



ISSN: 2091-2749 (Print)
2091-2757 (Online)

Submitted on: 2026 Mar 30
Accepted on: 2026 Jun 23

<https://doi.org/10.3126/jpahs.v13i1.96120>

Termination of pregnancy for congenital fetal malformations: a retrospective review in a tertiary care center

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Abstract

Introduction: Congenital fetal anomalies are a significant cause of perinatal morbidity and mortality, affecting approximately 3% of pregnancies worldwide. This study aimed to evaluate the patterns of congenital anomalies, methods of detection, and outcomes of pregnancy termination.

Method: A retrospective study was conducted in the Department of Obstetrics and Gynaecology, Patan Hospital, Nepal, from April 2021 to March 2025. Medical records of 78 women who underwent termination of pregnancy for congenital fetal malformations were reviewed. Maternal demographics, diagnostic modalities, type of anomaly, time taken for decision-making, method of termination, and outcomes were analyzed.

Result: Most women were aged 21–30 years (71.8%), and 56.4% were primigravida. Fetal anomalies were most commonly diagnosed between 21 and 30 weeks of gestation (61.5%), predominantly during the anomaly scan (51.3%). Central nervous system anomalies were the most common (33.3%), followed by multisystem anomalies (30.8%) and congenital heart disease (19.2%). More than half of the women (51.3%) decided to terminate the pregnancy within three days of diagnosis. Medical termination was the predominant method (88.5%), followed by Foley's catheter induction (7.7%). Most women achieved expulsion within 24 hours.

Conclusion: The anomaly scan was the most common diagnostic modality, with the central nervous system being the most frequently affected system. Medical termination was effective and feasible, with high success rates. Wider implementation of first-trimester screening and streamlined referral pathways may facilitate earlier diagnosis and management of congenital fetal anomalies.

Keywords: Anomaly scan; Congenital anomalies; Pregnancy termination; Prenatal diagnosis



How to Cite: Timalisina G, Gurung P, Jaiswal E, Dangol C, Gautam N, Timalisina A. Termination of pregnancy for congenital fetal malformations: a retrospective review in a tertiary care center. *J Patan Acad Health Sci.* 2026 Jun;13(1):80-85.

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Introduction

Congenital fetal anomalies are a major cause of perinatal morbidity and mortality and may result in lifelong disability. Approximately 3% of pregnancies are affected by genetic or structural anomalies worldwide.¹ Early prenatal detection is essential for parental counselling, pregnancy management, and informed decision-making. Routine antenatal ultrasonography has improved the prenatal diagnosis of congenital anomalies, enabling timely intervention and management.^{2–5}

Despite advances in prenatal screening, delayed diagnosis and management remain common in low- and middle-income countries because of limited access to antenatal care, late booking of pregnancy, inadequate expertise in anomaly screening, and referral delays.⁶ Central nervous system anomalies are among the most frequently detected congenital malformations, followed by gastrointestinal, renal, and cardiovascular anomalies.^{7,8} For severe fetal anomalies, termination of pregnancy may be considered using medical, mechanical, or surgical methods depending on gestational age and clinical circumstances.⁹

However, there is limited evidence from Nepal regarding the spectrum of congenital fetal anomalies, timing of diagnosis, parental decision-making, and outcomes of pregnancy termination. Such information is important for improving prenatal screening services, counselling, referral pathways, and termination protocols.

Therefore, this study aimed to evaluate the clinical profile of women undergoing termination of pregnancy for congenital fetal anomalies, identify the common anomalies detected, assess gestational age at diagnosis and time taken for decision-making, and determine the methods and outcomes of pregnancy termination at a tertiary care hospital in Nepal.¹⁰

Method

This was a retrospective descriptive study conducted in the Department of Obstetrics and Gynaecology at Patan Hospital, a tertiary care teaching hospital under Patan Academy of Health Sciences (PAHS), Lagankhel, Lalitpur, Nepal. The study period was from April 2021 to March 2025. Ethical approval was obtained from the Institutional Review Committee (IRC) of Patan Academy of Health Sciences (approval reference number: drs2512192179).

All women who underwent termination of pregnancy due to prenatally diagnosed congenital fetal anomalies during the study period were included. Pregnancies complicated by Rh isoimmunization, cases referred to other centres for termination, and

records with incomplete or missing essential data were excluded. Termination of pregnancy was defined as the intentional medical or surgical interruption of pregnancy following prenatal diagnosis of structural or functional fetal anomalies that are incompatible with life, associated with severe disability, or significantly affecting postnatal quality of life.

