

Incidence and Risk Factors of Embryo-logically Diagnosed Congenital Hydrocephalus: A Study of Ultrasonography Finding in 2nd and 3rd Trimesters

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Abstract

Objective: To determine the incidence of congenital hydrocephalus (CHC) detected by ultrasonography during antenatal check-ups and to assess associated risk factors in the second and third trimesters of pregnancy.

Methodology: A descriptive cross-sectional study was conducted in the Departments of Radiology and Obstetrics & Gynaecology at Pak Red Crescent Medical and Dental College and Teaching Hospital, Dina Nath, Multan Road, Kasur. Pregnant women in the second and third trimesters presenting for routine antenatal ultrasound examination, irrespective of age and parity, were included in the study. Ultrasonography was performed using high-resolution real-time ultrasound machines with colour Doppler in all participants. The ventricular atrial width was measured at the level of the glomus of the choroid plexus during scanning. Congenital hydrocephalus was considered present when the atrial width of the lateral ventricles was found to be greater than 10 mm. Data were collected using a self-structured proforma containing study-related variables.

Results: The mean maternal age was 28.34 ± 6.44 years. Congenital hydrocephalus was identified in 5 out of 2335 women, while spina bifida was detected in 3 out of 2335 women. Among the suspected risk factors, inadequate intake of folic acid, advanced maternal age, and consanguinity were found to be the most important factors and showed significant associations with both congenital hydrocephalus and spina bifida ($p < 0.05$). Hypertension, diabetes, and infections did not show any significant association with either anomaly ($p > 0.05$).

Conclusion: The incidence of congenital hydrocephalus was approximately 5 (0.2%) out of 2335 pregnancies diagnosed by ultrasonography. Inadequate folic acid intake, advanced maternal age, and consanguinity were identified as the most common suspected risk factors during the second and third trimesters of pregnancy.

Keywords: Congenital hydrocephalus, Risk factors, Ultrasonography

Cite this article as: Nahra RA, Rasul N, Sibtain S, Tahir E, Abid M, Mahmood S. Incidence and Risk Factors of Embryo-logically Diagnosed Congenital Hydrocephalus: A Study of Ultrasonography Finding in 2nd and 3rd Trimesters. J Soc Obstet Gynaecol Pak. 2026; 16(1):42-46 DOI. 0.71104/jsogp.v16i1.1033

Introduction

Congenital hydrocephalus (CH) reflects a set of multifactorial, complex, and heterogeneous disorders of brain indicated by abnormal circulation or absorption of cerebrospinal fluid (CSF), which results in ventriculomegaly (VM), a most common addressed fetal brain abnormality.¹ Although exact prevalence remains unidentified, estimates of congenital hydrocephalus cases have been suggested to range from 1% to 5% in general population, while affecting 02 to 08 neonates per 10,000 live births.^{2,3} The global prevalence of

hydrocephalus varies by geographic, genetic, and sociodemographic factors.²⁻⁴ Global annual incidence of hydrocephalus among infants has been estimated to be over 383,000 fresh cases, with case-fatality levels reflecting 4%-87% deaths among individuals affected by hydrocephalus.⁴ A range of etiological factors can contribute to congenital hydrocephalus development, including vitamin deficiency, viral infections, developmental abnormalities, genetic predisposition, and environmental factors.^{5,6} In Pakistan around 6% to

Authorship Contribution:^{1,5} Substantial contributions to the conception or design of the work or the acquisition, ²Final approval of the study to be published, ^{4,6}Drafting the work or revising it critically for important intellectual content.

Funding Source: none

Conflict of Interest: none

Received: July 18, 2025

Revised: Nov 05, 2025

Accepted: Dec 11, 2025

9% of perinatal deaths are attributed to congenital conditions and these anomalies have been associated with maternal, genetic, sociodemographic factors, and metabolic and cardiovascular conditions.⁷

Ultrasonography remains a major diagnostic modality for the detection of cerebral ventriculomegaly and congenital hydrocephalus in obstetrical practice due to its convenience, safety, and acceptable diagnostic accuracy, particularly during the second and third trimesters compared with the first trimester.⁸⁻¹⁰ In this regard, standardized measurement of the lateral ventricles for the early identification of ventriculomegaly facilitates monitoring of disease progression, detection of associated anomalies, and assists in clinical decision-making regarding prognosis and management.¹¹

A recent study reported the detection of various brain anomalies on prenatal ultrasonography, including neural tube defects in 75.3% of cases, isolated central nervous system anomalies in 74.3%, combined central nervous system and extracranial anomalies in 5.2% and 21.9%, respectively, and ventriculomegaly in 11.0% of cases, with an overall mortality rate reaching up to 43%.¹²

Despite advancements in prenatal imaging techniques and diagnostic approaches, findings across studies remain heterogeneous. This highlights the need for population-based research incorporating second- and third-trimester sonographic evaluations to determine more accurate estimates of the incidence of congenital hydrocephalus and its associated risk factors. Therefore, the present study was conducted to determine the incidence of congenital hydrocephalus and to identify related maternal and fetal risk factors in our population. Such evidence may help improve early diagnosis, support the development of preventive strategies, and enhance antenatal counselling and management, ultimately contributing to better fetomaternal outcomes.

