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ORIGINAL ARTICLE

Clinico-epidemiological profile of disorders of sex development at a tertiary referral hospital in South Kalimantan, Indonesia

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ABSTRACT

BACKGROUND
Disorders of sex development (DSD) comprise a group of complex conditions that pose significant diagnostic and clinical issues. This study aimed to describe the clinical spectrum, temporal patterns, and geographical distribution of DSD cases at a tertiary referral hospital in South Kalimantan, Indonesia.

METHODS
This retrospective descriptive study analyzed medical records of patients with clinical features suggestive of DSD at Ulin General Hospital, Banjarmasin, between 2022 and 2025. Clinical diagnoses were established by a consultant pediatric endocrinologist based on phenotypic evaluation, consistent with approaches used in other Indonesian centers with limited access to karyotyping. Variables assessed included clinical diagnosis, age, sex, year of diagnosis, and domicile, whilst spatial analysis used Geographic Information System (GIS) software.

RESULTS
Among the 100 cases analyzed, severe hypospadias dominated (45 cases, 45%), followed by primary amenorrhea (28 cases, 28%) and micropenis (13 cases, 13%). Males accounted for 67% of cases, females for 32%, and one case had undetermined sex. Mean age at diagnosis was 9.41 ± 7.32 years (range: neonatal period to 35.4 years). Geographic analysis revealed notable disparities, with Banjarmasin contributing 40% of cases, while other districts each accounted for approximately 5%.

CONCLUSIONS
The findings indicate delayed diagnosis, potential underdiagnosis in rural areas, and geographic disparities that likely reflect differences in access to tertiary services rather than true occurrence variation. This highlights the importance of evaluating boys presenting with proximal penile hypospadias or micropenis and girls with primary amenorrhea for timely intervention. Strengthening early detection and expanding diagnostic capacity are essential to improve DSD clinical outcomes.

Keywords: Disorders of sex development, clinical spectrum, epidemiology

INTRODUCTION

Disorders of sex development (DSD) is an umbrella term used to describe congenital conditions in that the development of chromosomal, gonadal, or anatomical sex deviates from the typical male or female developmental pathways.[1,2] The term "disorders of sex development" covers a broad spectrum of conditions with diverse etiologies that include genetic, hormonal, and environmental influences.[3] The clinical spectrum of DSD is wide, ranging from overt ambiguous genitalia detected at birth to more subtle presentations, comprising primary amenorrhea, that emerge during adolescence. Globally, the estimated prevalence of DSD is approximately 1 in 4,500 to 1 in 5,500 live births. This lack of epidemiological information substantially constrains effective healthcare planning and the delivery of appropriate DSD-related services.[1]

The management of DSD requires a multidisciplinary approach involving multiple specialties, including pediatric endocrinology, genetics, urology, gynecology, psychology, and surgery.[4] The complexity of DSD management extends beyond establishing an accurate clinical diagnosis and delivering appropriate medical treatment; it also encompasses the psychological and social dimensions associated with the condition.[5,6] Patients and their families often experience stigma, confused identity, and issues in making critical medical decisions.[7] Therefore, developing a thorough understanding of the DSD clinical spectrum within a specific population is essential for improving referral pathways and supporting integrated management.[8]

Understanding the local epidemiological profile of DSD, including their geographical and temporal distribution, is crucial for effective health-service planning. Conducting spatiotemporal analysis can help identify regions with high case burdens, track temporal shifts, and reveal potential environmental or social contributors, thereby enabling more targeted and strategic interventions.[9]

Disorders of sex development are most frequently diagnosed in newborns with atypical ("ambiguous") genitalia, that cannot on cursory examination be called male or female sex organs. Another type of DSD may be found if the karyotype matches the phenotype, or of there are associated renal disorders or hernias. Other DSD examples are adolescents without puberal features and those with virilization, estrogenization or primary amenorrhea. Some DSD manifest as infertile adults or are found accidentally during examination for unrelated complaints. These diverse presentations underscore the complexity of DSD and the need for careful evaluation and support for affected individuals and their families.[8]

Previous studies in other Indonesian regions have described the clinical profile of DSD. Maritska et al.[9] in Palembang reported hypospadias, undescended testis, and congenital adrenal hyperplasia as the most frequent clinical diagnoses among 173 DSD patients, although chromosomal analysis was unavailable in nearly all cases. Similarly, Maitsa et al.[10] in Yogyakarta identified hypospadias and ambiguous genitalia as the predominant presentations in a pediatric DSD cohort. These findings are consistent with the general understanding of DSD prevalence but are limited by the absence of geographical mapping and temporal analysis. The present study differs from previous reports in two important respects. It integrates spatial analysis using Geographic Information System (GIS) technology to map case distribution across districts of South Kalimantan, thereby identifying potential disparities in access to care.