Data were extracted retrospectively from hospital medical records using a structured pro-forma. Variables included maternal demographics (age, gravidity, parity), gestational age at diagnosis, interval between diagnosis and decision for termination, and relevant maternal comorbidities such as diabetes mellitus, hypertension, and hypothyroidism. Diagnostic modalities recorded included nuchal translucency/nasal bone scan, biochemical screening, anomaly scan, and fetal echocardiography. Outcome variables included method of termination (medical termination, Foley catheter induction, or operative intervention) and fetal outcomes such as type and system of anomaly, presence of gross malformations, fetal weight, and sex.

Data confidentiality and privacy were strictly maintained. Patient identifiers were removed during data extraction, and each case was assigned a unique study code. Access to data was restricted to the research team only. Data were analyzed using Microsoft Excel 2013.

Result

A total of 78 women were included in the study. The majority were aged 21–30 years 56 (71.8%), followed by 31–40 years 16 (20.5%). More than half of the participants were primigravida accounted for 44 (56.41%), multigravida women with parity G2–G5 accounted for 33 (42.30%). Regarding risk factors, 50 (64.1%) women had no identifiable comorbidities. Among those with medical conditions, hypothyroidism was the most common 11 (14.1%), followed by diabetes nine (11.53%) and hypertension six (7.69%).

Most congenital anomalies were diagnosed between 21 and 30 weeks of gestation 48 (61.54%), while 19 (24.36%) were diagnosed at 10–20 weeks and 11 (14.10%) after 30 weeks. Most of the patient 40 (51.28%) made the decision for termination of pregnancy within 1–3 days of diagnosis. As most of the cases were diagnosed in our center and genetic counselling by obstetrician and paediatric counselling and additional investigations required were available in our centre so most of the decisions for termination were made earlier, Table 1.

Among first-trimester screening parameters, nuchal translucency/nasal bone (NT/NB) assessment was

missed by the majority of women 55 (70.51%). Twenty-three women had done the NTNB scan among

Table 1. Demographic and obstetric characteristics of mothers undergoing pregnancy termination (N=78)

Variables	Category	f (%)
Age (Years)	≤20	3(3.85%)
	21–30	56(71.79%)
	31–40	16(20.51%)
	>40	3(3.85%)
Gravida	G1	44(56.41%)
	G2–G5	33(42.31%)
	>G5	1(1.28%)
Risk Factors	Diabetes	9(11.54%)
	Hypertension	6(7.69%)
	Hypothyroidism	11(14.10%)
	Age >35 years	10(12.82%)
	None	50(64.10%)
Gestational Age at Time of Diagnosis (Weeks)	10–20	19(24.36%)
	21–30	48(61.54%)
	>30	11(14.10%)
Time Taken for Decision of Termination (Days)	1–3	40(51.28%)
	4–6	21(26.92%)
	≥7	17(21.79%)

which, 19(24.36%) had normal findings, while 4(5.13%) showed abnormal results. Similarly, biochemical markers (Dual and Quadruple) were not performed in 60 (76.92%) women; 13 (16.67%) had normal results and 5 (6.41%) had abnormal findings. Most of the anomalies were identified during anomaly scan where 40 (51.28%) demonstrates abnormal findings and 23 (29.49%) reported as normal; it was not performed in 15 (19.23%) cases. Some of the women underwent fetal echocardiography as an additional investigations, among those evaluated, 12 (15.38%) had normal findings and 9 (11.54%) had abnormal results, Table 2.

Table 2. Results of prenatal screening and diagnostic investigations for congenital anomalies (N=78)

Factors	Result	f (%)
NT/NB	Normal	19(24.36%)
	Abnormal	4(5.13%)
	Not done	55(70.51%)
Biochemical Markers	Normal	13(16.67%)
	Abnormal	5(6.41%)
	Not done	60(76.92%)
Anomaly Scan	Normal	23(29.49%)
	Abnormal	40(51.28%)
	Not done	15(19.23%)
Fetal Echocardiography	Normal	12(15.38%)
	Abnormal	9(11.54%)

Most of the foetus, 23 (29.49%), has multiple congenital anomalies, followed by congenital heart disease in 15 (19.23%), Acrania in 11 (14.10%) cases while dysplastic kidney and other anomalies each

accounted for 7 cases (8.97%). Cystic hygroma and holoprosencephaly was present in 6 cases (7.69%) and 5 cases (6.41%) respectively. Gastroschisis/omphalocele was the least common specific anomaly, seen in 4 cases (5.13%). The nervous system was the most affected 26 (33.33%), followed by involvement of multiple system 24 (30.77%). Cardiovascular anomalies accounted for 15 cases (19.23%), renal system involvement for 8 cases (10.26%) and Gastrointestinal anomalies were identified in 5 cases (6.41%), while no pulmonary system anomalies were reported during the study period, Table 3,4.

