



Pattern of Congenital Anomalies in Pediatric Patients at dr. Doris Sylvanus General Regional Hospital: A One-Year Retrospective Study

¹ Rayma Hayati, ¹ Yhohan Thaihutu

¹ Paediatric Surgery Division, Department of Surgery, dr. Doris Sylvanus General Regional Hospital, Palangka Raya, Indonesia.

Correspondence : raymahayati@gmail.com

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ABSTRACT

Background : Congenital anomalies (CA) require early treatment and impair pediatric care. Knowing demographic group incidence, classification, and distribution of inherent anomalies helps treatment and resource allocation. **Aims :** This study examines forms and demographics of CA in children treated at dr. Doris Sylvanus Regional Hospital for a year. **Methods :** A retrospective study was carried out by analyzing 320 patient files between July 2023-June 2024 at dr. Doris Sylvanus General Regional Hospital. The classification system ICD-10 was used for the evaluation of frequency, gender, and geographical distribution of CA. **Results :** The obtained analysis revealed a higher incidence of CA in males than in females: 208 (65%) and 112 (35%), respectively. There were 72 (22.5%) cases of CA in the genital organs, the highest number found, followed by the circulatory system with 67 (20.9%) and digestive system with 44 (13.75%) cases. Most patients are coming from the city of Palangka Raya, where 197 cases were reported (61.56%), followed by Gunung Mas with 28 cases (8.75%) and East Kotawaringin with 20 cases (6.25%). Several diagnoses are often made, such as Down syndrome, Hirschsprung disease, and congenital heart malformation. **Discussion :** The findings provide evidence of the need for specific health interventions in regions that seem to have higher rates of CA, including the city of Palangka Raya. The gender bias found in this research is supported, and its presence is to be further deeply probed. Timely detection of focused therapeutic management is critical in getting better outcomes among children born with CA. **Conclusion :** The analysis indicated a significant prevalence of abnormalities in the genital and circulatory systems, highlighting the urgent requirement for specialized health services to address these issues at an earlier stage in life. The prevalence of congenital abnormalities is greater in males and in urban regions like Palangka Raya, where access to health services is more prevalent. This disparity in access to health services between urban and rural areas is prevalent globally and significantly hinders the attainment of universal health coverage across all regions of Indonesia. This study's findings align with a broader investigation in the area, indicating that disparities in access to health services and health outcomes are influenced by socioeconomic, geographic, and demographic factors.

Keywords: Congenital Anomalies (CA), Pediatric Care, ICD-10 Classification, Gender Distribution, Regional Incidence

INTRODUCTION

Congenital anomalies (CA), often known as birth defects, are structural or functional abnormalities that appear at birth and have a significant impact on newborn and pediatric health. These anomalies can be induced by genetics, the environment, or a combination of the two, and they can have serious consequences for many organ systems, raising the risk of illness, disability, and death. CA are defined by the World Health Organization (WHO) as structural or functional problems that develop during intrauterine life and can be identified prenatally, at birth, or later in infancy. According to WHO figures from 2016, congenital abnormalities kill over 303,000 newborns worldwide each year. Congenital abnormalities affect roughly 3% of all live births. In 2015, congenital malformations accounted for 20% of all infant mortality in the United States and have subsequently emerged as the primary cause of baby death, following a decrease in other more common causes of mortality, including preterm birth, low birth weight, bacterial sepsis, and neonatal respiratory distress.(1,2) According to a meta-analysis of 25 studies from nine different countries, the estimated prevalence of CA is 20–21 per 1,000 live births. The highest prevalence, 43 per 1,000 people (95% CI: 14.89, 71.10), was found in southern Africa, according to the meta-analysis. Down syndrome had the lowest frequency at 0.62 per 1,000 (95% CI: 0.40, 0.84), while musculoskeletal system disorders were the most common among all types, occurring at a rate of 3.90 per 1,000 (95% CI: 3.11, 4.70). (Adane et al. 19)(3)