This study therefore aims to describe the spectrum of clinical presentations of DSD, analyze temporal patterns of DSD cases from 2022 to 2025, and map the spatial distribution to delineate the regional epidemiological profile.

METHODS

Research design

This retrospective descriptive study used patient medical record data and study data from Ulin Banjarmasin Regional General Hospital, Banjarmasin, South Kalimantan, Indonesia. The medical records covered the period from 2022 to 2025, while the study data were collected and analyzed from August to November 2025.

Study subjects

The study population included all patients at Ulin Banjarmasin Regional General Hospital, from 2022 to 2025, who were diagnosed with conditions whose clinical presentation suggest DSD or related genitourinary anomalies. The inclusion criteria comprised patients diagnosed with ambiguous genitalia, severe hypospadias, bilateral undescended testis (UDT), micropenis, clitoromegaly, clinical congenital adrenal hyperplasia (CAH), primary amenorrhea, Turner syndrome, Klinefelter syndrome, and other DSD-related clinical conditions managed under the supervision of a pediatric endocrinologist. The exclusion criteria included patients with incomplete medical records (missing age, diagnosis, or domicile) and patients with acquired genital anomalies due to trauma or complications of circumcision rather than congenital DSD.

Sample size determination

A total sampling technique was applied, whereby all patients who met the inclusion criteria during the study period were included in the analysis, resulting in a total of 100 confirmed DSD cases.

Measurements

In this study, given the unavailability of routine karyotyping and molecular genetic testing at our center during the study period, the diagnosis of DSD was established based on clinical phenotype and physical examination that are consistent with the clinical signs of DSD. Clinical diagnoses were assigned by a consultant pediatric endocrinologist using phenotypic criteria. Consequently, the cases reported here represent a clinical spectrum of DSD, and the diagnostic groupings reflect clinical presentation rather than confirmed genetic etiologies.

Statistical analysis

Data analysis was divided into two main components. First, clinical spectrum analysis was performed, in that categorical variables were summarized as frequencies and percentages, whilst numerical variables were expressed as mean ± standard deviation (SD) or median (range), depending on data distribution. Descriptive statistical analyses were conducted using SPSS software version 25.0. Second, spatial analysis was carried out by geocoding patient addresses to generate thematic (choropleth) maps using Geographic Information System (GIS) software. This approach was used to visualize case density across districts and cities and to identify potential geographic clustering of DSD cases in South Kalimantan.

Ethical clearance

This study received ethical approval by the Health Research Ethics Committee (KEPK) of Ulin General Hospital, Banjarmasin, under approval number 110/VIII-Reg Riset/RSUDU/25. Patient confidentiality was maintained by using codes throughout the data collection and analysis.

RESULTS

A total of 100 cases with diverse clinical presentations of DSD were identified at Ulin General Hospital, Banjarmasin. All diagnoses were established based on clinical outcome, without confirmation by chromosomal testing. The distribution of diagnoses demonstrated a clear bimodal pattern, consisting of congenital genital abnormalities in predominantly male children and disorders of secondary sexual development in predominantly female adolescents and young adults,

Table 1 summarizes the distribution of assigned sex and mean age across DSD diagnostic categories. The overall mean age at diagnosis was 9.41 ± 7.32 years, ranging widely from the neonatal period (0 months) to 35.4 years of age. Male patients accounted for 67% of cases, with a mean age at diagnosis of 8.07 ± 6.50 years, whilst female patients comprised 32% of cases and had a higher mean age of 13.94 ± 8.52 years. One newborn presenting with ambiguous genitalia was assigned an undetermined sex at the time of evaluation.

The clinical diagnostic outcome showed that severe hypospadias was the most common presentation, accounting for 45 cases (45%). Of these, 35 cases (35%) involved isolated severe hypospadias, whilst 10 cases (10%) were associated by comorbidities suggestive of complex disorders of sex development (DSD). Undescended testis (UDT) was the most frequent comorbidity, observed in 7 cases.

Primary amenorrhea was the second most common condition, by 28 cases (28%). Among these, 23 cases were classified as isolated primary amenorrhea, 4 cases were diagnosed as Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome, and 1 case had suspect Turner syndrome, pending further diagnostic confirmation.

