with at least one ultrasound scan during pregnancy.
- Willing to provide informed consent.

Exclusion Criteria:

- Women who were not pregnant or not willing to participate

Ethical Consideration

This study followed the ethical principles for research at Superior University. Informed written consent was obtained from all patients and information remained confidential and anonymous with the knowledge that the patient could withdraw from the study at any time. No hazards were related to the study with a possible advantage of discovering birth abnormalities early by means of the non-invasive procedure of sonography. Confidentiality was respected throughout the course of the study and publication.

DATA COLLECTION PROCEDURE

Questionnaire data were collected which included demographic variables, risk factors for the mother, BMI, consumption of folic acid and medical history. Relevant clinical records and past ultrasonic studies were consulted. All participants that met criteria for the study had standardized obstetric ultrasonography for the investigation of congenital anomalies and structural abnormalities. Data are classified and recorded into an ultrasonic data sheet system.

DATA ANALYSIS PROCEDURE

Statistical analysis The entered data was analyzed by SPSS version 30. Descriptive analysis of the data presented by frequencies and percentages. Maternal risks factors were assessed for association with congenital anomalies using Pearson chi-square test and $p < 0.05$ was considered significant. For the

usefulness of prenatal sonography sensitivity, specificity, positive predictive value and negative predictive value were measured.

Conflict of Interest

The authors declare no conflict of interest.

Financial Disclosure

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Authors' Contributions

Zoya Nazir Khan: Conceptualization, data collection, data analysis, and manuscript writing. M. Adnan Hafeez: Study supervision, methodology development, critical revision of the manuscript, and final approval of the submitted version.

Results

Results Nineteen ninety six pregnant ladies were performed prenatal ultrasonographic examinations. Congenital anomaly detection occurred in 32(16.3%)examinations whereas there was no detectable anomaly detection in here maiming ones. Maternal characteristics, gestational age, gestational-body mass index, consanguinity, folic acid take , vitality of the fetus , type of ultrasonic test , skill of the personnel, fluid amniotic status and pertinent family history, obstetric history are demonstrated in table I.

Table I. Baseline characteristics and distribution of ultrasound-detected congenital anomalies (n=196)

Variable/category	Total, n (%)	Detected CA, n (%)
18-20	26 (13.3)	3 (11.5)
20-25	76 (38.8)	10 (13.2)
25-30	56 (28.6)	6 (10.7)
30-35	38 (19.4)	13 (34.2)
18-22 weeks	90 (45.9)	19 (21.1)
22-26 weeks	58 (29.6)	11 (19.0)
26-30 weeks	33 (16.8)	1 (3.0)
30-34 weeks	15 (7.7)	1 (6.7)

Normal	92 (46.9)	13 (14.1)
Underweight	21 (10.7)	0 (0.0)
Overweight	59 (30.1)	11 (18.6)
Obese	24 (12.2)	8 (33.3)
Consanguineous marriage: Yes	84 (42.9)	18 (21.4)
Consanguineous marriage: No	112 (57.1)	14 (12.5)
Folic acid intake: Yes	135 (68.9)	16 (11.9)
Folic acid intake: No	61 (31.1)	16 (26.2)
Fetal viability: Alive	183 (93.4)	25 (13.7)
Fetal viability: Not alive	13 (6.6)	7 (53.8)
Type of scan: Anomaly	135 (68.9)	21 (15.6)
Type of scan: First trimester	6 (3.1)	5 (83.3)
Type of scan: Targeted	55 (28.1)	6 (10.9)
Operator: Consultant	97 (49.5)	19 (19.6)
Operator: Sonographer	99 (50.5)	13 (13.1)
Amniotic fluid: Normal	154 (78.6)	15 (9.7)
Amniotic fluid: Oligohydramnios	16 (8.2)	2 (12.5)
Amniotic fluid: Polyhydramnios	25 (12.8)	15 (60.0)
Previous child with congenital anomaly: Yes	14 (7.1)	5 (35.7)
Previous child with congenital anomaly: No	182 (92.9)	27 (14.8)
Family history of congenital anomaly: Yes	27 (13.8)	7 (25.9)
Family history of congenital anomaly: No	169 (86.2)	25 (14.8)

Mothers in the oldest aged category(>35 year old) had a higher proportion of ultrasonic determined congenital anomaly; also obese women.

The probability of anomaly detection was significantly higher in mothers whom took oral folic acid and previous anomaly history. The gestational age, congenital familial disease, Abortion, the history of parity, having pregnancy the history of son is also a cause of the existence of malformation but didn't make

any significant statistical difference in relation to other factors. Anomaly detection was significantly associated with amniotic fluid status, viability and ultrasonic test, polyhydramnios and non-viable fetus were more associated with malformed fetus. Ultrasound procedure correlated significantly with abnormality in ultrasound detected. Operator level is not correlated significantly with anomaly detected (table II).

