RESEARCH ARTICLE

Pregnant women's knowledge about birth defects and their associated risk factors in Quetta

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Abstract

Objective: To quantify pregnant women's knowledge of congenital abnormalities, their associated risk factors and preventative measures, and to determine the association of knowledge level with socio-demographic factors.

Method: The multi-centre, cross-sectional study was conducted from October 2023 to March 2024 in Quetta, Pakistan, after approval from the ethics review committee of the Quetta Institute of Medical Sciences, and comprised pregnant women aged at least 18 years who were attending antenatal clinics. Data was collected using a questionnaire. The participants' knowledge was assessed through 21 close-ended questions. The association of knowledge score with a range of socio-demographic factors was explored. Data was analysed using SPSS 26.

Results: Of the 640 mothers, 303(47.3%) were aged 25-34 years, 131(20.5%) had completed secondary school education, 540(84.4%) were housewives, 373(58.3%) were married within their families, and 447(69.8%) lived in urban areas. There were 349(54.5%) participants having moderate knowledge about birth defects, 207(32.3%) had high level of knowledge, and 84(13.1%) had poor knowledge. The knowledge level had significant association with education level (p=0.001) and residence area (p=0.006).

Conclusion: Pre-pregnancy counselling shall be made available at antenatal care centres. Maternal obesity and consanguinity shall be pointed out to pregnant women as risk factors for congenital anomalies.

Keywords: Congenital abnormalities, Pregnancy, Health knowledge, Attitudes, Practice, Risk factors, Antenatal care, Regression analysis. (JPMA 76: 551; 2026) **DOI: https://doi.org/10.47391/JPMA.23158**

Introduction

Congenital disorders are certain medical conditions that afflict the foetus during intrauterine life and are, therefore, present at birth either as grossly visible structural anomalies or dysfunctions of various organ systems of the body. Early neonatal screening can significantly reduce morbidity and mortality through early detection.[1]

About 2.4 million newborns succumb to death within 28 days of their birth every year due to birth defects (BDs), making it necessary to ascertain their risk factors, formulate better preventative healthcare strategies, and improve curative methods.[2]

Pre-conception counselling and antenatal care (ANC) promote healthy nutrition, folate intake, and avoidance of tobacco/environmental hazards. The 2016 ANC model of the World Health Organisation (WHO) recommends eight or more visits covering maternal and foetal assessment, nutrition education, and routine symptom management.[3]

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In Pakistan's least-populated province, Balochistan, neonatal mortality is 63 per 1,000 live births, and maternal mortality is 298 per 100,000.[4] With 85% rural dwellers, but only 96 rural centres and 60 maternal and child health (MCH) centres, healthcare access is poor, with a doctor-to-patient ratio of 1:1000 and nurse-to-patient ratio of 1:50.[5,6] Sociocultural barriers and restricted mobility further delay access to care. Educating pregnant women on risk factors and prevention can enable primary prevention of congenital malformations.

The current study was planned to quantify pregnant women's knowledge of BDs, their associated risk factors, and preventative measures, and to assess the association of knowledge levels with sociodemographic variables.

Subjects and Methods

The multi-centre, cross-sectional study was conducted from October 2023 to March 2024 in Quetta, Pakistan. Quetta is one of the 36 districts in the province of Balochistan, having a population of over 2.5 million.[7] There are approximately 66 public healthcare facilities in Quetta; 6 civil hospitals, 4 military hospitals, 2 rural health centres (RHCs), 32 basic health units (BHUs), 12 civil dispensaries, and 10 MCH centres.[8] No accurate number is available of the private hospitals operating in Quetta.

After approval from the ethics review committee of the

Quetta Institute of Medical Sciences, Quetta, the sample size was calculated using the single population proportion formula with a 50% proportion (p=0.5), 95% confidence level (Zα/2=1.96), and 5% margin of error (d=0.05).[9] The formula used was:

$$n = \frac{\left(\frac{Z\alpha}{2}\right)^2 p\,(1-p)}{d^2}\ (n) = \frac{(1.96)^2 * 0.5\,(1-0.5)}{(0.05)2} = 385$$

A design effect of 1.5 was applied to account for potential variability in knowledge levels across five tertiary care hospitals, reflecting site-specific differences in patient demographics and healthcare access. Although not a formal cluster sampling design, this adjustment enhanced the robustness of the estimate. After accounting for a 10% non-response rate, the final sample size was adjusted to 636 and was rounded off to 640.[10,11]

The sample was raised using consecutive non-probability sampling technique from 3 public-sector hospitals — Civil Hospital Quetta, Bolan Medical Complex Hospital, and Combined Military Hospital (CMH) — and 2 private-sector hospitals — Heart and General Hospital, and City International Hospital.