The prevalence of CA exhibits significant regional diversity throughout Indonesia. A study conducted at RSUP Sanglah Denpasar revealed that 19.3% of neonates were born with CA. The digestive system experienced the most significant impact, representing 40.1% of all cases, succeeded by chromosomal disorders at 11.5% and nervous system anomalies at 10.2%. A comprehensive study conducted at RSUP Dr. Mohammad Hoesin Palembang over a span of three years revealed a total of 432 instances of CA. The digestive system emerged as the most commonly affected, representing 60.2% of the cases, while the circulatory system accounted for 11.1%, and the musculoskeletal system comprised 10.4%. This study underscored the significance of possessing region-specific data to effectively guide healthcare policy formulation. Localized studies demonstrate variations in the types and prevalence of CA. Southern Africa exhibits the highest occurrence rates, while detailed studies in Indonesia reveal significant regional variations in the effects on organ systems, with certain areas displaying

comparatively higher frequencies of specific anomalies. (4,5) This inconsistency indicates potential influences from local environmental factors, access to medical care, and regional genetics that may alter the identification and prevalence of such anomalies. While these facts are valuable, the understanding of CA across various regions, particularly those with inadequate healthcare and reporting systems, is remarkably lacking.

In Indonesia, particularly in less urbanized areas such as Central Kalimantan, there exists a notable lack of data regarding CA. Dr. Doris Sylvanus General Regional Hospital, a prominent referral institution in Central Kalimantan, is essential in pediatric healthcare, particularly in the treatment of CA. The lack of specific local data obstructs the appropriate evaluation of these illnesses, creating obstacles for healthcare planning, early diagnosis, and intervention. This study is to examine the prevalence of CA in pediatric patients admitted to dr. Doris Sylvanus General Regional Hospital over the course of one year. This research aims to identify the organ systems most frequently affected and the regions where they are most commonly found, thus addressing information gaps and offering significant epidemiological insights. This data is essential for improving medical prevention strategies, formulating public health measures, and reducing the impact of diseases in specific areas. This research will also provide a basis for further research and policy formulation, promoting better health care outcomes for children affected by CA.

METHODS

This research, conducted via a retrospective analysis at dr. Doris Sylvanus General Regional Hospital in Central Kalimantan, examined CA in pediatric patients. From July 2023 to June 2024, a review of medical records for 320 pediatric patients diagnosed with CAs was conducted to ascertain prevalence and patterns. The records were classified according to the International Classification of Diseases, Tenth Revision (ICD-10), encompassing a range from nervous system malformations to chromosomal abnormalities. Demographic data, including gender, and geographical origin, were collected from various districts of Central Kalimantan. The data were analyzed descriptively and expressed as percentages, subsequently presented in tables and figures to illustrate the frequency and distribution of anomalies, gender differences, and geographical trends. The study adhered to ethical standards, receiving Hospital Ethics Committee approval and ensuring strict confidentiality of patient data. This provided essential insights into the influence of CAs on regional pediatric healthcare, informing future healthcare

RESULTS

This retrospective analysis was performed at dr. Doris Sylvanus General Regional Hospital from July 2023 to June 2024. The research examined a group of 320 pediatric identified with various congenital abnormalities. This study clarifies the distribution of abnormalities about their classification, gender, and geographic location.

