Research Article

Feasibility Study of Integrated Service for Congenital Abnormalities in A Low-Middle Income Country

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Abstract

Objective: To explore the occurrence of congenital anomalies identified at Dr. Cipto Mangunkusumo Hospital, a leading tertiary referral center in Indonesia. The findings emphasize the necessity for comprehensive and integrated healthcare services to improve the management and outcomes for affected newborns.

Methods: Data were retrospectively analyzed from a one-year period of prenatal screening at Dr. Cipto Mangunkusumo General Hospital, comprising 323 patients. The analysis included variables such as maternal age, date of birth, obstetric history, gestational age, and detected fetal anomalies.

Results: Pregnant women aged 30–39 years accounted for the majority of congenital anomaly cases. Most abnormalities were detected during the second trimester (13–28 weeks of gestation). Central nervous system defects were the most common (159 cases), followed by cardiovascular abnormalities (130 cases). In total, 607 anomalies were recorded in 494 patients during the screening period.

Conclusion: This research reveals the frequency of congenital anomalies detected over a year at Dr. Cipto Mangunkusumo Hospital. The findings underscore the importance of timely prenatal diagnostics and well-structured perinatal care, particularly in high-level referral hospitals. Strengthening early screening and intervention efforts may enhance health outcomes for both mothers and infants.

Keywords: birth defects, congenital anomalies, prenatal diagnostics, referral hospital.

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INTRODUCTION

Congenital abnormalities refer to structural or functional anomalies that originate during intrauterine development and are present at birth. These abnormalities are one of the leading contributors to high infant mortality rates in Indonesia.[1] The country's infant mortality rate is estimated at 32 deaths per 1,000 live births, while the neonatal mortality rate is approximately 19 per 1,000 live births. According to the World Health Organization (WHO) South-East Asia Region, the prevalence of congenital anomalies in Indonesia is around 59.3 per 1,000 live births. With an estimated 5 million births annually, this equates to roughly 295,000 cases of congenital abnormalities each year [2,3] In addition to causing neonatal deaths, these conditions are also significant contributors to stillbirths and spontaneous abortions. For infants who survive, congenital abnormalities often result in long-term disabilities or chronic health issues. Therefore, early detection through integrated medical interventions is essential to minimize infant morbidity and mortality.[4]

The causes of congenital abnormalities are

multifactorial and may include genetic disorders, infections, and environmental exposures. Preventive measures such as vaccination, sufficient intake of folic acid and iodine, avoiding teratogenic substances like alcohol, and promoting healthy maternal behaviors can significantly reduce the risk. Several maternal risk factors have been identified, including alcohol use, exposure to tobacco smoke, high parity, advanced maternal age, medication use during pregnancy, consanguinity, a history of children or relatives with congenital anomalies, low socioeconomic status, gestational diabetes, and prior pregnancy loss.[5]

At Dr. Cipto Mangunkusumo Hospital, a national tertiary referral center, congenital and fetal anomalies are observed across nearly all organ systems. Each month, dozens to hundreds of new fetal anomaly cases are detected among pregnant women attending the fetomaternal outpatient clinic for prenatal screening. This study aims to investigate the range and frequency of congenital abnormalities diagnosed at Dr. Cipto Mangunkusumo General Hospital and to emphasize the importance of integrated healthcare from early detection to condition-specific management. Fetomaternal ultrasound serves as a critical tool in identifying anomalies during the prenatal period. Routine ultrasound examinations during pregnancy have been shown to reduce neonatal mortality by enabling the early detection of life-threatening anomalies, particularly congenital heart defects. In addition, postnatal screening plays a vital role in diagnosing abnormalities that may not be apparent during prenatal assessment.[6]

However, in Indonesia, the practice of neonatal screening continues to face significant challenges, including limited epidemiological data, ethical considerations, low public awareness and family involvement, a shortage of trained healthcare professionals, and the absence of standardized screening protocols and formal training for medical staff.[7] The treatment of congenital abnormalities depends on the specific condition and may involve pharmacological interventions such as corticosteroids—or surgical approaches, including procedures like spina bifida repair.[1]

This research aims to provide a comprehensive overview of the prevalence and variety of congenital abnormalities identified over a one-year period at Dr. Cipto Mangunkusumo General Hospital. By mapping the burden and characteristics of these conditions, this study seeks to support the advancement of well-integrated, multidisciplinary healthcare services for their effective management. Furthermore, this study also functions as a preliminary evaluation of the feasibility of implementing a structured national program for managing birth defects, taking into account the clinical, logistical, and ethical aspects of service delivery. This dual objective is intended to contribute to future policies and practices aimed at improving outcomes for neonates affected by congenital abnormalities and supporting their families.

