Pakistan Journal of Intensive Care Medicine
eISSN: 2708-2261; pISSN: 2958-4728
www.pjicm.com
DOI: https://doi.org/10.54112/pjicm.v5i02.127
*Pak. J. Inten. Care Med., volume 5(2), **2025**: 127*
Original Research Article

CONGENITAL ABNORMALITIES IN NEWBORNS OF CONSANGUINEOUS AND NON-CONSANGUINEOUS PARENTS

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(Received, 06th May 2025, Revised 10th June 2025, Accepted 06th July, Published 14th July 2025)

ABSTRACT

Background: *Congenital abnormalities contribute significantly to neonatal morbidity and mortality worldwide. Consanguinity is recognized as a potential risk factor for various structural and genetic malformations, yet its impact on the prevalence of specific anomalies in local populations remains underreported.* ***Objective:*** *To determine the frequency of congenital abnormalities in newborns of consanguineous versus non-consanguineous parents.* ***Study Design:*** *Cross-sectional study.* ***Setting:*** *Department of Obstetrics and Gynecology, Saidu Medical College, Swat, Pakistan.* ***Duration of Study:*** *November 1, 2024, to May 1, 2025.* ***Methods:*** *A total of 207 women with fetuses diagnosed with congenital malformations were enrolled. Diagnoses included clubfoot, congenital heart disease, cleft palate, polycystic kidney disease, and anencephaly, confirmed through physical examination and relevant diagnostic tests. Frequencies of anomalies were compared between consanguineous and non-consanguineous groups using the chi-square test, with p-values <0.05 considered statistically significant.* ***Results:*** *The mean maternal age was 27.52 ± 7.80 years. Congenital heart disease was significantly more common among consanguineous cases (17.4%) than non-consanguineous cases (3.6%) (p = 0.001). Similarly, cleft palate occurred in 15.9% vs. 2.9%, polycystic kidney disease in 7.2% vs. 1.4%, and anencephaly in 5.8% vs. 0.7%, respectively.* ***Conclusion:*** *Congenital abnormalities were significantly more frequent among newborns of consanguineous parents compared to those of non-consanguineous parents. These findings highlight the importance of genetic counseling and public health interventions in populations with high rates of consanguineous marriages.*

Keywords: Congenital Anomalies, Consanguinity, Neonatal Malformations, Genetic Risk, Pakistan, Prenatal Counseling

INTRODUCTION

Consanguinity, defined as the practice of marriage between people who are closely related genetically, has garnered considerable interest in both medical and genetic fields due to its implications for descendants. This practice, noticed in certain societies and cultures, brings forth considerable concerns about heightened risk for genetic disorders in children resulting from these unions (1, 2). The main medical issue related to consanguineous marriages (CM) is a higher risk of genetic disorders. In instances where closely related people engage in reproduction, the likelihood increases that both parents have an identical genetic mutation. This scenario heightens the probability of recessive genetic conditions manifesting in their offspring (3). In CM, the chance of both parents possessing the same recessive gene increases significantly when contrasted with non-consanguineous marriages. The prevalence of autosomal recessive disorders has become notably higher in populations in which consanguinity is commonly observed (4). Furthermore, consanguinity may result in increased expression of harmful genes, subsequently diminishing total genetic diversity within the family. The decrease in genetic diversity may lead to wider consequences that extend beyond disorders linked to specific genes (5).

A study revealed that the overall prevalence of congenital anomalies had been 3.7%, with a rate of 3.2% noticed among live births as well as 15.7% among stillbirths. Another study identified congenital anomalies in 3.61% of the overall 2188 infants examined in the study (6, 7). A study revealed that approximately one-third of infants were admitted to the unit each consecutive year throughout the study period due to congenital disabilities (8). A study on the distribution and pattern of congenital anomalies discovered that neural tube defects seemed the most prevalent anomaly in their findings (9). The study identified neural tube defects as the most prevalent congenital disabilities, taking place at a rate of 4-15 per 10,000 live births (10, 11). A study reported that club feet occur in 16.0% of consanguineous marriages and 3.0% of non-consanguineous marriages (12). Also, the prevalence of congenital anomalies was approximately 3% (13).