Table 1. Clinical spectrum and demographical characteristics of DSD (n=100)

Clinical spectrum	n (%)	Male (M) [n / Age]	Female (F) [n / Age]	Undeter-mined [n /Age]	Total Age (years)
Severe hypospadias (Total)	45 (45)	45 (5.82±3.61)	-	-	5.82 ± 3.61
Isolated	35 (35)	35 (5.61±3.45)	-	-	5.61 ± 3.45
With comorbidity	10 (10)	10 (6.66±4.28)	-	-	6.66 ± 4.28
with genital ambiguity	2 (2)	2 (12.54±1.76)	-	-	12.54 ± 1.76
with UDT/cryptorchidism	7 (7)	7 (5.16±3.60)	-	-	5.16 ± 3.60
with micropenis	1 (1)	1 (5.67)	-	-	5.67
Primary amenorrhea (Total)	28 (28)	-	28 (19.53±5.41)	-	19.53 ± 5.41
Primary amenorrhea	23 (23)	-	23 (19.71±5.62)	-	19.71 ± 5.62
Primary amenorrhea with MRKH	4 (4)	-	4 (22.10±6.32)	-	22.10 ± 6.32
Primary amenorrhea with suspect Turner syndrome	1 (1)	-	1 (15.00)	-	15
Other presentations (Total)	27 (27)	22 (3.42±3.95)	5 (0,6 ± 1,34)	1 (6.50)	4.89 ± 5.15
Micropenis	13 (13)	13 (5.54±5.12)	-	-	5.85 ± 5.12
Genital ambiguity	8 (8)	5 (6.11±5.32)	2 (0.00±0.00)	1 (6.50)	3.46 ± 3.62
Gynecomastia (adolescence)	1 (1)	1 (14.75)	-	-	1 (4.75)
CAH	2 (2)	-	2 (0.00±0.00)	-	0.00 ± 0.00*
Turner syndrome	1 (1)	-	1 (3.00)	-	3
Other DSD**	2 (2)	2 (1.58±2.12)	-	-	1.58 ± 2.12

Note: Data presented as n (%) and mean ± SD; UDT: undescended testis; MRKH: Mayer-Rokitansky-Küster-Hauser syndrome; CAH: congenital adrenal hyperplasia

Analysis of age at diagnosis identified three distinct clinical patterns. First, neonatal–infant presentations (0–1 year), comprising congenital adrenal hyperplasia (CAH) and genital ambiguity, were generally detected early; however, diagnosis still relied mainly on visual screening, devoid of routine support by 17-OHP testing. Second, childhood diagnoses (1–12 years) were predominantly cases of severe hypospadias, with a mean age at diagnosis of 5.82 ± 3.61 years. This finding describes a delayed diagnosis, considering that the optimal period for surgical intervention is before 2 years of age. Third, adolescent–adult presentations (>13 years) were primarily characterized by primary amenorrhea, with a mean age at diagnosis of 19.53 ± 5.41 years, reflecting a substantial diagnostic delay of approximately 3–5 years beyond the expected age of menarche.

Based on patient residence data, geographic analysis revealed pronounced disparities in the distribution of DSD cases across South Kalimantan during the 2022–2025 period. Banjarmasin accounted for 40% of all reported cases, a proportion substantially higher than that of other districts. Most other districts each contributed approximately 5% of cases, whilst Tanah Bumbu regency recorded the lowest proportion at 2%.