Methodology

This descriptive cross-sectional study was conducted in the Departments of Radiology and Obstetrics & Gynaecology at Pak Red Crescent Medical College and Teaching Hospital, Lahore. Study duration was one year from July 2023 to June 2024. All pregnant women in the second and third trimesters with singleton pregnancy, presenting for routine antenatal ultrasound examination, irrespective of age and parity, were included in the study. Women with multiple gestations (twin pregnancies), poor visualization due to maternal obesity or fetal position, the

presence of other major fetal anomalies, and those who were unwilling to participate were excluded.

A non-probability consecutive sampling technique was employed, and all eligible participants fulfilling the inclusion criteria were enrolled until the required sample size was achieved. Ultrasonographic examinations were performed using high-resolution real-time ultrasound machines with colour Doppler by a well-trained and experienced sonologist with a minimum of five years of professional experience. Standard obstetric ultrasound scanning protocols were followed. Particular attention was given to the evaluation of the third ventricle, lateral ventricles, cavum septi pellucidi, cerebral hemispheres, and posterior fossa structures.

The ventricular atrial width was measured at the level of the glomus of the choroid plexus during scanning. Congenital hydrocephalus was considered present when the atrial width of the lateral ventricles was found to be greater than 10 mm, either with or without associated findings such as a dangling choroid plexus, thinning of the cortical mantle, or enlargement of the third ventricle. Obstetric risk factors were assessed through direct maternal history and review of antenatal medical records. Data were collected using a self-structured proforma containing study-related variables, and statistical analysis was performed using SPSS version 23.

Results

The maternal average age was 28.34 ± 6.44 years, and average gestational age was 24.31 ± 5.29 weeks. Around More than half of the women were presented during the 2nd trimester (54.2%), while 45.8% were presented during third trimester of pregnancy. Out of all, 28.7% of cases had consanguinity; the satisfactory use of folic acid was noted 61.0% of women during pregnancy, while rest had history of inadequately intake of folic acid. The antenatal infections were found in 13.2% of the women, whereas the hypertensive and diabetics women were 19.2% and 14.3%, respectively. (Table I)

Out of one year presented pregnancies (2335) were evaluated by ultrasonography, where congenital hydrocephalus was found around 5/2335 (0.2%) of the women, while spina bifida was detected among 3 out of 2335 (0.1%) of the women. (Table II)

According to the suspected risk factors the inadequately given folic acid and advance maternal age were the found most important risk factors and which showed

Table I: Demographic and clinical variables of the patients. (n=2335)

Variables		N	%
Consanguinity	No	1664	71.3
	Yes	671	28.7
Use of folic acid	Adequate	1424	61.0
	Inadequate	911	39.0
Antenatal infection	No	2026	86.8
	Yes	309	13.2
Diabetes mellitus	No	2002	85.7
	Yes	333	14.3
Hypertension	No	1886	80.8
	Yes	449	19.2
Trimester of pregnancy	Second	1265	54.2
	Third	1070	45.8
Maternal mean age		28.34+6.44 years	
Mean gestational age		24.31+5.29 weeks	

Table II: Frequency of congenital disorders. (n=2335)

Congenital disorders	N	%
Congenital Hydrocephalus		
No	2330	99.8
Yes	05	0.2
Total	2335	100.0
Spina Bifida		
No	2332	99.9
Yes	03	0.1
Total	2335	100.0

significant correlations with both congenital hydrocephalus and the spina bifida $p < 0.05$. The

consanguinity was also significantly associated with congenital hydrocephalus only ($p = 0.011$). On the other hand, diabetes, and infections did not show the any significant correlation with either anomalies, $p > 0.05$ ($p > 0.05$), indicating the consanguinity, advanced maternal age and poor periconceptional folic acid intake play a key role congenital anomalies development. (Table III)

Discussion

This study included pregnant women in the second and third trimesters. The mean maternal age was 28.34 ± 6.44 years, and the mean gestational age was 24.31 ± 5.29 weeks. More than half of the women presented during the second trimester (54.7%), whereas 45.8% presented during the third trimester of pregnancy. Comparable findings were reported in a study conducted by Alia and Ahmed¹³, who evaluated antenatal ultrasound examinations in the second and third trimesters and documented a mean maternal age of 26.5 years and a mean gestational age of 24 weeks at diagnosis. Similarly, another study by Mahela and Talukdar¹⁴ reported a mean maternal age of 25.5 ± 6.15 years and a mean gestational age of 27 ± 6.42 weeks, suggesting a relatively late presentation of pregnant women for routine antenatal screening.