Those included were pregnant women aged at least 18 years attending ANC clinics at the participating hospitals who could comprehend the locally dominant languages (Urdu, Pashto, Balochi or Persian) either personally or through a trained interpreter. Healthcare professionals were excluded to prevent knowledge bias, and so were women with severe communication barriers that could compromise informed participation. Also excluded were those who did not volunteer to participate.

After taking informed consent from all the subjects, data was collected using a self-designed questionnaire that was adapted from literature.[12] Some questions were added in the light of certain studies,[11,13] while some others were removed to make the questionnaire relevant and culturally appropriate. The questionnaire was subjected to scrutiny by field experts to remove any ambiguities and ensure content validity of the tool. The questionnaire was developed in English language, and was also translated into Urdu language. Finally, the questionnaire was pilot-tested on a sample of 30 pregnant women to ensure the reliability of the tool. Cronbach's alpha (α) value obtained for the knowledge assessment part of the questionnaire was 0.764.

The questionnaire covered sociodemographic variables, including age, residence, ethnicity, maternal educational level, maternal occupation, husband's occupation, average monthly family income, and consanguinity. It also explored clinical variables the participants, including gravidity, parity, number of children, prior abortion, prior stillbirth, prior BD history, family history of BDs, number of ANC visits, ANC booking time, uptake of folic acid, and the reason for taking folic acid.

The dependent variable was knowledge of BDs, which was assessed by asking the participants 21 close-ended questions. Correct answers were scored 1 point, while 0 was given for each wrong or 'I don't know' answer. The maximum total score obtainable was 21; 8 points for specific knowledge, 10 points for knowledge of the risk factors, and 3 points for knowledge of preventative measures.

The scores on the knowledge domain were categorized into three levels based on approximate tertiles of the total possible score: 0-7=low, 8-14= moderate, and 15-21= high. This tertile-based categorisation was in line with Bello et al., who used a similar approach for their 12-point knowledge scale; 0-4=low,5-8+moderate, and 9-12=high.[12]

Data was transferred to Google Sheets and analysed using SPSS 26. Data was expressed as mean+/-standard deviation (SD), or as frequencies and percentages, as appropriate. Multiple linear regression was used with a Bonferroni-corrected threshold of p<0.0071 (0.05/7) for seven pre-specified predictors (age, education level, occupation, husband's occupation, residence area, ANC visits), excluding less relevant variables (ethnicity, family income, married) based on low correlation, to control Type I error. For the purpose of multiple linear regression analyses, the scores were treated as continuous variables (0-21). In the multiple linear regression analysis, assumptions were verified to ensure model validity. P<0.05 was considered significant. Multicollinearity was assessed using variance inflation factors (VIFs), all of which were <1.6, indicating no significant collinearity among the predictors. Normality of residuals was confirmed through visual inspection of a histogram and normal probability plot, which showed an approximately normal distribution (mean residual ≈0, SD ≈1). Homoscedasticity was evaluated via a scatterplot of standardised residuals against predicted values, revealing random dispersion without patterns. Linearity was supported by the absence of systematic trends in residual plots. No influential outliers were detected.

Results

Of the 640 mothers, 303(47.3%) were aged 25-34 years, 131(20.5%) had completed secondary school education, 540(84.4%) were housewives, 373(58.3%) were married within their families, and 447(69.8%) lived in urban areas (Table 1).

Overall, 425(66.4%) of the respondents had two to four

Table-1: Sociodemographic characteristics of the study participants (n=640).

Variables	n (%)
Age (years)	
≤ 25	242 (37.8)
25-34	303 (47.3)
> 34	95 (14.8)
Education Level	
No Formal Education	272 (42.5)
Matriculation	131 (20.5)
Intermediate	103 (16.1)
Graduate and above	134 (20.9)
Respondent's Occupation	
Housewife	540 (84.4)
Private employee	44 (6.9)
Government employee	39 (6.1)
Self-employed	17 (2.7)
Husband's Occupation	
Unemployed	72 (11.3)
Private employee	172 (26.9)
Government employee	171 (26.7)
Self-employed	225 (35.2)
Ethnicity	
Pathan	298 (46.6)
Baloch	165 (25.8)
Hazara	41 (6.4)
Others	136 (21.3)
Average Monthly Income (Pakistani Rupees)	
< 25,000	246 (38.4)
25,000 to 50,000	196 (30.6)
> 50,000	198 (30.9)
Married	
Consanguineous Marriage	373 (58.3)
Non-Consanguineous Marriage	267 (41.7)
Residence Area	
Rural	193 (30.2)
Urban	447 (69.8)

Table-2: Clinical characteristics of the study participants.