METHODS

This retrospective cohort study utilized data from 323 pregnant women treated at Dr. Cipto Mangunkusumo General Hospital over a one-year period, from March 2023 to March 2024. Information was sourced from electronic medical records of patients attending the outpatient fetomaternal and high-risk pregnancy clinics. The study aimed to describe the prevalence and types of congenital abnormalities diagnosed during pregnancy. These abnormalities were identified during the first, second, or third trimester through fetomaternal ultrasonography and categorized based on the affected system, including the central nervous system, facial structures, thorax, heart, abdomen, extremities, genitals, syndromes, and others. Maternal characteristics such as age and gestational age were also analyzed. To ensure data reliability and relevance, specific inclusion and exclusion criteria were applied during sample selection.

Subject Criteria

Inclusion Criteria

Pregnant women aged 18 years or older who were diagnosed with fetal congenital abnormalities at Dr. Cipto Mangunkusumo General Hospital between March 2023 and March 2024, consented to the use of their medical data, and had their data recorded during their first clinic visit.

Exclusion Criteria

Patients under 18 years of age; data collected from follow-up visits instead of the initial consultation; duplicate entries; cases without fetal congenital abnormalities; patients treated for placenta accreta spectrum (PAS) before July 2020 or after February 2024; records not approved for research use; and any repeated datasets.

Sample Size Determination
This study employed a total sampling approach to obtain a comprehensive dataset. All obstetric patients who met the inclusion criteria and were examined at the Fetomaternal Clinic between March 2023 and March 2024 were included in the analysis.

Procedures and Data Collection
Relevant clinical data were extracted from the hospital's electronic medical record system, focusing on patients who underwent fetomaternal ultrasonography during their initial clinic visit. All ultrasound assessments were part of routine clinical diagnostics at Dr. Cipto Mangunkusumo General Hospital. Only first-visit data were analyzed to prevent duplication and minimize confounding from follow-up examinations. The data were compiled into a structured database for analysis.

Study Parameters
This study applied a descriptive-analytic method using secondary data obtained from pregnant patients with diagnosed fetal congenital anomalies. Only first-visit data were considered, with emphasis on maternal age, gestational age at diagnosis, and the type of abnormality detected.

Data Analysis and Statistical Methods
This study was conducted following ethical approval from the Ethics Committee of the Faculty of Medicine, Universitas Indonesia, and Dr. Cipto Mangunkusumo Hospital. All data were organized into tables and analyzed using SPSS software. Descriptive statistics were used to present the frequency and distribution of the abnormalities, and additional analytical methods were applied to identify associations between clinical variables.

Ethical Approval
This study utilized retrospective data obtained from medical records at Dr. Cipto Mangunkusumo General Hospital. A total sampling technique was applied for all eligible patients who underwent prenatal screening at the fetomaternal outpatient clinic between March 2023 and March 2024. The study received ethical clearance from the Ethics Committee of the Faculty of Medicine, Universitas Indonesia, and RSCM. All data collection was conducted in accordance with ethical guidelines. Patient confidentiality was strictly maintained, with all information stored securely and inaccessible to unauthorized individuals. No personal identifiers were included or published in this study.