Table II. Associations between categorical variables and ultrasound-detected congenital anomalies

Variable	Pearson χ^2	df	p-value
Maternal age	11.18	3	0.011
Gestational age	7.10	3	0.069
BMI	9.74	3	0.021
Consanguineous marriage	2.80	1	0.094
Folic acid intake	6.36	1	0.012
Previous child with	4.15	1	0.042

congenital anomaly			
Family history of congenital anomaly	2.11	1	0.146
Amniotic-fluid status	39.80	2	<0.001
Parity	3.71	3	0.295
Abortion history	1.95	3	0.583
Gravidity	0.89	3	0.827
Fetal viability*	14.35	1	<0.001
Type of ultrasound scan	20.96	2	<0.001
Operator level	1.49	1	0.221
Maternal medical risk-factor category†	40.02	10	<0.001

Most frequent maternal medical risk factor was absent while renal diseases was less frequent. Anemia had the higher percentage among listed risk factors. Maternal medical risk factor

overall was significantly correlated with ultrasonic detected fetal anomaly (table III)

Table III. Maternal medical risk factors among participants and anomaly-positive examinations

Risk factor	Total n	Total %	Detected CA n	Detected CA %
None	105	53.6	9	28.1
Anemia	33	16.8	3	9.4
Gestational diabetes mellitus	19	9.7	4	12.5
Pregnancy-induced hypertension/pre-eclampsia	12	6.1	3	9.4
Chronic hypertension	7	3.6	2	6.3
Pregestational diabetes mellitus	5	2.6	4	12.5
Infection during pregnancy	5	2.6	3	9.4
Thyroid disease	4	2.0	1	3.1
Cardiac disease	3	1.5	1	3.1
Smoking/tobacco exposure	2	1.0	1	3.1
Renal disease	1	0.5	1	3.1
Total	196	100.0	32	100.0

Among the congenital anomaly, centralnervous systemmalformations had high prevalence(50%). Mal formations affecting cardiac, abdominal and renal organs are similar

in frequency(8,1%)and among them; musculoskeletal system anomalies had lowest prevalence(3.1%)(Table IV).

Table IV. Distribution of ultrasound-detected congenital anomalies by organ system (n=32)

Organ system	n	%
Central nervous system	11	34.4

Cardiac	5	15.6
Renal	4	12.5
Abdominal	4	12.5
Craniofacial	3	9.4
Musculoskeletal	1	3.1
Unclassified/unspecified	4	12.5
Total	32	100.0

Comparing the prenatal result with reference value the tested ultrasounds had an excellent diagnostic capability due to false one, false zero

and no error as showed in Table V sensitivity , specificity and predicted values in table V.

Table V. Diagnostic performance of prenatal ultrasonography against the recorded reference status

Measure	Value
True positive	32
False negative	1
True negative	163
False positive	0
Sensitivity	97.0% (95% CI 84.2-99.9)
Specificity	100.0% (95% CI 97.8-100)
Positive predictive value	100.0% (95% CI 89.1-100)
Negative predictive value	99.4% (95% CI 96.6-100)
Overall diagnostic accuracy	99.5% (95% CI 97.2-100)

Discussion

In this study, we reviewed prenatal diagnosis (by Ultrasound) and maternal/pregnancy related factors for congenital abnormalities detected in two tertiary-care hospitals in Lahore. The ultrasound-detected anomaly rate was 16.3%; this cannot be directly extrapolated to a general population as purposive sampling and tertiary referral might represent selection of high-risk pregnancies. In comparison to other recent studies of pregnancy-related factors and congenital abnormality detection, frequencies were relatively high. These reports included data from other tertiary centers where abnormality detection rate has varied from 4.1% during routine scans [3] to 5.49% from a tertiary care center^[4].

We found CNS related anomalies as the most frequent group as expected given the literature from Pakistan and worldwide. Brain abnormalities constitute a significant part of detected abnormalities. A study similar to ours done by Nahra et al., in Lahore, has illustrated importance of Ultrasound in detectability of CNS anomalies in second and third trimester. Out of the 33 detected anomalies none was

recorded under the null status in relation to classification status therefore distribution illustrated above belongs to the cases of detected abnormalities belonging to different classification groups.

Statistically significant relation between detection of anomalies and maternal BMI observed in the study, with relatively higher proportion of detected anomalies among obese and morbidly obese category compared to normal BMI categories. Similar observation has been reported in population-based studies looking at the association between maternal metabolic characteristics and congenital anomalies^[15-17]. Since BMI had only four categories there can be overlap within, and the lack of adjustment for confounders further makes it impossible to claim causality.