Table III: Analysis for post stratification. (n=2335)

Variables		Congenital Hydrocephalus		Total	p-value	Spina Bifida		Total	p-value
		No	Yes			No	Yes		
Age groups	18-25 years	1013	1	1014	0.043	1014	0	1014	0.028
		43.4%	0.0%	43.4%		43.4%	0.0%	43.4%	
		629	0	629		629	0	629	
26-32 years	26-32 years	269	0	269	0.043	269	0	269	0.028
		26.9%	0.0%	26.9%		26.9%	0.0%	26.9%	
		688	4	692		689	3	692	
33-40 years	33-40 years	295	2	297	0.043	295	1	296	0.028
		29.5%	0.2%	29.6%		29.5%	0.1%	29.6%	
		1663	1	1664		1663	1	1664	
Consanguinity	No	71.2%	0.0%	71.3%	0.011	71.2%	0.0%	71.3%	0.146
		669	2	671		669	2	671	
		28.7%	0.1%	28.7%		28.7%	0.1%	28.7%	
Yes	Yes	1998	4	2002	0.713	1999	3	2002	0.480
		85.6%	0.2%	85.7%		85.6%	0.1%	85.7%	
		332	1	333		333	0	333	
Diabetes mellitus	Yes	14.2%	0.0%	14.3%	0.713	14.3%	0.0%	14.3%	0.480
		1883	3	1886		1885	1	1886	
		80.6%	0.1%	80.8%		80.7%	0.0%	80.8%	
Hypertension	No	447	2	449	0.238	447	2	449	0.037
		19.1%	0.1%	19.2%		19.1%	0.1%	19.2%	
		1424	0	1424		1424	0	1424	
Folic acid use	adequate	61.0%	0.0%	61.0%	0.005	61.0%	0.0%	61.0%	0.030
		906	5	911		908	3	911	
		38.8%	0.2%	39.0%		38.9%	0.1%	39.0%	
Inadequate	Inadequate	2024	2	2026	0.062	2025	1	2026	0.076
		86.7%	0.1%	86.8%		86.7%	0.0%	86.8%	
		306	3	309		307	2	309	
Antenatal infection	Yes	13.1%	0.1%	13.2%	0.062	13.1%	0.1%	13.2%	0.076

According to the current study, out of all, 28.7% of cases had consanguinity, the satisfactory use of folic acid was noted 61.0% of women during pregnancy, while rest had history of inadequately intake of folic acid. Aligning with these findings, in the study of Khan et al.,¹⁵ who studies congenital anomalies among infants and reported consanguinity rate of 67.7%, which is higher than that of our study, and folic acid supplementation in 32.3% of women during pregnancy, which is lower than our finds. Similarly, in the study of Karim et al.¹⁶ higher consanguinity rate of 83.9% was found than our study findings, with 77.5% of women never consumed folic acid supplements during pregnancy, reflecting common pattern of lack of folic acid use. In current study, the antenatal infections were found in 13.2% of the women, whereas the hypertensive and diabetics women were 19.2% and 14.3%, respectively. Consistent findings were observed in the study carried out by Nahar et al.,¹⁷ who found gestational/uncontrolled diabetes among 16% and additional maternal conditions such as representing rash accompanied by fever in 12% of patients. Consistently in the study of Anbreen et al.,¹⁸ diabetes was present in 5.57% of patient, which was relatively lower than our findings.

In present study, out of one year presented pregnancies were evaluated by ultrasonography, where congenital hydrocephalus was found in around 0.2% of the women, while spina bifida was detected among 0.9% of the women. In agreement with these findings, in the study conducted by Mahela and Talukdar¹⁹ congenital abnormalities were found in 1.57% of pregnant women in their second or third trimester. Aligning with these findings, in a study conducted by Mahmoud et al.²⁰ hydrocephalus was detected in 0.4% of cases and spina bifida in 30% of hydrocephalus cases. Incidence of hydrocephalus their study was lower, while spina bifida cases were higher than those of our study. In our study cohort, according to the suspected risk factors the inadequately given folic acid and consanguinity were the found most important risk factors and which showed significant correlations with both congenital hydrocephalus and the spina bifida $p < 0.05$. Aligning with our study findings, in the study carried out by Maged et al.²¹ supplementation of folate was significantly associated with spina bifida ($p < 0.001$). They investigated consanguinity as a risk factor and did not find statistical significance. In another study conducted by Saleem et al.²² periconceptional supplementation of folic acid and high parental consanguinity were found to be significantly associated with neural tube defect ($p <$

0.01), suggesting that consanguinity and inadequate folic acid use are important risk factors in congenital anomalies.

In this study, the age of mothers, hypertension, diabetes, trimester of pregnancy and infections did not show the any significant correlation with either anomalies, $p > 0.05$ ($p > 0.05$), indicating the consanguinity and poor periconceptional folic acid intake play a key role congenital anomalies development. In agreement with our findings, in the study of Karim et al.²³ although, inadequate use of folic and consanguinity were the most frequent risk factors, use of folic, consanguinity, maternal age and diabetes were not significantly correlated with congenital anomalies ($p > 0.05$).

Conclusion

Incidence of CHC was observed around 5(0.2%)/2335 pregnancy diagnosed by ultrasonography during 2nd and 3rd trimester of pregnancy, along with the significance risk factors as low folic acid intake, advanced maternal age and three cases of spina bifida were found with neural tube defect also related to folic acid deficiency.

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