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PREVALENCE AND SPECTRUM OF INTRAUTERINE CONGENITAL ANOMALIES DIAGNOSED BY ULTRASONOGRAPHY ACROSS ALL TRIMESTERS AT A TERTIARY REFERRAL CENTER IN WEST NUSA TENGGARA HOSPITAL: A 2024-2025 CROSS- SECTIONAL STUDY

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ABSTRACT

Introduction: Congenital anomalies are a major cause of perinatal morbidity and mortality worldwide. Prenatal ultrasonography enables early detection of fetal structural abnormalities, supporting appropriate management planning and obstetric counseling. However, data on the prevalence and spectrum of intrauterine congenital anomalies in West Nusa Tenggara remain limited.

Objective : To determine the prevalence and spectrum of intrauterine congenital anomalies detected by ultrasonography and to describe associated maternal and obstetric characteristics at a tertiary referral center in West Nusa Tenggara Provincial Hospital during 2024–2025.

Methods: A hospital-based cross-sectional study was conducted at the fetomaternal clinic from 2024 to 2025. A total of 717 pregnancies undergoing obstetric ultrasonography were included using consecutive sampling. Prevalence was calculated with 95% confidence intervals (CI), and related variables were analyzed descriptively.

Results: Seventy-eight cases of congenital anomalies were identified, yielding a prevalence of 10.9% (95% CI: 8.6–13.2%). Most cases occurred in women aged 26–30 years (30.77%) and multigravida (67.95%). The majority were detected in the second trimester (53.85%). Central nervous system anomalies were most common (30.76%), followed by cardiovascular anomalies (20.51%). Most cases involved a single organ system (74.35%), and delivery was predominantly by cesarean section (73.08%).

Conclusions: The prevalence in this tertiary referral center is higher than regional estimates, likely reflecting referral bias. Central nervous system and cardiovascular anomalies were the most common. Strengthening early prenatal screening and developing population-based registries are essential.

Keywords: Congenital anomalies; tertiary referral center; prevalence; antenatal ultrasonography.

INTRODUCTION

Intrauterine congenital anomalies (birth defects or congenital malformations) are defined as single or multiple structural abnormalities identified during pregnancy and confirmed at birth.^{1–3} According to data from the World Health Organization (WHO) in 2023, approximately 240,000 newborns die each year due to congenital anomalies.⁴ The prevalence of congenital anomalies is estimated to range from 3% to 6% of live births.^{5,6} Other studies have reported that congenital anomalies represent the fifth

leading cause of neonatal mortality, with a prevalence of 2% to 3% of live births.^{7,8} Congenital anomalies contribute to spontaneous abortion, stillbirth, perinatal mortality, as well as long-term morbidity and the need for intensive neonatal care.^{9,10} In Asia, the prevalence of congenital anomalies has been reported within the WHO South-East Asian Region Newborn and Birth Defects Database (SEAR-NBBD). The reported prevalence varies widely (0.21%–9.68%), likely due to differences in study design, classification systems, and reporting methods.¹¹

Congenital anomalies are classified as single or multiple based on the number of affected organ systems, including the central nervous, cardiovascular, pulmonary, gastrointestinal, musculoskeletal, genitourinary, and orofacial systems.¹²⁻¹⁵ Maternal obstetric factors—such as maternal age, parity, plurality, residence, comorbidities, medication exposure, and mode of delivery—are associated with the occurrence, detection, and outcomes of congenital anomalies.^{14,16-21} Advances in ultrasonography have enhanced prenatal detection across all trimesters, enabling detailed anatomical assessment, classification by organ involvement, and prognostic evaluation through serial monitoring.^{14,22-24} However, incidence rates reported in tertiary referral centers are often higher due to the concentration of high-risk cases.

West Nusa Tenggara, as a region characterized by an archipelagic geography and unequal distribution of healthcare facilities, faces unique challenges in maternal and fetal healthcare services. Data on the prevalence and spectrum of intrauterine congenital anomalies diagnosed by ultrasonography in this region remain limited, despite their importance for planning screening strategies, improving the quality of antenatal care, and developing evidence-based maternal and perinatal health policies grounded in local data.

OBJECTIVE

This study aimed to determine the prevalence and spectrum of intrauterine congenital anomalies detected by ultrasonography across all trimesters of pregnancy at

a tertiary referral center during the 2024–2025 period, as well as to analyze maternal obstetric characteristics associated with these cases.

METHODS

This study was a hospital-based cross-sectional study conducted at the Fetomaternal Clinic of a tertiary referral center in West Nusa Tenggara Province during 2024–2025. All pregnancies undergoing fetomaternal ultrasonographic examination during the study period were included using a consecutive sampling method. Intrauterine congenital anomalies were defined as fetal structural abnormalities detected by ultrasonography from the first to the third trimester. In cases that progressed to delivery, the diagnosis was confirmed clinically or through additional investigations. Pregnancy termination was performed in cases of severe or lethal anomalies following appropriate counseling.