Congenital Anomalies Distribution

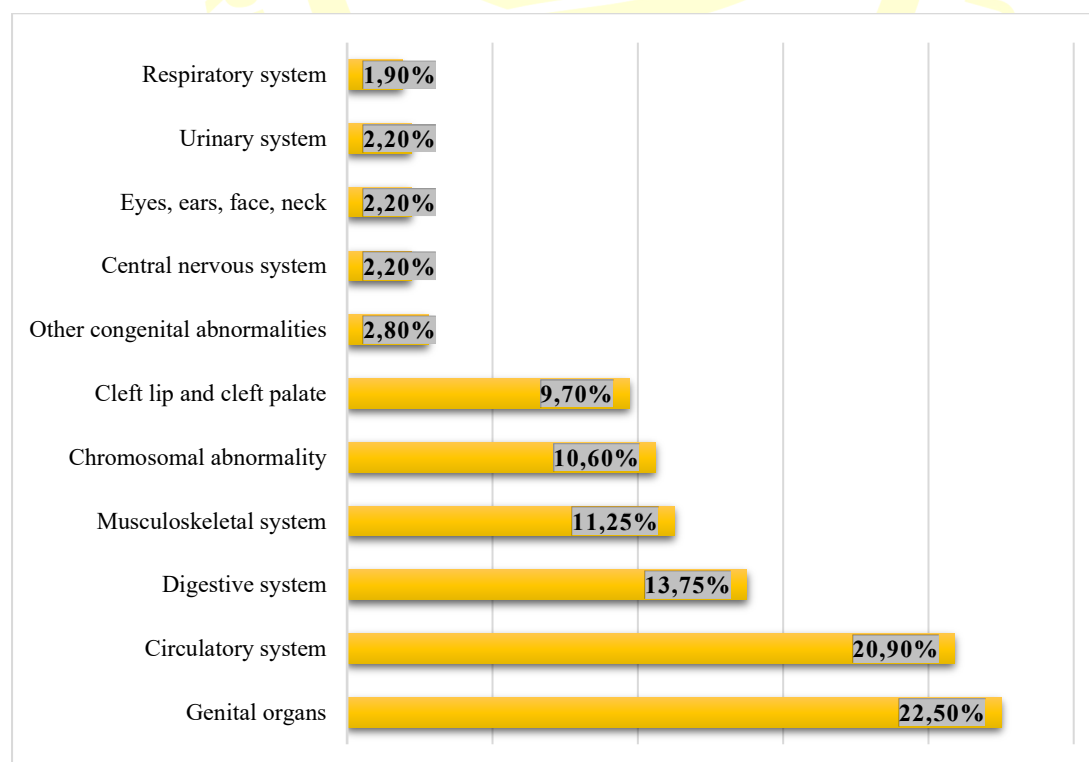
The following table discusses frequency of types of congenital abnormalities in infants and children at dr. Doris Sylvanus General Regional Hospital in the period July 2023-June 2024. During the period from July 2023 to June 2024 at dr. Doris Sylvanus General Regional Hospital, the prevalence of CA was notably highest in the genital organs, accounting for 22.5% with a total of 72 cases. This was closely followed by abnormalities in the circulatory system, which represented 20.9% with 67 cases, and those in the digestive system, which made up 13.75% with 44 cases. Malformations of the musculoskeletal system, including conditions like clubfoot, were observed in 11.25% of the cases, totaling 36 instances. Meanwhile, chromosomal abnormalities not classified elsewhere, such as Down syndrome, represented 10.6% with 34 cases recorded. Cleft lip and palate were identified in 9.7% of the subjects, totaling 31 cases. Other congenital malformations were observed less frequently, accounting for 2.8% with 9 cases. Malformations of the nervous system, eyes, ears, face, and neck, as well as the urinary system, each represented 2.2% with 7 cases respectively. The least frequent were respiratory system malformations, noted at 1.9% with 6 cases. This distribution illustrates the variety and prevalence of congenital conditions addressed at the hospital throughout the year.

Table 1 Distribution of Subjects According to Type of Congenital Abnormalities

ICD-10 Classification	Number (n)	Percentage (%)
Q00-Q07 Congenital malformations of the nervous system	7	2,2%
Q10-Q18 Congenital malformations of the eyes, ears, face, and neck	7	2,2%

Q20-Q28 Congenital malformations of the circulatory system	67	20,9%
Q30-Q34 Congenital malformations of the respiratory system	6	1,9%
Q35-Q37 Cleft lip and cleft palate	31	9,7%
Q38-Q45 Congenital malformations of the digestive system	44	13,75%
Q50-Q56 Congenital malformations of genital organs	72	22,5%
Q60-Q64 Congenital malformations of the urinary system	7	2,2%
Q65-Q79 Congenital malformations and deformations of the musculoskeletal system	36	11,25%
Q80-Q89 Other congenital malformations	9	2,8%
Q90-Q99 Chromosomal abnormality, not elsewhere classified	34	10,6%
Total	320	100%