RESULTS

Table 1. Maternal Age and Gestational Age Distributions

Variable	n	(%)
Age		
≥ 18	131	40.6
≥ 30	154	47.7
≥ 40	38	11.8
Gestational age		
Trimester 1	131	40.6
Trimester 2	154	47.7
Trimester 3	38	11.8

The data presented in Table 1 are derived from inpatients at the Fetomaternal and High-Risk Pregnancy Clinic of Dr. Cipto Mangunkusumo General Hospital. This table highlights the distribution of maternal and gestational age among the study participants. Maternal age was categorized into three groups, based on existing literature indicating an increased risk of congenital abnormalities beginning at age 30, with a significantly higher risk observed in mothers aged 40 and above.[8]

In this study, the majority of congenital abnormalities were observed in the 30–39 years age group, which accounted for 47.7% of cases. This was followed by the 18–29 years group with 40.6%, and lastly, mothers aged 40 years and above, comprising 11.8% of the total cases. Regarding gestational age, the highest proportion of congenital anomalies was detected during the second trimester, representing 47.7% of the cases.

In total, 607 congenital abnormalities were identified among 494 patients, indicating that some patients had multiple anomalies. Table 2 shows the distribution of these anomalies according to the affected anatomical system, presented both by total number of cases and number of affected patients. The most commonly affected system was the central nervous system (CNS), which accounted for 26.2% of all cases and was present in 26.7% of the patients. Within this category, CNS anomalies were the most frequent (26.2% of cases, involving 132 patients), followed by facial anomalies, which represented 9.4% of cases. Cardiovascular anomalies were the second most prevalent group, making up 21.4% of all

cases and affecting 16.8% of the patients. This highlights the significant clinical burden posed by congenital heart defects in the study population.

Table 2. Congenital Abnormalities Distribution of Total Patient and Cases

Variable		Total Cases	(%)	Total Patient	(%)
Head and Neurological System	CNS	159	26.2	132	26.7
	Facial Structure	57	9.4	44	8.9
Cardiorespiratory System	Respiratory	9	1.5	8	1.6
	Cardiovascular	130	21.4	83	16.8
Gastrointestinal System	Abdominal	53	8.7	51	10.3
	Urogenital	52	8.6	46	9.3
Syndrome	Syndrome	22	3.6	22	4.5
Others	Extremities	41	6.8	38	7.7
	Others	84	13.8	70	14.2
TOTAL		607	100	494	100

Abdominal and urogenital anomalies were found in relatively similar proportions, contributing 8.7% and 8.6% of the total identified cases, respectively. Congenital abnormalities involving the extremities and syndromic presentations were also significant, accounting for 6.8% and 3.6% of all cases.

The "syndrome" category refers to a set of congenital anomalies that follow a distinct and recognizable pattern, often arising from a shared underlying cause be it genetic, environmental, or pathophysiological in nature.[9,10] This includes well-documented syndromes such as chromosomal or genetic disorders, as well as specific fetal conditions with syndromic features, such as Potter sequence, which is commonly associated with severe oligohydramnios and bilateral renal agenesis. Another example is twin-to-twin transfusion syndrome (TTTS), typically occurring in monochorionic twins, where the Quintero staging system is used to assess the severity of intertwin blood flow imbalance.[9,10] These syndromes are categorized separately from isolated structural defects due to their multi-system involvement and complex pathophysiology.

The "Others" category encompassed a variety of anomalies not classified under the primary anatomical systems and represented 13.8% of total cases, affecting 14.2% of patients. This reflects the diversity and diagnostic challenges associated with congenital disorders that do not fit neatly into conventional classifications.

Overall, the findings point to a notably high prevalence of central nervous system and cardiovascular anomalies within the study population, which is consistent with global epidemiological trends. These results emphasize the critical need for early prenatal screening and coordinated multidisciplinary care, especially within tertiary referral centers, to improve detection and outcomes in cases of congenital abnormalities.

DISCUSSION

Integrated maternal-fetal care is crucial for improving health outcomes in pregnancies complicated by congenital anomalies. A comprehensive approach comprising systematic prenatal screening, precise diagnosis, coordinated multidisciplinary management, timely therapeutic interventions, and close perinatal monitoring plays a key role in reducing perinatal morbidity and mortality.[11] Prenatal ultrasonography remains the cornerstone for detecting fetal anomalies, enabling early clinical decision-making and timely interventions that contribute to better neonatal survival and long-term prognosis. It is estimated that over 50% of congenital anomalies can be diagnosed prenatally, and early surgical treatment has been shown to significantly reduce complications and improve outcomes.[11]