A total of 717 ultrasonographic examinations were performed by fetomaternal obstetrics and gynecology consultants during the study period, all of which were included in the prevalence estimation. Cases identified as congenital anomalies on antenatal ultrasonography but subsequently found to be normal at delivery were excluded from the study. Cases detected antenatally were confirmed at the time of delivery. Pregnancy termination was performed in cases with lethal anomalies following adequate counseling.

Data were collected from medical records using a structured form. Maternal demographics included age, education, and residence, while obstetric profiles comprised parity, plurality, gestational age at diagnosis, risk factors, fetal prognosis, pregnancy outcome, and mode of delivery. The spectrum of anomalies was classified based on organ systems, including the central nervous, cardiovascular, gastrointestinal, genitourinary, orofacial, musculoskeletal, and pulmonary systems. Congenital anomalies were classified by organ system and categorized as single or multiple. Data were analyzed descriptively using IBM SPSS Statistics version 25. Prevalence was

calculated as the proportion of cases among total pregnancies with 95% confidence intervals, and categorical variables were presented as frequencies and percentages.

RESULTS

Characteristics of Study Participants

A total of 717 pregnancies underwent obstetric ultrasonographic examination at the Fetomaternal Clinic of a tertiary referral center. Of these, 78 cases were identified as having intrauterine congenital anomalies, yielding a prevalence of 10.9% (95% CI: 8.6%–13.2%). The majority of mothers with congenital anomalies were in the 26–30 years age group (n=24, 30.77%), followed by those aged 31–35. The lowest frequency was observed in the 36–40 years age group (n=2, 2.56%). Most respondents had completed senior high school or equivalent (n=27, 34.62%), followed by those with a bachelor's degree (n=23, 29.49%). Based on residence, the majority of patients were from urban areas (n=51, 65.38%), while a smaller proportion in rural areas (n=27, 34.62%). Maternal demographic characteristics are presented in Table 1.

The majority of cases occurred in multigravida women (n=53, 67.95%) with singleton pregnancies (n=76, 97.44%). Congenital anomalies were most frequently detected during the second trimester (n=42, 53.85%). The most common risk factors identified were a history of abortion (n=11, 14.10%) and infections during pregnancy (n=7, 8.97%). Fetal viability was classified as lethal in 33 cases (42.31%) and non-lethal in 45 cases (57.69%). Most pregnancies were managed conservatively (n=42, 53.84%), and the predominant mode of delivery was cesarean section (n=57, 73.08%). Obstetric characteristics of the respondents are presented in Table 2.

Maternal Demographics	Frequency (n)	%
Maternal Age (Years)		
<16	4	5.13
16-20	5	6.41
21-25	17	21.79
26-30	24	30.77
31-35	21	26.92
36-40	2	2.56
>40	5	6.41
Maternal Education Level		
No formal education	1	1.28
Primary school	6	7.69
Junior high school	14	17.95
Senior high school	27	34.62
Diploma (D3)	6	7.69
Bachelor's degree (S1)	23	29.49
Master degree (S2)	1	1.28
Residence		
Urban	51	65.38
Rural	27	34.62

Tabel 1. Maternal demograp

Obstetric Profile	Frequency (n)	%
Parity		
Primigravida	25	32.05
Multigravida	53	67.95
Plurality		
Singleton	76	97.44
Multiple	2	2.56
Gestational Age at Diagnosis (weeks)		
First Trimester (0-12)	4	5.13
Second Trimester (13-28)	42	53.85
Third Trimester (29-42)	32	41.03
Risk Factors		
History of abortion	11	14.10
Metabolic disorders in pregnancy	4	5.13
Infection in pregnancy	7	8.97
Oligohydramnion	2	2.56
Polihydramnion	6	7.69
No risk factors	48	61.54
Fetal Prognosis		
Lethal	33	42.31
Non lethal	45	57.69
Pregnancy Outcome		
Termination	32	41.02
Conservative management	42	53.84
Abortion	1	1.28
IUFD	3	3.84
Mode of Delivery (MOD)		
Cesarean section	57	73.08
Vaginal delivery	21	26.92

Tabel 1. Profil Obstetri pada 78 Kasus
Kelainan Kongenital Intrauterin

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Spectrum of Congenital Anomalies by Organ System