Figure 1 Distribution of Congenital Anomalies based on the ICD-10 system



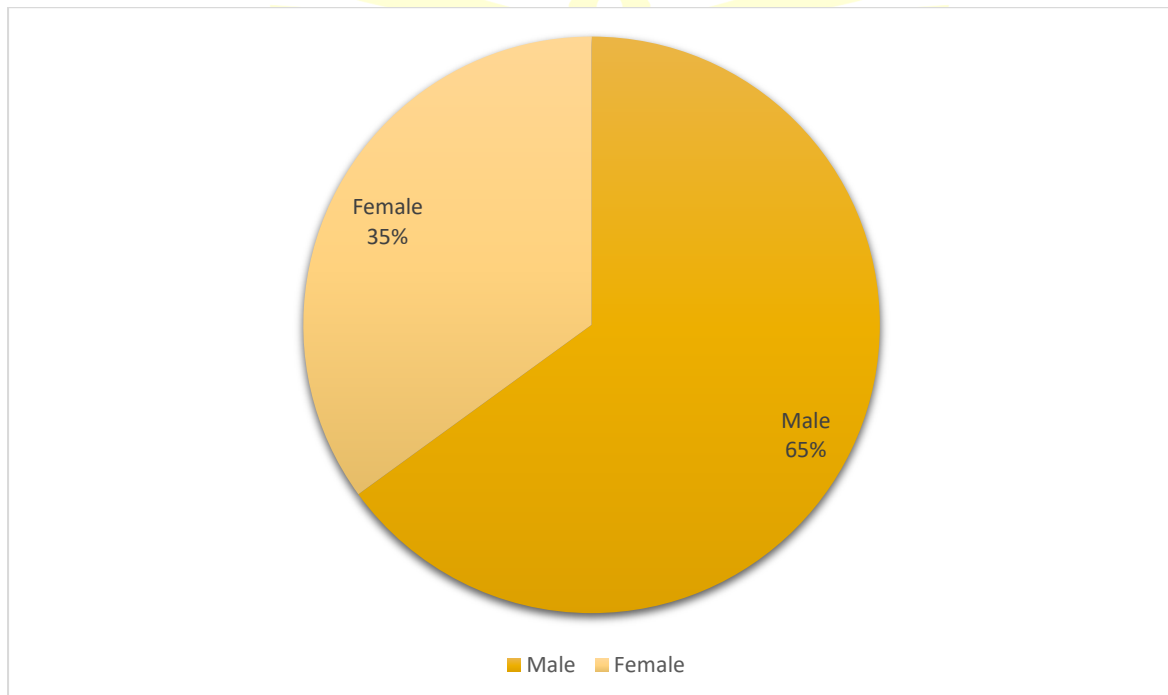
Gender Distribution

The analysis showed a higher prevalence of CA in male patients, with 208 cases (65%), compared to 112 cases (35%) in female patients.