This study aims to determine the magnitude of congenital abnormalities in newborns associated with consanguinity. The findings of this study will be helpful for resource allocation for the management of congenital abnormalities. Secondly, if congenital anomalies are found to be higher among couples with consanguineous marriages, strategies could be devised to raise public awareness and discourage such marriages through workshops, thereby minimizing the burden of CM in our resource-poor country.

METHODOLOGY

The cross-sectional study was conducted in the Department of Obstetrics & Gynecology at Saidu Medical College, Swat, from 01-November-2024 to 01-May-2025. An ethical certificate was obtained from the hospital. We determined a sample of 207 patients based on the frequency of clubfoot in consanguineous marriages, 16.0%12, with a 5% margin of error and a 95% confidence level. Non-probability consecutive sampling was used to select participants.

Participants were enrolled from the outpatient department (OPD) and emergency units of the hospital. We selected mothers of fetuses with congenital malformations detected via ultrasound during the antenatal period, visible anomalies at birth, or anomalies identified post-birth through ultrasound or X-ray. Women were aged between 15 and 40 years and had a gestational age of 24 weeks or more. Diabetic mothers of fetuses with congenital malformations, mothers exposed to radiation during the first trimester, and those exposed to infectious agents known to cause congenital anomalies were not included.

Data collection commenced after consent was secured from all participants. A proforma was used to record maternal age, method of delivery, number of births, history of stillbirths and miscarriages,

gravidity, parity, history of infertility, family history of congenital malformations, and parental consanguinity. Neonatal characteristics such as sex and the presence of congenital malformations were also documented. The anomalies were classified according to the International Classification of Diseases to ensure diagnostic standardization.

We examined several congenital malformations, such as clubfoot, characterized by the foot pointing downward and inward upon physical examination. Congenital heart disease is identified by clinical features such as rapid heartbeat (>100 bpm), rapid breathing (>60 breaths/min), edema in the extremities or periorbital region, fatigue, cyanosis, or feeding difficulties, with confirmation via echocardiography, revealing structural or functional defects. Cleft palate was diagnosed based on feeding difficulties, nasal regurgitation, or hypernasal speech, verified by a pediatrician's physical assessment of the palatal cleft. Polycystic kidney disease was detected through ultrasonography, showing enlarged hyperechoic kidneys with microcysts and loss of corticomedullary differentiation, and anencephaly confirmed by prenatal ultrasound after 14 weeks, either by the absence of cerebral tissue above the orbits or the presence of characteristic "frog eye" or "Mickey Mouse" morphology in the coronal plane.

Data analysis was conducted with SPSS 15. Quantitative variables were evaluated as mean ± standard deviation. Categorical variables were evaluated using frequencies and percentages. Congenital anomalies were assessed according to consanguinity. Stratification of congenital malformations was conducted based on demographics and clinical history. The chi-square test was applied for assessment and stratification, with a p-value ≤ 0.05 considered statistically notable.

RESULTS

Mean maternal age was 27.52±7.80 years. Mean gestational age was 29.02±3.24 weeks. The mean gravidity was 2.28±0.96, and parity was 1.71±0.75. Neonatal gender distribution showed that there were 108 (52.2%) males and 99 (47.8%) females. Vaginal delivery was the most common method, 147 (71.0%), while cesarean sections were performed in 60 (29.0%) cases. Consanguineous marriages were identified in 69 (33.3%) cases, while 138 (66.7%) were non-consanguineous. The clinical history can be seen in Table 1.

We observed clubfoot in 19 (9.2%) neonates, congenital heart disease in 17 (8.2%), cleft palate in 15 (7.2%), polycystic kidney disease in 7 (3.4%), and anencephaly in 5 (2.4%). Clubfoot was more prevalent in neonates with consanguineous parents (13, 18.8%) than non-consanguineous ones (6, 4.3%) (P = 0.001). Congenital heart disease occurred in 12 (17.4%) consanguineous cases and 5 (3.6%) non-consanguineous cases (P = 0.001), and cleft palate was present in 11 (15.9%) and 4 (2.9%) cases, respectively (P = 0.001). Polycystic kidney disease and anencephaly also showed higher frequencies in consanguineous groups 5 (7.2%) and 4 (5.8%) compared to non-consanguineous 2 (1.4%) and 1 (0.7%) (P = 0.03), (P = 0.02). Stratification of congenital anomalies with demographics and clinical history can be seen from Table 4 to Table 11.