Based on organ systems, central nervous system anomalies were the most frequently identified (n=24, 30.76%), followed by cardiovascular anomalies (n=16, 20.51%) and gastrointestinal anomalies (n=9, 11.53%). Genitourinary anomalies accounted for 4 cases (5.12%), while orofacial and musculoskeletal anomalies were each observed in 2 cases (2.56%). Pulmonary anomalies were the least common, with 1 case (1.28%). The majority of cases involved a single organ system (n=58, 74.35%). The complete distribution of the spectrum of congenital anomalies is presented in Table 3

Table 2. Spectrum of Single Congenital Anomalies by Organ System

Organ System	Congenital Anomaly	Frequency (n)	Percentage (%)
Singleton Organ System			
Central Nervous System	Hydrocephalus	5	30.76%
	Ventriculomegaly	5	
	Holoprosencephaly	3	
	Meningocele	1	
	Anencephaly	6	
	Mikrocephaly	2	
	Meningoencephalocele	1	
	Encephalocele	1	
cardiovaskular	VSD (Ventricular Septal Defect)	4	20.51%
	ASD (Atrial Septel Defect)	1	
	Hemangioma	2	
	Hydrops fetalis	3	

Organ System	Congenital Anomaly	Frequency (n)	Percentage (%)
	PAC (Premature Atrial Contraction)	1	
	Heart Block grade III	1	
	Pericardial Effusion	2	
	Ebstein anomaly	1	
	Cardiomegaly	1	
Gastrointestinal	Omphalocele	5	11.53%
	Duodenal atresia	3	
	Gastroschisis	1	
Genitourinary	Renal agenesis	3	5.12%
	Megavesica	1	
Orofacial	Cleft lip	1	2.56%
	Cleft palate	1	
Musculoskeletal	Achondroplasia	1	2.56%
	Phocomelia	1	
Pulmonary	Pulmonary hypoplasia	1	1.28%
Singleton organ system		58	74.35
Multiple organ system		20	25.64

DISCUSSION

Maternal sociodemographic factors and intrauterine congenital anomalies

Most congenital anomaly cases occurred in the 26–30 years age group ($n=24$, 30.77%), differing from reports in Nepal that showed higher incidence among adolescent pregnancies.²⁵ Studies from India reported the highest distribution in the 18–30 years age group, while population-based data from Iran and China also demonstrate an association between maternal age, congenital anomalies, and perinatal outcomes.^{29–33} These variations indicate that facility-based distributions often reflect the underlying reproductive age structure, and risk assessment requires comparison with total births across age groups.

Most respondents had completed senior high school ($n=27$, 34.62%). Lower educational attainment has been associated with a higher frequency of congenital anomalies, while higher maternal education correlates with reduced perinatal mortality due to birth defects.^{33,34} The predominance of secondary-level education in this study suggests that congenital anomalies are not limited to lower educational groups, highlighting the potential role of biological, genetic, and referral factors.

Most cases originated from urban areas, likely reflecting better access to ultrasonography and tertiary referral systems. Studies from Iran and Hunan have shown that residence is associated with mortality due to congenital anomalies, with higher perinatal mortality reported in rural populations ($OR=1.69$ and $OR=2.03$, respectively).^{31,33} Rural residence has also been linked to consanguinity, which may increase the risk of congenital anomalies.^{34–36} These findings suggest that, in hospital-based studies, geographic distribution primarily reflects healthcare access and referral patterns rather than true population-level biological risk.

Association between obstetric profile and intrauterine congenital anomalies

Based on the findings of this study, the majority of cases occurred in multigravida women ($n=53$, 67.95%). This finding is consistent with hospital-based studies from India, which reported a higher proportion of cases among multigravida compared to primigravida women.^{26,32} Data from Iran have also identified parity greater than two as an independent risk factor for congenital anomalies.²⁹ However, several studies have reported a higher distribution of cases among women with low to moderate parity.^{29,30} These variations suggest that parity distribution in facility-based studies is strongly influenced by the local obstetric population structure. Therefore, interpretation of risk requires comparison with the total number of births stratified by parity.

The majority of pregnancies were singleton ($n=76$, 97.44%). Multifetal pregnancies have been associated with a higher risk of morbidity and congenital anomalies, particularly in monochorionic complications.³³ Studies from India have also identified multifetal pregnancy as a frequently discussed risk factor.³² In the context of this study, plurality does not appear to be a major contributor to the distribution of congenital anomaly cases.

The highest rate of detection was observed in the second trimester ($n=42$, 53.85%). This finding is consistent with the established concept that the second-trimester anatomical survey represents the optimal window for detection, with most anomalies identified before 24 weeks of gestation.^{27,29,35,36} Previous studies have reported that congenital anomalies contribute to 42.0% of perinatal mortality, with 96.16% of cases resulting in selective termination.³² The predominance of second-trimester detection in this study may reflect the antenatal care system in Indonesia, where ultrasonographic examination by specialists is commonly recommended during the second trimester. This practice likely increases the probability of detecting congenital anomalies at this gestational age.