Table 2 Distribution of Subjects by Gender

Gender	Number (n)	Percentage (%)
Male	208	65%
Female	112	35%
Total	320	100%

Figure 2 Distribution of Patients by Gender



Geographic Distribution

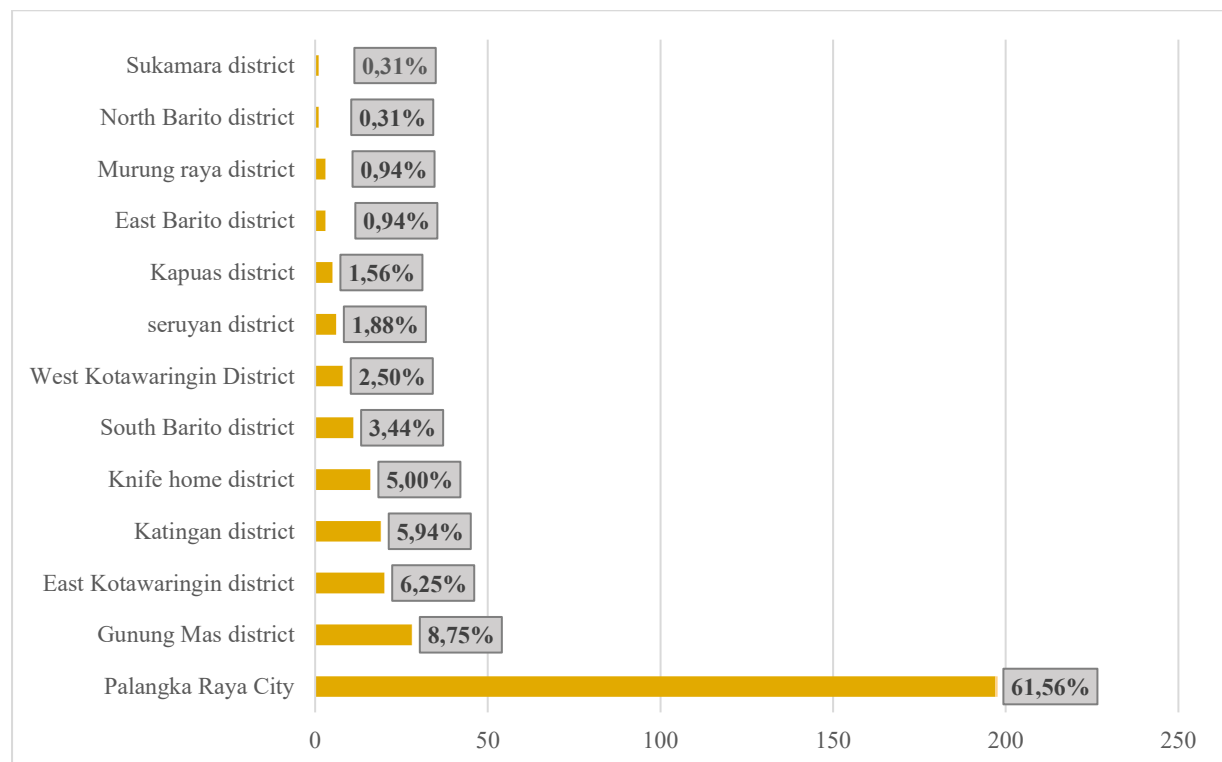
During the geographical analysis of CA at dr. Doris Sylvanus General Regional Hospital from July 2023 to June 2024, Palangka Raya City exhibited the highest incidence, accounting for 197 cases (61.56%), signifying a substantial regional concentration. Gunung Mas District recorded 28 cases (8.75%), while East Kotawaringin District reported 20 cases (6.25%). Katingan District had 19 cases (5.94%), Pulang Pisau District 16 cases (5.00%), and

South Barito District 11 cases (3.44%). West Kotawaringin District reported 8 cases (2.50%), Seruyan District 6 cases (1.88%), Kapuas District 5 cases (1.56%), while both East Barito and Murung Raya Districts each recorded 3 cases (0.94%). The lowest number of instances occurred in North Barito and Sukamara Districts, each reporting only 1 case (0.31%). This pattern underscores the considerable heterogeneity in the prevalence of congenital abnormalities among various districts, with a pronounced majority in the center metropolitan area of Palangka Raya.

Table 3 Distribution of Subjects by District

No	District/City	Number (n)	Percentage (%)
1	Palangka Raya City	197	61,56%
2	Gunung Mas district	28	8,75%
3	East Kotawaringin district	20	6,25%
4	Katingan district	19	5,94%
5	Pulang Pisau district	16	5,00%
6	South Barito district	11	3,44%
7	West Kotawaringin District	8	2,50%
8	Seruyan district	6	1,88%
9	Kapuas district	5	1,56%
10	East Barito district	3	0,94%
11	Murung raya district	3	0,94%
12	North Barito district	1	0,31%
13	Sukamara district	1	0,31%