Variables	n (%)
Gravidity	
1	118 (18.4)
2-4	425 (66.4)
≥5	97 (15.2)
Parity (n=474)	
1	106 (16.6)
2-4	296 (46.3)
≥5	72 (11.3)
Still Birth (n=474)	
Yes	129 (27.2)
No	345 (72.8)
Defected child delivered (n=474)	
Yes	100 (21.1)
No	374 (78.9)
History of birth defects in family	
Yes	218 (34.1)
No	422 (65.9)
Abortions (n=474)	
Yes	235 (49.6)
No	239 (50.4)
Antenatal care booking time	
< 20 weeks of gestation	449 (70.2)
> 20 weeks of gestation	191 (29.8)
Antenatal Care Visits	
<4	379 (59.2)
≥4	261 (40.8)
Took iron-folate (folic acid) during antenatal care (ANC)	
Yes	494 (77.2)
No	146 (22.8)
Reason to take iron-folate (folic acid) (n=494)	
To prevent anaemia	328 (66.4)
To support foetal growth	97 (19.6)
To prevent postpartum haemorrhage	14 (2.8)
To prevent congenital anomalies	55 (11.1)

Table-3: Cumulative knowledge scores of the study participants.

Variable	n (%)
Specific Knowledge of birth defects	
High	331 (51.7)
Low	309 (48.3)
Knowledge of risk factors of birth defects	
High	402 (62.8)
Low	238 (37.2)
Overall knowledge of birth defects	
High	207 (32.3)
Moderate	349 (54.5)
Low	84 (13.1)

Specific knowledge categorised as low (< 4 points) or high (≥ 4 points) based on the questions asked.

pregnancies, while 296(46.3%) had given birth two to four times. Of the 474(74%) respondents who had given birth, 129(27.2%) had still-births, 235(49.6%) had abortions, and 100(21.1%) had given birth to a child with BD, while 218(34.1%) of the total respondents had a history of children with BDs in their families.

More than two-third of the respondents 449(70.2%) had visited an ANC clinic in the first 20 weeks of gestation, and 379(59.2%) had less than four ANC visits during the course of their latest pregnancy. Overall, 494(77.2%) respondents said they had taken iron-folate during their latest pregnancy (Table 2).

There were 349(54.5%) participants having moderate knowledge about BDs, 207(32.3%) had high level of knowledge, and 84(13.1%) had poor knowledge (Table 3). The mean overall knowledge score was 12.4±4.2.

Of the total, 464(72.5%) participants knew that BDs can be treated or managed, 375(58.6%) were aware that BDs can be genetic in origin, 532(83.1%) were aware that taking un-prescribed medications before or during pregnancy can cause BDs, 293(45.8%) were able to identify maternal age ≥40 years as a risk factor for congenital anomalies, 274(42.8%) identified maternal obesity as a risk factor, and 485(75.8%) believed that regular check-ups during pregnancy can reduce the risk of BDs (Table 4).

The knowledge level had significant association with education level (p=0.001) and residence area (p=0.006). No significant association of knowledge level was found with age, occupation, husband's occupation, consanguineous marriage and number of ANC visits (Table 5).

Table-4: The participants specific knowledge, knowledge about risk factors, and knowledge about prevention of birth defects (BDs).