The most frequently identified risk factor in this study was a history of abortion ($n=11$, 14.10%). A history of spontaneous abortion in previous pregnancies has been associated with fetal chromosomal abnormalities.⁴⁰ Infection during pregnancy was the second most commonly identified risk factor in this study, which is consistent with the established concept that maternal infections can increase the risk of congenital anomalies.^{40,42} However, screening for infections particularly TORCH infections is not routinely performed in Indonesia, largely due to its relatively high cost.

The majority of deliveries were performed via cesarean section ($n=57$, 73.08%). This finding is consistent with previous studies demonstrating that congenital anomalies are associated with higher rates of cesarean delivery, as well as increased neonatal morbidity and adverse outcomes at birth.⁴¹ A study from Iran reported that a history of cesarean section was associated with mortality due to congenital anomalies (OR 2.13), likely reflecting high-risk pregnancies.³³ In this study, fetal viability was classified as lethal in 33 cases (42.31%) and non-lethal in 45 cases (57.69%). All lethal cases were planned for termination; however, one case resulted in spontaneous abortion prior to the procedure. Previous studies have shown that congenital anomalies encompass a wide spectrum of severity, ranging from mild conditions to lethal abnormalities. The greater the

severity of the anomaly, the higher the risk of perinatal and neonatal mortality. Lethal congenital anomalies are known to contribute significantly to infant mortality, both in utero and during the early neonatal period^{38,44}

Spectrum of intrauterine congenital anomalies by organ system

Based on organ systems, central nervous system anomalies were the most frequently identified (n=24, 30.76%), followed by cardiovascular (n=16, 20.51%) and gastrointestinal anomalies (n=9, 11.53%). This pattern is consistent with reports from India, which demonstrated a predominance of nervous system anomalies (43%) and circulatory system anomalies (21.7%).²⁹ Another study similarly reported that central nervous system anomalies (33.72%) were the most common, followed by cardiovascular and gastrointestinal anomalies.⁹ In contrast, other studies from India have reported gastrointestinal anomalies (28.3%) as the most dominant, followed by musculoskeletal (24.53%) and central nervous system anomalies (20.13%), with a lower proportion of cardiovascular anomalies (6.29%).³⁷ A predominance of gastrointestinal anomalies has also been reported (43.9%), with significant contributions from central nervous, musculoskeletal, chromosomal, and cardiovascular systems.²⁶ Additionally, pulmonary, genetic, and genitourinary anomalies have been reported to have relatively low prevalence (<10 per 10,000 births).³⁸ Based on organ involvement, the majority of cases in this study were limited to a single organ system (n=58, 74.35%), while multisystem involvement was observed in 20 cases (25.64%). This pattern is comparable to findings from India (74.4%) and Iran (82.35%), whereas studies from China reported 57.8% single-system involvement and 40.6% multiple congenital anomalies.^{26,31,33}

Overall, the spectrum of anomalies in this study was predominantly composed of central nervous system and cardiovascular anomalies, with most cases involving a single organ system. This pattern is consistent with reports from tertiary referral centers, although it may vary depending on epidemiological context, registry systems, and healthcare service characteristics across different countries. The prevalence of congenital anomalies in this study, calculated using the proportion formula, was 10.9% (95% CI: 8.6%–13.2%), which is higher than regional estimates.

This study has several limitations that should be considered when interpreting the findings. First, as a hospital-based study, the results may not fully represent the national population. In addition, the study was conducted in a tertiary referral center, which may lead to a higher observed prevalence of congenital anomalies compared to the general population due to referral bias.

Another limitation is that the identification of certain risk factors, such as genetic abnormalities and specific infections, is not routinely performed, primarily due to the relatively high cost and limited coverage by the national health insurance system. Furthermore, the analysis of associations between individual risk factors and the occurrence of congenital anomalies could not be conducted optimally.

CONCLUSION AND RECOMMENDATIONS

The prevalence of intrauterine congenital anomalies detected by ultrasonography at this tertiary referral center was 10.9% (95% CI: 8.6%–13.2%). The anomalies were predominantly related to the central nervous and cardiovascular systems, with most cases involving a single organ system and detected during the second trimester.

The relatively high prevalence observed in this study is likely influenced by referral bias, as tertiary hospitals tend to receive a higher proportion of high-risk pregnancies and severe cases. Strengthening ultrasonographic screening—particularly during the second trimester improving antenatal care coverage, and optimizing folic acid supplementation are essential strategies. Furthermore, the development of population-based congenital anomaly registries is crucial to obtain more representative estimates and to reduce referral bias.

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