Figure 3 Distribution of Subjects by District



DISCUSSION

Unveiling Patterns: The Predominance of Genital and Circulatory Anomalies

The retrospective study conducted at dr. Doris Sylvanus General Regional Hospital from July 2023 to June 2024 revealed a high prevalence of CA, with genital and circulatory system abnormalities being the most dominant findings. The most common abnormalities among the 320 pediatric patients examined were sexual organ anomalies, which made up 22.5% (72 instances) of the total. These were closely followed by circulatory system anomalies (20.9%; 67 cases) and digestive system anomalies (13.75%; 44 cases). This high prevalence suggests a need for further investigation into potential genetic and environmental factors contributing to their occurrence. The findings align with previous studies in Indonesia and globally, which report similar trends. For instance, research at Kathmandu Medical College identified genitourinary anomalies as the most common (24.2%), followed by musculoskeletal (21.2%) and circulatory system anomalies (18.2%). Similarly, a study in Vietnam found congenital heart disease to be the predominant defect, accounting for 52.3% of cases, with a prevalence rate of 200.9 per 10,000 live births, followed by musculoskeletal and digestive

system irregularities. Additionally, a study conducted at Gulf Medical College Hospital and Research Center reported genitourinary and musculoskeletal anomalies as the most frequently affected systems. (6,7)

In contrast, a study at RSUP Sanglah Denpasar indicated that 19.3% of assessed newborns had CA, with digestive system disorders being the most common (40.1%), followed by chromosomal abnormalities (11.5%) and neurological anomalies (10.2%). Similarly, a three-year study at RSUP dr. Mohammad Hoesin Palembang recorded 432 cases of CA, with digestive disorders accounting for the highest proportion (60.2%), followed by circulatory system diseases (11.1%) and musculoskeletal conditions (10.4%). Furthermore, research at Dr. Soetomo General Hospital indicates that the three organ systems most frequently affected in foetal congenital anomalies include the digestive system (22 instances; 37.9%). Cleft lip and cleft palate (20 instances; 34.5%); circulatory and musculoskeletal system (each 12 cases; 20.7%).(4,5,8) These variations across different geographical locations may be attributed to genetic predispositions, environmental exposures, healthcare accessibility, and differences in diagnostic methodologies. The results of this study emphasize the need for early detection and targeted healthcare interventions, particularly in regions with high prevalence rates, to improve outcomes for children born with CA.

Analyzing Anomalies: Gender Influences in Congenital Disorders

Recent investigations indicate a significant disparity in the prevalence of CA between male and female infants, with 208 cases (65%) identified in males and 112 cases (35%) in females. Subsequent investigations have revealed a comparable surplus of males. The relationship between prematurity and low birth weight and the heightened occurrence of congenital malformations is well-established in the literature. It is possible that these two factors may represent consequences rather than sources of congenital malformations. (6)

In conjunction with analyses conducted via the Live Birth Information System database of the Minas Gerais Health Secretariat in Brazil, three factors emerged as particularly significant concerning newborns and their association with CA: low birth weight, male gender, and a high-risk 5-minute Apgar score. CA is intricately linked to the rates of morbidity and mortality in infants, particularly during the neonatal period, which is characterized by significant transformations and heightened susceptibility for the newborn. Consequently, the examination, comprehension, and intervention in cases of CA are crucial for timely diagnosis,

the distribution of specialized resources required for the care of the child, and the enhancement of both the health and quality of life for the newborn and their family.(9)

Black et al. (2020) identified The prevalence of males surpassed that of females in 10 out of 14 anomalies, including Hirschsprung's disease, omphalomesenteric duct, congenital diaphragmatic hernia, anorectal malformation, malrotation, esophageal atresia, congenital pulmonary airway malformation, intestinal atresia, omphalocele, and gastroschisis. The prevalence of duodenal atresia and intestinal duplication exhibited no discernible variation between the sexes. There was a notably greater occurrence of biliary anomalies, specifically atresia and choledochal cyst, among females. (10)

Pugnaroni et al. (2023) examine the nuances associated with gender and their ramifications for Congenital Heart Defects (CHD), emphasizing that aortic stenosis and transposition of the great arteries are more commonly observed in males, while females with CHD predominantly present with atrial septal defects. Women typically present with congenital heart defects that are comparatively less severe than those observed in men. The development of the heart can be influenced by hormonal variations, as well as a complex interplay of sex-specific genetic and environmental factors. Variations in parental hormones at the time of conception have been proposed as a potential explanation for the observed outcomes. Some congenital heart defects that are specific to sex may arise from the distinct influences of X and Y chromosome genes in the early stages of heart structure development.(11)