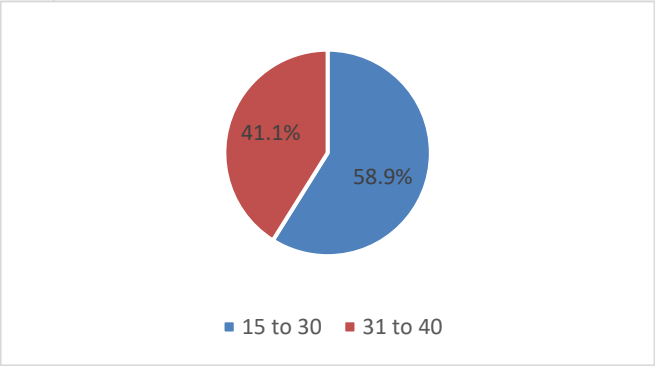

Figure 1: Maternal age distribution (Years)

Table 1: Demographics and clinical history

Demographics and clinical history		N	%
Neonatal gender	Male	108	52.2%
	Female	99	47.8%
Method of delivery	Vaginal	147	71.0%
	Caesarean section	60	29.0%
No of births	0 to 2	172	83.1%
	> 2	35	16.9%
Stillbirth and miscarriage	Yes	39	18.8%
	No	168	81.2%
Gravidity	1 to 3	188	90.8%
	> 3	19	9.2%
History of infertility	Yes	24	11.6%
	No	183	88.4%
Consanguinity	Consanguineous	69	33.3%
	Non-consanguineous	138	66.7%

Table 2: Congenital abnormalities

Congenital abnormalities		N	%
Clubfoot	Yes	19	9.2%
	No	188	90.8%
Congenital Heart Disease	Yes	17	8.2%
	No	190	91.8%
Cleft Palate	Yes	15	7.2%
	No	192	92.8%
Polycystic kidney	Yes	7	3.4%
	No	200	96.6%
Anencephaly	Yes	5	2.4%
	No	202	97.6%

Table 3: Congenital abnormalities according to consanguinity

Congenital abnormalities		Consanguinity				P value
		Consanguineous		Non-consanguineous		
		N	%	N	%	
Clubfoot	Yes	13	18.8%	6	4.3%	0.001
	No	56	81.2%	132	95.7%	
Congenital Heart Disease	Yes	12	17.4%	5	3.6%	0.001
	No	57	82.6%	133	96.4%	
Cleft Palate	Yes	11	15.9%	4	2.9%	0.001
	No	58	84.1%	134	97.1%	
Polycystic kidney	Yes	5	7.2%	2	1.4%	0.03
	No	64	92.8%	136	98.6%	
Anencephaly	Yes	4	5.8%	1	0.7%	0.02
	No	65	94.2%	137	99.3%	

[Citation: Sabahat, K., Naveed, P. (2025). Congenital abnormalities in newborns of consanguineous and non-consanguineous parents. *Pak. J. Inten. Care Med. 5(2), **2025**: 127*. doi: https://doi.org/10.54112/pjicm.v5i02.127]

Table 4: Stratification of congenital abnormalities with maternal age

Congenital abnormalities		Age distribution (Years)				P value
		15 to 30		31 to 40		
		N	%	N	%	
Clubfoot	Yes	11	9.0%	8	9.4%	P > 0.05
	No	111	91.0%	77	90.6%	
Congenital Heart Disease	Yes	8	6.6%	9	10.6%	P > 0.05
	No	114	93.4%	76	89.4%	
Cleft Palate	Yes	10	8.2%	5	5.9%	P > 0.05
	No	112	91.8%	80	94.1%	
Polycystic kidney	Yes	3	2.5%	4	4.7%	P > 0.05
	No	119	97.5%	81	95.3%	
Anencephaly	Yes	3	2.5%	2	2.4%	P > 0.05
	No	119	97.5%	83	97.6%	