Table 3. Distribution of commonly observed congenital anomalies among fetuses diagnosed with congenital anomalies (N=78)

Congenital Anomaly	f (%)
Cystic hygroma	6(7.69%)
Acrania	11(14.10%)
Gastroschisis/Omphalocele	4(5.13%)
Holoprosencephaly	5(6.41%)
Congenital heart disease	15(19.23%)
Dysplastic kidney	7(8.97%)
Multiple anomalies	23(29.49%)
Other anomalies	7(8.97%)

Table 4. Distribution of congenital anomalies according to the affected organ system among fetuses diagnosed with congenital anomalies (N=78)

Organ System Affected	f (%)
Nervous system	26(33.33%)
Cardiovascular system	15(19.23%)
Gastrointestinal system	5(6.41%)
Pulmonary system	-
Multisystem involvement	24(30.77%)
Renal system	8(10.26%)

The anomaly scan, routinely performed between 18 and 22 weeks of gestation, was the primary modality for detecting congenital anomalies. As the most detailed fetal ultrasonographic examination, it identified most anomalies, while some anomalies that developed later in gestation were detected during subsequent obstetric ultrasound examinations.

Medical methods 69 (88.46%) was the most common method employed for termination of pregnancy while Foley's catheter induction was used in six (7.69%) and caesarean section in 3 cases (3.84%). Regarding use of medical methods, all of them were induced with mifepristone whereas misoprostol requirement was different in different individuals, most women required two doses 24 (34.78%), followed by three doses in 14 (20.28%) and five doses in 13 (18.84%) cases. A single dose was sufficient in seven (10.14%) cases whereas nine (13.04%) cases required four doses and only two cases (2.56%) required more than five doses of misoprostol. Majority of cases 61 (78.20%), expelled the product of conception within a day while 10 (12.82%) cases required 1–7 days and five (6.41%) women took more than seven days. Surgical

evacuation (ERPC) was required in 20 (25.64%) cases, whereas 58 (74.35%) cases did not require further intervention, Table 5.

Table 5. Maternal and procedural outcomes following medical termination of pregnancy for fetal congenital anomalies (N=69)

Outcome		f (%)
Mode of termination	Medical method	69(88.46%)
	Foleys induction	6(7.69%)
	Cesarean section	3(3.84%)
Doses of misoprostol required	1	7(10.14%)
	2	24(34.78%)
	3	14(20.28%)
	4	9(13.04%)
	5	13(18.84%)
	>5	2(2.56%)
Duration taken for expulsion(days)	0-1	61(78.20%)
	46204	10(12.82%)
	>7	5(6.41%)
Requirement of ERPC	Yes	20(25.64%)
	No	58(74.35%)

Gross malformations like anencephaly, omphalocele were observed in 11 (16.41%) cases while the majority, 67 (85.89%) cases did not exhibit externally visible malformations. Thirty-five fetuses (44.87%) were male and 23 (29.48%) were female, whereas sex could not be determined in 20 cases (25.64%), likely due to early gestational age or advanced maceration. Regarding fetal weight distribution, more than half of the fetuses 44 (56.41%) weighed less than 500 gm at the time of expulsion, 119 (4.35%) fetuses weighed between 500–1000 gm, six (7.69%) weighed 1000–1500 gm four (5.12%) weighed 1500–2000 g. Only a small proportion three (3.84%) had higher birth weights of 2000–2500 gm, and two (2.56%) cases of more than 2500 gm, Table 6.

Table 6. Fetal outcome after termination of pregnancy(N=78)

Fetal Outcome		f (%)
Gross malformation	Yes	11(16.41%)
	No	67(85.89%)
Sex of fetus	Male	35(44.87%)
	Female	23(29.48%)
	Unknown	20(25.64%)
Weight of fetus(gm)	<500	44(56.41%)
	500-1000	19(24.35%)
	1000-1500	6(7.69%)
	1500-2000	4(5.12%)
	2000-2500	3(3.84%)
	>2500	2(2.56%)