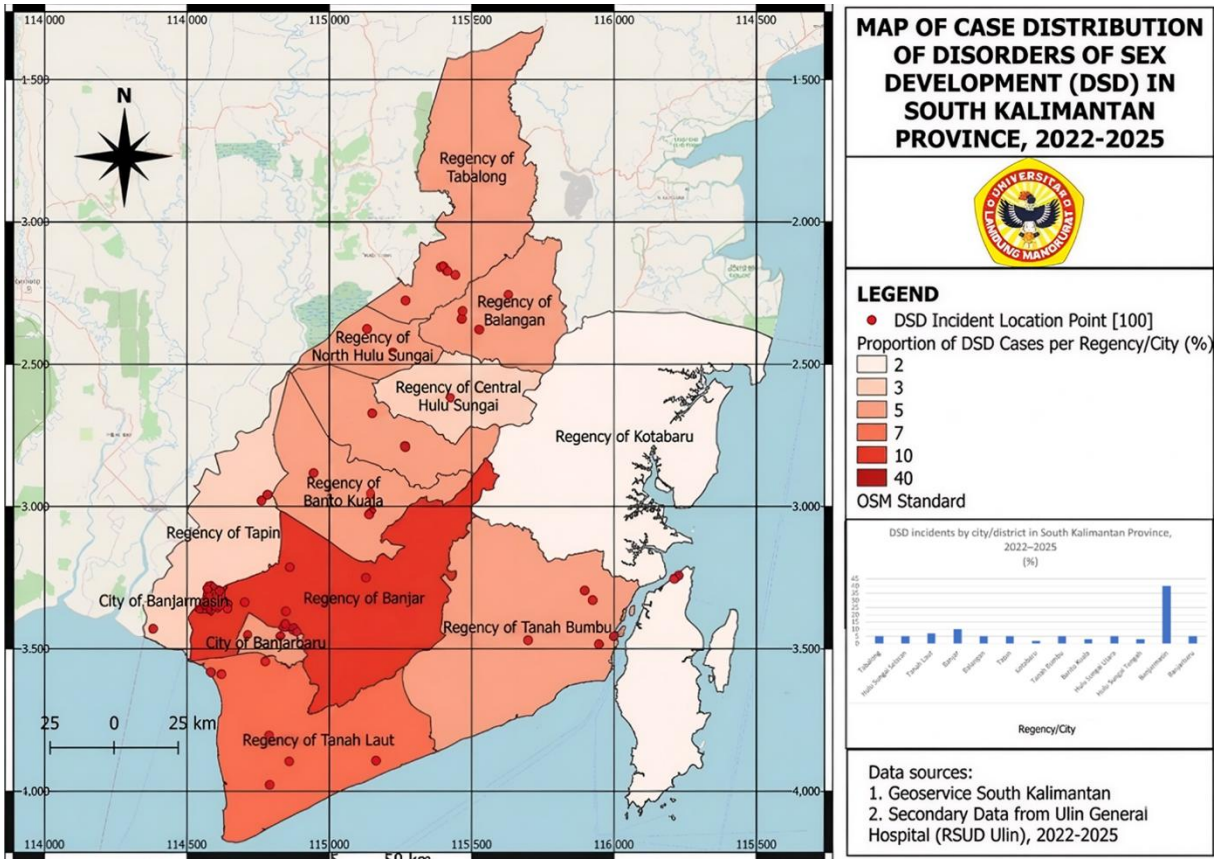

Figure 1. Map of distribution of **c**linically diagnosed DSD cases based on physical phenotypicsSigns in South Kalimantan Province, 2022-2025

DISCUSSION

Our analysis of 100 cases over a four-year period (2022–2025) revealed three key outcomes. First, severe hypospadias was the most common clinical presentation (45%), followed by primary amenorrhea (28%) and micropenis (13%). Second, the age at diagnosis ranged widely from the neonatal period to 35.4 years, with an overall mean age of 9.41 ± 7.32 years, indicating substantial delays in detection. Third, geographic analysis demonstrated marked disparities, with 40% of cases concentrated in Banjarmasin, whilst peripheral districts each accounted for only 2–5% of reported cases.

The predominance of severe hypospadias in our cohort aligns by outcome with other Indonesian centers. A study conducted in Palembang similarly listed hypospadias, undescended testes (UDT), and CAH as the most common diagnoses.[10] Meanwhile, data from Yogyakarta in the same year identified hypospadias and ambiguous genitalia as the predominant clinical presentations.[11] These outcomes are consistent with the present study, in that hypospadias was the predominant clinical presentation. In the absence of karyotypic confirmation, this phenotype warrants evaluation as a potential manifestation of undervirilization, clinically consistent with a 46, XY DSD etiology.[10,11]

Globally, hypospadias is recognized as a relatively common congenital condition in male infants, with an incidence ranging from 1 in 125 to 1 in 250 male births.[12,13] This condition arises from disrupted embryologic development of the urethra, influenced by a combination of genetic and environmental factors, that may contribute to the observed increase in cases.[14]

In cases of 46,XX DSD, congenital adrenal hyperplasia (CAH) has been reported as the most common clinical finding (60%) in previous Indonesian study,[15] presenting in both salt-wasting and simple virilizing forms. The underlying pathophysiology involves an enzymatic defect most commonly 21-hydroxylase deficiency that leads to excessive androgen production and subsequent virilization of the external female genitalia.[15,16]

The wide age range at diagnosis (0–35.4 years) reflects not only the phenotypic variability of DSD but, more importantly, highlights systemic weaknesses in early detection.