Variables	Yes n (%)	No n (%)	I don't Know n (%)
Participant's specific Knowledge of birth defects			
Birth defect is a disease acquired by pregnant women	201 (31.4)	230 (35.9)	209 (32.7)
It can be acquired by the developing foetus in the womb	356 (55.6)	93 (14.5)	191 (29.8)
It can be transmitted by contact with an affected child	99 (15.5)	372 (58.1)	169 (26.4)
Most birth defects are preventable	355 (55.5)	107 (16.7)	178 (27.8)
Most birth defects can be treated or managed	464 (72.5)	80 (12.5)	96 (15)
Most birth defects can be managed with physiotherapy	318 (49.7)	111 (17.3)	211 (33)
Most birth defects can be corrected with surgery	378 (59.1)	93 (14.5)	169 (26.4)
Most birth defects are genetic in origin	375 (58.6)	106 (16.6)	159 (24.8)
Participant's knowledge of risk factors of birth defects			
Using some un-prescribed medications can cause BDs	532 (83.1)	63 (9.8)	45 (7)
BDs occur due to smoking before or during pregnancy	468 (73.1)	104 (16.3)	68 (10.6)
BDs can occur due to advanced maternal age (≥40 years)	293 (45.8)	196 (30.6)	151 (23.6)
BDs can occur due to consuming smokeless tobacco	363 (56.7)	168 (26.3)	109 (17)
BDs can occur due to taking X-rays during pregnancy	365 (57)	120 (18.8)	155 (24.2)
Consanguineous marriage can cause BDs in children	430 (67.2)	146 (22.8)	64 (10)
Infection during pregnancy can cause BDs	372 (58.1)	123 (19.2)	145 (22.7)
Stress and depression during pregnancy can cause BDs	461 (72)	70 (10.9)	109 (17)
BDs can occur due to maternal obesity	274 (42.8%)	236 (36.9%)	130 (20.3)
BDs can occur due to chronic illness of the mother	472 (73.8%)	76 (11.9%)	92 (14.4%)
Participant's knowledge of preventative measures of birth defects			
Intake of iodized salt reduces the risk of BDs	183 (28.6%)	126 (19.7%)	331 (51.7)
Regular Check-ups during pregnancy helps prevent BDs	485 (75.8%)	78 (12.2%)	77 (12)
Intake of folic acid reduces the risk of BDs	398 (62.2)	53 (8.3)	189 (29.5)

Table-5: Multiple regression analysis exploring the effect of sociodemographic characteristics on the participants' overall knowledge of birth defects (BDs).

Variable	B	Std. Error	*t*-test	*p*-value
(Constant)	11.129	0.483	23.061	0
Age	0.198	0.24	0.825	0.410
Education Level	0.592	0.172	3.439	0.001
Occupation	0.32	0.25	1.282	0.200
Husband's Occupation	-0.291	0.166	-1.753	0.080
Marrying within family	0.222	0.338	0.658	0.511
Residence area (Urban/rural)	1.014	0.371	2.732	0.006
Number of antenatal care (ANC) visits	0.225	0.342	0.657	0.511

Discussion

In Pakistan, 3.2% deaths of children aged <5 years are caused due to congenital anomalies and the proportion is suspected to be higher in Balochistan.[14] To the best of our knowledge, the current study is the first conducted in Balochistan to assess the knowledge level of pregnant women regarding BDs. The study found a large majority of the participants to be moderately or highly knowledgeable about congenital anomalies, with 207(32.3%) of the women having high level of knowledge regarding BDs. This is comparable to a similar study conducted in northern Iran (32%).[15] These findings were, however, lower than those reported from Saudi Arabia (51%),[16] Sri Lanka (56.4%),[17] India (63.5%),[18] Egypt (76%) (16), Ethiopia (49.2%)[11] and Ghana (42.6%).[12] The variation can be explained to some extent by the difference in methods of data-collection, analysis and the participants' education level.

The current findings identified significant association for overall knowledge with education level (p=0.001) and area of residence (p=0.006). However, there was no significant relationship of knowledge level with age, occupation, husband's occupation, ethnicity, family income, consanguineous marriage and number of ANC visits (p>0.05). A study found significant association between the overall knowledge of the participants and their education level, but reported no significant relationship with the number ANC visits, which was in line with the current findings. However, the same study found a significant relationship of overall knowledge with age,[14] which was inconsistent with the current findings.

According to the 2023 census, 2.3%, 4.6% and 1.6% of the population of Quetta comprised Urdu-speaking, Punjabi-speaking and Sindhi-speaking people, respectively.[7] These settler communities and the Hazara population outperformed Pathans and Balochis in the current study, reflecting disparities. Besides, female literacy in Balochistan has been reported to be 21%, with 42.5% lacking formal education.[19] In the current study, the participants with graduate and higher level of education had the most knowledge about BDs.

There were 540(84.4%) current participants who were housewives and lacked an independent source of income. Balochistan is mainly a tribal region, and the current study mostly had ethnic Pathan (46.6%) and Baloch (25.8%) subjects whose cultural values dictate women to be housewives, and men to be the breadwinners of the family.