Discussion

Current recommendations from the Society for Maternal-Fetal Medicine and American College of Obstetricians and Gynecologists advise a first

trimester dating ultrasound at 10–13⁺ weeks followed by a detailed fetal anomaly scan at 18–22 weeks of gestation. First-trimester ultrasonography primarily establishes fetal viability, accurate gestational dating, and chorionicity in multiple pregnancies, whereas the second-trimester scan enables systematic assessment of fetal structural abnormalities and placental location.¹¹ In our cohort, however, a substantial proportion of women did not undergo first-trimester screening, including nuchal translucency and nasal bone assessment. due to lack of awareness, healthcare facilities & it require expert clinician, advanced lab and good socioeconomic status of the patient. As a result, certain major anomalies such as acrania and anencephaly were detected only during the second-trimester anomaly scan. This finding underscores the pivotal role of the routine anomaly scan in antenatal care, particularly in settings where early pregnancy screening uptake remains suboptimal. The anomaly scan provides a comprehensive structural evaluation of the fetus and remains one of the most effective strategies for early detection of congenital anomalies and guiding subsequent prenatal management.^{12,13}

In the present study, most women reached a decision regarding termination within three days following diagnosis; however, a subset required more than seven days. This delay was largely attributable to the need for additional investigations including fetal MRI for central nervous system anomalies, fetal echocardiography for suspected congenital heart disease—and multidisciplinary counselling involving fetal medicine specialists and paediatricians, particularly in pregnancies with advanced gestational age. The increasing complexity of prenatal diagnosis and the need for confirmatory imaging may prolong the decision-making process in cases of late-detected anomalies. Therefore, establishing a structured and expedited referral pathway is essential to facilitate timely evaluation and counselling, thereby minimizing delays that may approach legal gestational limits for termination.¹⁴

A study done in South Africa reported that severe central nervous system abnormalities accounted for 44 cases (77.2%), while multiple system involvement was observed in 19 cases (33.3%).¹⁵ Consistent with these findings, central nervous system anomalies constituted the most frequent abnormalities in our cohort, followed by multisystem involvement. Cardiovascular and renal anomalies were comparatively less common. This distribution reflects the well-recognised predominance of neural tube defects and other central nervous system anomalies among prenatally detected congenital malformations.

Medical termination of pregnancy using mifepristone in combination with prostaglandins or their analogues remains the most widely used and

effective approach for second-trimester termination due to its accessibility, safety, and high success rate.¹⁶ Misoprostol (a prostaglandin E1 analogue) is extensively utilised for cervical ripening and labour induction; however, its use may be associated with uterine hyperstimulation and, rarely, uterine rupture, particularly in women with a prior caesarean section. Careful patient selection and close monitoring are therefore essential when employing misoprostol in such clinical contexts.^{17,18}

Mechanical methods such as transcervical Foley catheter insertion represent an alternative strategy for induction of labour. The Foley catheter promotes cervical ripening through mechanical dilatation and stimulation of endogenous prostaglandin release.¹⁹ In our study, medical methods were used for termination in the majority of cases (88.46%), while Foley catheter induction accounted for 7.69% of cases. Caesarean delivery was required in three cases (3.84%), reflecting situations where vaginal termination was not feasible or complications arose.

A Brazilian study reported a mean induction-to-abortion interval of 2.3 days, with 81.8% of abortions occurring within the first 72 hours.²⁰ In contrast, our study demonstrated a shorter induction-to-expulsion interval in most cases, with 78.20% achieving expulsion within 24 hours. However, 12.82% required 1–7 days and 6.41% required more than seven days, necessitating a second cycle of misoprostol. These findings highlight the variability in induction outcomes and emphasize the importance of individualized management protocols for second-trimester termination.

Conclusion

Prenatal diagnosis of congenital anomalies was primarily achieved through ultrasonography, particularly the NT/NB scan and anomaly scan highlighting the importance of this scan routinely. Central nervous system anomalies were the most commonly detected abnormalities, followed by multisystem and congenital heart defects which require further confirmatory evaluation and counselling which delay the time for diagnosis and decision for termination. Medical termination using mifepristone and misoprostol was the predominant method, with successful expulsion occurring within 24 hours in most cases, demonstrating its effectiveness and feasibility in clinical practice.

Acknowledgement

None

Conflict of Interest

None

Funding

None

Author Contribution

Concept, design, planning: GT, PG, EJ, CD, NG, AT; Literature review: GT, AT; Data collection: GT, NG; Data analysis: GT, CD; Draft manuscript: GT, PG, AT; Revision of draft: GT, PG, EJ; Final manuscript: GT, PG, EJ, CD, NG, AT; Accountability of the work: GT, PG, EJ, CD, NG, AT; Guarantor: GT.

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