Table 5: Stratification of congenital abnormalities with neonatal gender

Congenital abnormalities		Neonatal gender				P value
		Male		Female		
		N	%	N	%	
Clubfoot	Yes	10	9.3%	9	9.1%	P > 0.05
	No	98	90.7%	90	90.9%	
Congenital Heart Disease	Yes	11	10.2%	6	6.1%	P > 0.05
	No	97	89.8%	93	93.9%	
Cleft Palate	Yes	6	5.6%	9	9.1%	P > 0.05
	No	102	94.4%	90	90.9%	
Polycystic kidney	Yes	4	3.7%	3	3.0%	P > 0.05
	No	104	96.3%	96	97.0%	
Anencephaly	Yes	3	2.8%	2	2.0%	P > 0.05
	No	105	97.2%	97	98.0%	

Table 6: Stratification of congenital abnormalities with method of delivery

Congenital abnormalities		Method of delivery				P value
		Vaginal		Caesarean section		
		N	%	N	%	
Clubfoot	Yes	13	8.8%	6	10.0%	P > 0.05
	No	134	91.2%	54	90.0%	
Congenital Heart Disease	Yes	12	8.2%	5	8.3%	P > 0.05
	No	135	91.8%	55	91.7%	
Cleft Palate	Yes	11	7.5%	4	6.7%	P > 0.05
	No	136	92.5%	56	93.3%	
Polycystic kidney	Yes	4	2.7%	3	5.0%	P > 0.05
	No	143	97.3%	57	95.0%	
Anencephaly	Yes	3	2.0%	2	3.3%	P > 0.05
	No	144	98.0%	58	96.7%	

Table 7: Stratification of congenital abnormalities with the number of births

Congenital abnormalities		No of births				P value
		0 to 2		> 2		
		N	%	N	%	
Clubfoot	Yes	15	8.7%	4	11.4%	P > 0.05
	No	157	91.3%	31	88.6%	
Congenital Heart Disease	Yes	16	9.3%	1	2.9%	P > 0.05
	No	156	90.7%	34	97.1%	
Cleft Palate	Yes	10	5.8%	5	14.3%	P > 0.05
	No	162	94.2%	30	85.7%	
Polycystic kidney	Yes	6	3.5%	1	2.9%	P > 0.05
	No	166	96.5%	34	97.1%	
Anencephaly	Yes	4	2.3%	1	2.9%	P > 0.05
	No	168	97.7%	34	97.1%	

Table 8: Stratification of congenital abnormalities with stillbirth and miscarriage

Congenital abnormalities	Stillbirth and miscarriage	P value

		Yes		No		
		N	%	N	%	
Clubfoot	Yes	3	7.7%	16	9.5%	P > 0.05
	No	36	92.3%	152	90.5%	
Congenital Heart Disease	Yes	4	10.3%	13	7.7%	P > 0.05
	No	35	89.7%	155	92.3%	
Cleft Palate	Yes	2	5.1%	13	7.7%	P > 0.05
	No	37	94.9%	155	92.3%	
Polycystic kidney	Yes	2	5.1%	5	3.0%	P > 0.05
	No	37	94.9%	163	97.0%	
Anencephaly	Yes	0	0.0%	5	3.0%	P > 0.05
	No	39	100.0%	163	97.0%	

Table 9: Stratification of congenital abnormalities with stillbirth and miscarriage

Congenital abnormalities		Gravidity				P value
		1 to 3		> 3		
		N	%	N	%	
Clubfoot	Yes	17	9.0%	2	10.5%	P > 0.05
	No	171	91.0%	17	89.5%	
Congenital Heart Disease	Yes	16	8.5%	1	5.3%	P > 0.05
	No	172	91.5%	18	94.7%	
Cleft Palate	Yes	12	6.4%	3	15.8%	P > 0.05
	No	176	93.6%	16	84.2%	
Polycystic kidney	Yes	7	3.7%	0	0.0%	P > 0.05
	No	181	96.3%	19	100.0%	
Anencephaly	Yes	4	2.1%	1	5.3%	P > 0.05
	No	184	97.9%	18	94.7%	

Table 10: Stratification of congenital abnormalities with a history of infertility