Further, only 17.4% of women in Balochistan attend ANC clinics more than four times during the course of a pregnancy.[20] In this study, 261(40.8%) women attended ANC clinics >4 times during the course of their latest pregnancy, which suggests that pregnant women in Balochistan are more attentive towards prenatal care. The current study found that women who attended ANC clinics >4 times had higher knowledge scores.

The risk of occurrence and recurrence of neural tube defects can be reduced by intake of supplements containing folic acid.[21] Only 28.6% of the current subjects knew iodine's role in preventing congenital anomalies. It is believed that the cost and availability of iodised salt limits its use. However, 62.2% of the subjects associated folic acid with risk reduction, reflecting obstetric emphasis. Only 355(55.5%) of the current women were aware that BD occurrence can be prevented to a large extent. This finding highlights the lack of awareness regarding the subject amongst the population studied.

Many of the current women 464(72.5%) were aware that

BDs can be treated or managed postnatally, which was in line with earlier findings.[11,15] Physiotherapy and surgery can help in the management and correction of BDs. Only 318(49.7%) and 378(59.1%) of the current participants were aware of the role of physiotherapy and surgery in the management and correction of BDs.

Moreover, 375(58.6%) of the current respondents were aware that BDs can be genetic in origin. This observation was lower compared to other questions due to limited knowledge of genetics and its terminologies among the general population.

The current findings revealed that 532(83.1%) and 468(73.1%) subjects were aware of the hazardous effects of taking over-the-counter medications and smoking before pregnancy or during pregnancy on the foetus, respectively. These findings were similar to those reported by Masoumeh, et al.[15] but were higher than those reported from Ethiopia.[11]

The risk of acquiring chromosomal disorders increases with the advancement of maternal age,[22] while maternal obesity is a risk factor for foetal and maternal complications of pregnancy.[23] The current findings revealed that only 293(45.8%) and 274(42.8%) participants were aware of these factors, respectively. These findings were slightly higher than those observed in Ethiopia (32.1% and 26.4%, respectively).[11] A study in Iran found a large majority of the participants (94.6%) to be aware of the pregnancy risks of advanced maternal age[14] but only 23.3% were aware of the effects of maternal obesity on the development of a child.[15]

Consanguineous marriage is identified as one of the leading causes of BDs in children.[24] A study conducted in the region found that the highest number of child mortality (61.9%) and abnormalities (32.1%) were observed among the children of first-cousin marriages.[25] Another study concluded that consanguinity is an established risk factor for developing intellectual disabilities in children.[26] In Pakistan, more than half of ever-married women aged 15-49 years are married to their first cousins. When second-cousin marriages are included, the number rises to 61.2% from 50.3%.[27] It has been established by multiple studies that such marriages cause congenital anomalies in the offspring.[20,21,28] In the current study, 373(58.3%) participants had married in their family, while 430(67.2%) correctly identified consanguineous marriages as a contributory risk factor for congenital anomalies. This finding was much lower than the finding reported by a study in Iran (82%).[15] The lack of formal education and awareness among women in Balochistan may explain the high incidence of consanguinity.

Lower maternal iodine status has been associated with lower birthweight and greater probability of the infant being small for gestational age.[29] Only 183(28.6%) of the current participants were aware that intake of iodised salt reduces the risk of BDs, while 331(51.7%) did not know anything about it, and 126(19.7%) reported negatively to the question. This finding was expected as majority of the population of Balochistan does not use salt that is fortified with iodine due to lack of awareness and the high price of iodised salt compared to the conventional salt sold in the region. Furthermore, 398(62.2%) of the current participants knew that taking folic acid before or during pregnancy reduces the risk of BDs. This finding was higher compared to the awareness about iodised salt because of the increased emphasis on the intake of folic acid by obstetricians and gynaecologists.

Regular prenatal check-ups were recognised as preventative by 75.8% of the current subjects. Regarding causes, 41.2% subjects cited scientific origins, while 30.2% attributed defects to God's will, highlighting the influence of religious beliefs.

The current study has limitations owing to its cross-sectional design which did not allow the assessment of temporal relations. Furthermore, data was collected at the time of ANC check-ups, which may have introduced a social desirability bias among the respondents.

Additional research is recommended on the subject. Targetted education and pre-conception counselling, particularly for women in rural areas with lower levels of education, are also essential. Further research should also explore effective strategies to enhance maternal knowledge and health outcomes.

Conclusion

Most participants had moderate to high knowledge of BDs and risk factors, especially among educated and urban women. However, awareness of advanced maternal age, obesity, consanguinity and iodised salt remained low. There is a need to increase mass awareness about the risks associated with consanguineous marriages.

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