Mapping Out Disparities: Geographical Variations in Healthcare Accessibility

The extensive geography of the Indonesian archipelago presents significant challenges for the equitable distribution of healthcare services. The uneven distribution of healthcare facilities in economically robust urban areas results in disparities in access to medical services across various regions of the country. The uneven geographical distribution influences the utilization of medical services in hospitals, with more populated or developed regions presumed to possess superior healthcare infrastructure. Mahmudiono and Laksono (2021) indicate that healthcare access in specific regions of Indonesia is influenced by factors such as socio-economic status, education level, marital status, occupation, and medical insurance ownership. The disparities are particularly evident when comparing healthcare utilization rates

between urban centers and rural areas, where there is a scarcity of facilities that are often under-resourced.(12)

The disparities in healthcare access are exacerbated by national economic policies that predominantly prioritize more economically developed regions. Laksono et al. (2023) state that economically active regions possess superior healthcare facilities, resulting in a self-reinforcing cycle whereby affluent areas accumulate more healthcare resources, while less affluent regions fall significantly behind. This underscores the necessity of achieving equilibrium in healthcare infrastructural development to ensure that investments are not exclusively directed toward developed regions. Addressing these imbalances requires not only government intervention but also collaboration between public and private sectors in financing and developing healthcare facilities in underserved regions.(13,14)

Navigating Healthcare Accessibility: Urban vs. Rural Divide

The disparity in health services is notably highlighted by the prevalence of CA in Palangka Raya, representing 61.56% of all cases examined in this study. Urban centers typically possess hospitals and health facilities, whereas rural areas often experience developmental delays, leading to lower case rates in North Barito and Sukamara. Both districts reported a single case each, representing 0.31 percent of the overall congenital abnormalities. This disparity illustrates the extent to which urban residents have access to hospitals in comparison to those living in less densely populated regions. This trend is observed globally. Research conducted in sub-Saharan Africa indicates that women residing in urban areas are over one and a half times more likely to utilize health services compared to their counterparts in rural areas. Research conducted in China indicates that older adults residing in rural areas utilize health services less frequently than their urban counterparts, highlighting the significant disparity in health service access between urban and rural regions. (14,15) Comparative insights are essential for customizing public health policies to meet the distinct needs of various regions, particularly in the integration of effective global health strategies aimed at decreasing the prevalence of CA. This approach aids in developing policies that reflect global best practices while being aligned with local socio-economic and environmental circumstances.

Understanding Multiple Congenital Anomalies: Insights from Recent Studies

The occurrence of multiple congenital anomalies complicates the assessment of paediatric cases associated with systemic illnesses of diverse origins. A review at Dr. Doris Sylvanus Regional Hospital from July 2023 to June 2024 identified 320 children diagnosed with CD. Seventeen patients (5.3%) exhibited a range of congenital anomalies, encompassing circulatory, gastrointestinal, genitourinary, musculoskeletal, and craniofacial disorders.

Geleta and Seyoum (2024) conducted a study involving 502 patients with Down syndrome (DS), revealing that 75.1% presented with at least one congenital disorder, while 12.8% exhibited multiple disorders. Patients with Down syndrome frequently exhibit disorders such as heart disease linked to congenital defects in the digestive and genitourinary systems. Common congenital heart defects include patent ductus arteriosus (PDA), ventricular septal defect (VSD), and atrioventricular septal defect (AVSD). The findings align with data from Dr. Doris Sylvanus Hospital, where seven patients (41.2%) exhibited a combination of heart defects (PDA, pulmonary artery stenosis), gastrointestinal malformations (Hirschsprung's disease and anal atresia), and craniofacial clefts (cleft lip) associated with SD.(16) An additional six patients (35.3%) had congenital heart disease with other systemic malformations. These results are consistent with a study conducted in Northern Ireland that analyzed the frequency of CHD, which had a high incidence of CHD with extracardiac defects that may indicate some embryonic pathology or possible underlying syndromic factors. (17)