Technological advancements in both prenatal imaging and fetal surgery have greatly improved the ability to diagnose and treat structural anomalies in utero. In specific cases, such as fetal myelomeningocele or lower urinary tract obstruction (LUTO), intrauterine surgical

interventions have demonstrated superior neonatal and neurodevelopmental outcomes when compared to postnatal treatment.[12,13] These procedures yield the best results when performed in specialized tertiary care centers that have access to multidisciplinary teams, including fetal medicine specialists and pediatric surgeons highlighting the importance of centralized and coordinated care models.[12,13]

Despite these advancements, notable challenges remain. There is a significant lack of national-level data on the incidence and clinical outcomes of specific congenital anomalies, which hinders the development of evidence-based healthcare policies.[7] Additional barriers include ethical concerns regarding fetal interventions, limited awareness among families, shortages of specialized perinatal care providers, and insufficient formal training programs for healthcare professionals.[1]

Treatment options for congenital anomalies range from medical therapies to complex surgical interventions. For instance, obstructive uropathies such as posterior urethral valves or congenital renal tumours often require early neonatal surgical correction.[14] Central nervous system anomalies, including hydrocephalus, encephalocele, and myelomeningocele, are typically addressed through neurosurgical intervention, either prenatally or postnatally, depending on the severity and feasibility of the procedure.[15]

Similarly, thoracic and gastrointestinal anomalies such as Congenital Cystic Adenomatoid Malformation (CCAM), Congenital Diaphragmatic Hernia (CDH), and intestinal atresias generally require a series of surgical procedures and specialized perioperative care.[16] Outcomes for congenital heart defects, including transposition of the great arteries and ventricular septal defects, have improved significantly due to advances in surgical techniques, such as the arterial switch operation and septal defect repair.[17]

In addition to surgical management, non-invasive and medical therapies also play essential roles. Antenatal corticosteroids, for example, are commonly used to accelerate fetal lung maturation in cases of CDH, while postnatal support such as mechanical ventilation or Extracorporeal Membrane Oxygenation (ECMO) is often necessary for critically ill newborns.[18] In less severe conditions such as mild hydronephrosis, conservative management including regular monitoring and prophylactic antibiotics may be adequate.[19] On the other hand, for neonates with non-operable or syndromic heart defects, treatment often focuses on medical stabilization, such as prostaglandin infusion, and palliative care.[20]

Despite best efforts, certain congenital anomalies remain lethal or incompatible with life. Conditions such as anencephaly, bilateral renal agenesis, and severe chromosomal disorders like trisomy 13 or trisomy 18 often require a palliative, family-centered approach, focusing on quality of life and compassionate end-of-life care.[21]

Strengths and Limitations of the Study

This study benefits from a large, diverse patient population drawn from a national referral hospital, which allows for in-depth analysis of congenital anomalies across various organ systems. The involvement of multiple specialties also enhances the clinical relevance and applicability of the findings. However, several limitations should be acknowledged. The retrospective nature of the study restricts the ability to draw causal conclusions. Furthermore, long-term follow-up data were not available for many cases, limiting the assessment of neurodevelopmental or health outcomes beyond the perinatal period. Lastly, the absence of national prevalence data for specific congenital conditions highlights the urgent need for robust, systematic surveillance programs and birth defect registries in Indonesia.

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CONFLICT of INTEREST

Authors declare that they have no conflict of interests.

CONCLUSION

Amid continuing efforts to improve outcomes for neonates with congenital anomalies, there is an increasing need to align clinical management with evidence-based practices that consider

both feasibility and long-term impact. A clear understanding of the prevalence and types of congenital abnormalities is critical for enhancing neonatal care strategies.

At Dr. Cipto Mangunkusumo General Hospital, central nervous system anomalies were the most frequently identified (26.2%), followed by cardiovascular defects (21.4%), as well as gastrointestinal and urogenital abnormalities, each accounting for approximately 8–9% of cases. These findings reflect the diverse and complex nature of cases seen at a tertiary referral institution and emphasize the importance of an integrated, multidisciplinary approach one that carefully weighs the practicality of intervention with the potential for favorable long-term outcomes.

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