Statistical significance was observed between folic acid ingestion and abnormality detection with relatively lower proportion of detected abnormalities among women using folic acid for periconceptional supplementation. This finding is in agreement with a systemic review which demonstrated the use of folic acid during periconception reduces congenital

abnormalities risk^[12]. Although in this study the intake was subjective, dose not defined, adherence not established and the actual biochemical levels for folate are not recorded.

Strong association was found between detection of anomalies and liquor amount status and this is largely attributable to the role of polyhydramnios which is associated with fetal structural deformities, but itself triggers the examination due to secondary implication leading to subsequent abnormality detection. Previous child-related issues were significant and cases involving pregnancy of woman with existing history were compared. Cases relating to consanguinity and family history were not found to be statistically significant, however this remained so only after reanalysis of the data. Associations between Maternal age, Smoking, Consanguinity have been reported in other Pakistani based studies of childhood mortality related to congenital abnormalities^[9]. Shahmirzady et al. Reported on maternal variables identified to contribute risk to newborns presented with congenital anomalies^[18]. CNS related anomalies were also important among cases presented by Mahdi et al.^[6] of abnormality detection. A study conducted in Pakistani population regarding diagnosis of fetal abdominal abnormalities revealed discrepancy in spectrum of organ systems reported with considerable variety^[10]. The issue of age-related anomaly diagnosis appears problematic: whereas, Goetzinger et al. Indicate a decreased risk of significant congenital malformations during normal pregnancies for older maternal age^[21], Almeida et al. Have cited association of pregnancy problems and congenital anomalies.

No statistically significant finding were made related to GA category after analysis adjusted to the valid 42table since the examinations in largest group were done at 18-22 weeks and this may indeed represent the examination protocol rather than be dependent on maternal/pregnancy related variables or abnormality presence. There exist significant variations on the sensitivity with reference to the specific abnormality as well as to the stage of pregnancy, standardized protocols are likely to increase ultrasound performance^[8,10,11]. Hence current finding supports the hypothesis

for the presence of standard protocol however, it is not indicative of any optimum GA.

The obtained values were 97.0 for sensitivity and 100.0 for specificity. However these values cannot be assumed to be a general performance evaluation of Ultrasound. These values vary depending on the sensitivity within the spectrum of anomalies, gestation, skill of operator, ultrasound equipment and confirmation data. Wide range of confidence interval of 97.0 may also be interpreted due to smaller numbers of reference positive cases. There was no documented uniform protocol for genetic or post-natal confirmation in all pregnancy therefore, the data of ultrasound findings cannot be relied upon purely due to bias in test detection without confirmation^[5,7]. A distinction between 32 found ultrasound and 33 referable anomalies was maintained through out data since this has made them comparable entities rather than to allow contamination of tested positive anomalies with referable positives and this may be one of the main reason for high sensitivity.

Operator experience was also evaluated but no statistical significance in anomaly detection rates was recorded after data re-analysis. The disparity of association of abnormality with operators can exist either because of variability of case mixes where only more risky cases are assigned to specialist or it can influence the sensitivity from the other side that only well trained specialist are given more risky assignments.

The present study was cross-sectional and was conducted on purposive sampling in two tertiary care hospitals at Lahore, Pakistan, so results cannot be generalized and causality cannot be inferred. Most of the variables analyzed had sparse data in some rows which does not provide reasonable data for an asymptotic chi-square- approximation. There can be recall bias in Maternal/Pregnancy related variables that were verbally communicated by participants in this case the mother. None of all suspected pregnancies underwent genetic analysis or post-natal confirmation in standard protocol there was distinction between 32 abnormalities seen on US scan and 33 abnormality detected by referable anomalies and comparison was made

between them to confirm sensitivity and specificity.

Conclusion

In the tertiary-care setting tertiary-care sample at Lahore the frequency of diagnosis of congenital anomaly by prenatal ultrasonography was 16.3% in observed pregnancies. At conventionally-derived thresholds the following parameters were positively associated with the risk of congenital anomaly; BMI category, use of folic acid and amniotic fluid at delivery and history of a previously born child with a congenital anomaly as well as the nature of scan and fetal viability, whereas others gestational-age category, consanguinity, gravidity, parity, history of abortions, family history and operators level did not show a significant associations after recalculation from viable contingency table. Recorded sensitivity (97.0%), specificity (100.0%), predictive accuracy (100.0%), and global efficiency (99.5%) of the Ultrasound showed high effectiveness of these investigations which lends support to routine assessment of all pregnant women with structured examination techniques while stressing the importance of large multicenter studies with independent external postnatal as well as genetic referents to compare the data.

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