Congenital abnormalities		History of infertility				P value
		Yes		No		
		N	%	N	%	
Clubfoot	Yes	2	8.3%	17	9.3%	P > 0.05
	No	22	91.7%	166	90.7%	
Congenital Heart Disease	Yes	2	8.3%	15	8.2%	P > 0.05
	No	22	91.7%	168	91.8%	
Cleft Palate	Yes	2	8.3%	13	7.1%	P > 0.05
	No	22	91.7%	170	92.9%	
Polycystic kidney	Yes	1	4.2%	6	3.3%	P > 0.05
	No	23	95.8%	177	96.7%	
Anencephaly	Yes	0	0.0%	5	2.7%	P > 0.05
	No	24	100.0%	178	97.3%	

DISCUSSION

The findings of this study showed notable associations between consanguinity and congenital anomalies, which align with trends observed in similar research across varied populations. Maternal age in our research averaged about 27.52±7.80 years, which is comparable to the demographic reported by Fatema et al, where the majority of participants were aged 20–25 years (14). A striking observation in our data was the higher frequency of specific anomalies among consanguineous pregnancies. Clubfoot, for example, occurred in 18.8% of consanguineous cases compared to 4.3% in non-consanguineous ones. This disparity mirrors the results of Mosayebi et al, who reported musculoskeletal anomalies in 22.0% of consanguineous neonates, nearly double the rate in non-consanguineous cases (15). Similarly, congenital heart disease was more frequently observed in our consanguineous group (17.4%) compared to non-consanguineous (3.6%), a trend consistent with Yunis et al, who noted a 1.8–3.9-fold increased risk of cardiac defects in first-cousin unions (12). These observations underscore the increased genetic risk conferred by a similar gene pool, particularly for autosomal recessive conditions.

Neural tube defects (NTDs), though less frequent in our study, were notably higher in consanguineous pregnancies (5.8%) than in non-consanguineous (0.7%). This aligns well with Butt et al, who identified NTDs as the most frequent anomaly in consanguineous Pakistani mothers, with anencephaly alone accounting for 42.86% of cases (16). The stark contrast between our anencephaly rates and those reported by Butt et al may be due to their smaller sample of 24 patients. Nevertheless, both studies highlight the risks of consanguineous pregnancies to severe CNS malformations, reinforcing the need for targeted interventions such as mandatory folic acid fortification.

The predominance of cleft palate in consanguineous neonates (15.9%) compared to non-consanguineous (2.9%) further supports the role of recessive gene expression. This finding resonates with Butt et al's work, where cleft lip/palate constituted 9.52% of anomalies, and they reported that cousin marriages were at 83.33% risk of having congenital anomalies.[16] The recurrence of such patterns across studies suggests that craniofacial defects are common in consanguinity, likely due to homozygous mutations in developmental genes.

Polycystic kidney disease, though rare in our cohort (7.2% consanguineous vs. 1.4% non-consanguineous), aligns with Anbreen et al.'s report of renal anomalies in 11.11% neonates; their study had around 67.67% cases of consanguinity (17).

Methodologically, our study relied on clinical diagnoses without genetic testing, which is a limitation of our study. Future studies should integrate chromosomal analysis.

CONCLUSION

In conclusion, the frequency of congenital abnormalities in newborns of consanguineous parents was significantly higher than that of non-consanguineous parents. Our study showed that consanguineous marriages can considerably increase the risk of congenital abnormalities in newborns, which highlights the urgent need for genetic counseling and enhanced prenatal screening in such populations.

DECLARATIONS

Data Availability Statement

All data generated or analysed during the study are included in the manuscript.

Ethics approval and consent to participate

Approved by the department Concerned. (IRB)

Consent for publication

Approved

Funding

Not applicable

CONFLICT OF INTEREST

The authors declare no conflict of interest.

AUTHOR CONTRIBUTION

KOMAL SABAHAT (Postgraduate Resident -OBG GYNAE***)***
Data Collection, Conception of Study, Development of Research Methodology Design, Data Entry, Data Analysis, Manuscript Drafting, Review of Manuscript, and Final Approval of Manuscript.
PARVEEN NAVEED (Assistant Professor OBS-GYNAE***)***
Critical input, and Final Approval of Manuscript.

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