This study discovered one patient (5.9%) with concurrent annular pancreas, hypospadias, and congenital heart deformity, presumably linked to multiple endocrine or genetic problems. Abnormalities in the genital region commonly associated with Disorders of Sex Development (DSD) are sometimes linked to additional congenital disorders. Markosyan (2021) noted that DSD is often associated with Turner and Klinefelter syndromes, as well as numerous chromosomal anomalies and possible genetic and hormonal modifications. The congenital DSD anomalies identified in many countries may be ascribed to environmental influences, particularly endocrine disruptors and maternal DSD. (18)

Furthermore, one patient (5.9%) displayed gastrointestinal and genital abnormalities; one case involved congenital laryngomalacia and talipes calcaneovalgus, while another presented with microphthalmia and talipes calcaneovalgus. These cases illustrate the need for thorough genetic assessment and proactive planning. The differing rates and trends of congenital anomalies across studies suggest that complex genetic factors, maternal health,

external influences, and local healthcare services play a role in congenital disorders. (17,18) Comparative global findings with studies at dr. Doris Sylvanus Hospital highlight the urgent need for improved prenatal screening services, paediatric surgical subspecialists, and accessible genetic testing. Concentrating on the establishment of registration for congenital abnormalities and the analysis of external risk factors is essential for improving prevention and early intervention strategies moving forward.

Limitations

This study, while comprehensive, has significant deficiencies. The retrospective analysis complicates the establishment of causality between identified congenital abnormalities and associated risk variables. Furthermore, the data were obtained from a singular regional hospital, rendering broader regional or national comparisons quite challenging. Moreover, the utilization of hospital records may be deceptive, as numerous instances of congenital defects remain untreated or receive treatment outside the hospital, necessitating an acknowledgment of potential bias. This may result in an inadequate portrayal of the actual incidence rate. Unintentional mistakes in medical records and classifications about abnormalities may compromise the dependability of the examined data.

Future Research

To improve representativeness and external validity, research should be expanded to more Indonesian locales. Prospective investigations may reveal congenital anomaly causes and risk factors. Future research should also improve congenital anomaly data collection and analysis, including creating a national registry to track changes over time and across areas. This will improve our understanding of this illness over time. Interdisciplinary research that include genetic, environmental, and health services are needed to understand congenital anomaly systems. Understanding how improved health service access and policy implementation affect congenital anomaly incidence and consequences might inform public health issues and resource allocation. These limitations, particularly the focus on specific populations and the scope of study, can be used to help subsequent studies understand congenital defects and enhance measures to lessen their health impact, especially on Indonesian children.

CONCLUSION

This study analyzed the incidence and patterns of congenital abnormalities among pediatric patients at dr. Doris Sylvanus Regional General Hospital over a one-year period, with a focus on variations by geography and gender.

The analysis indicated a significant prevalence of abnormalities in the genital and circulatory systems, highlighting the urgent requirement for specialized health services to address these issues at an earlier stage in life. The prevalence of congenital abnormalities is greater in males and in urban regions like Palangka Raya, where access to health services is more prevalent. This disparity in access to health services between urban and rural areas is prevalent globally and significantly hinders the attainment of universal health coverage across all regions of Indonesia. This study's findings align with a broader investigation in the area, indicating that disparities in access to health services and health outcomes are influenced by socioeconomic, geographic, and demographic factors to address these challenges, it is essential for health policymakers and stakeholders to formulate strategies that target existing gaps and enhance the quality of services provided to children.

This entails increasing financial investment in health facilities that are disadvantaged in terms of equity, improving accessibility to transportation for patients, and broadening health insurance coverage to alleviate the economic strain on families. Furthermore, enhanced monitoring and research into the etiology of congenital abnormalities are essential for their comprehension and effective management. This study highlights the necessity of developing an integrated and responsive health policy to ensure that all children, irrespective of their geographical location or socioeconomic status, receive the necessary care and attention. Implementing these measures, along with insights gained from the policies of other nations, will enhance health services for children with congenital disorders in Indonesia.

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