In our study, hypospadias was diagnosed at a mean age of approximately 5.82 ± 3.61 years, despite clinical guidelines recommending that corrective treatment be performed before 12 months of age to reduce the risk of complications, including infertility and later psychosocial difficulties.[17] The primary contributing factors include limited parental awareness and an underdeveloped pediatric screening system.[18]

In our cohort, the diagnosis of primary amenorrhea occurred at a mean age of 19.53 ± 5.41 years, reflecting a delay of approximately 3–5 years beyond the expected age of menarche. This delay appears to be largely attributable to limited awareness among parents and healthcare providers regarding the clinical indicators of the condition. Primary amenorrhea is defined as the absence of menstruation by age 14 in individuals devoid of secondary sexual characteristics, or by age 16 in persons with or without secondary sexual characteristics.[19] Primary amenorrhea can result from several underlying causes, including genetic abnormalities, such as Turner syndrome, and anatomical conditions, such as Müllerian agenesis. [20]

One study showed that only about 25% of infants with ambiguous genitalia were referred to a physician within the first month of life, whilst in most other cases the condition was identified much later, even as late as puberty.[21] Early detection is essential because it allows timely intervention by a multidisciplinary team, including endocrinologists, surgeons, genetic counselors, and psychologists, to address the complex nature of DSD.[22]

The distribution of DSD cases shows marked geographic differences. Banjarmasin accounts for 40% of all reported cases, a proportion far higher than that of other districts. This pattern does not represent a genuinely higher prevalence, but instead reflects Banjarmasin's role as a tertiary referral center equipped with more complete diagnostic resources, specialist availability, and support services that are not yet distributed evenly across the region.

Most other districts reported case proportions of around 5%, a consistent pattern that suggests potential systemic underdiagnosis. The low figures may arise from limited neonatal screening, insufficient capability among healthcare workers to recognize the diverse clinical features of DSD, and socio-cultural barriers that hinder reporting. Tanah Bumbu recorded only 2% of cases, which is the lowest rate, that should be monitored as a possible indication of weak case detection or strong stigma that prevents patients from accessing healthcare services.

The disparity in case distribution carries significant medical and social consequences. Patients living outside major referral centers are at greater risk of delayed diagnosis, suboptimal treatment, and limited psychosocial support. Addressing these gaps requires strengthening referral systems, expanding early detection capacity inside of primary healthcare facilities, and providing education to reduce stigma, steps that are essential for building an inclusive and equitable DSD service landscape across all regions.[23]

The low prevalence of complex genetic conditions comprising Turner syndrome, that accounted for only 1% of the cohort, should be interpreted with caution. Although Turner syndrome is a rare condition, occurring in approximately 1 in 2,500 live female births, the identification of only a single case inside of a cohort of 100 patients by DSD may not accurately reflect its true prevalence in the referral population.[24] This minimal number may result from diagnostic limitations. Accurate identification of DSD cases often depends on advanced genetic assessments, including karyotyping and molecular analysis.

Diagnoses in our study were made solely on clinical outcome, without confirmation by chromosome testing, that should be the primary diagnostic modality according to the 2006 Chicago Consensus.[25,26] A study conducted at dr. Mohammad Hoesin Hospital identified 19 clinical variations that met the criteria for DSD across a wide age range. The situation in Palembang closely mirrors that in Banjarmasin, where access to comprehensive diagnostic testing and integrated management remains limited.[10] In both settings, cytogenetic data confirming DSD are either unavailable or extremely scarce, resulting in significant underreporting.

Optimal DSD management requires a fully integrated, multidisciplinary approach. One of the major issues faced by regional referral hospitals is the limited availability of a complete team of specialists. Ideally, patients with DSD should be managed by a multidisciplinary team (MDT) consisting of pediatric and general

endocrinologists, surgeons (including urologists and pediatric surgeons), genetic counselors, psychologists, and religious leaders. However, shortages of specialized human resources and expertise in regional hospitals often hinder the establishment of such comprehensive teams.[27]

In addition, achieving an accurate diagnosis depends heavily on the availability of specialized diagnostic tools. Chromosomal and genetic analyses, along with advanced endocrine hormone testing, are often unavailable at the regional level, requiring specimens or patients to be referred to larger medical facilities, an approach that is both time-consuming and costly. These delays may postpone diagnosis, surgical intervention, and essential counseling, particularly in cases of ambiguous genitalia where timely sex determination is crucial for supporting the psychological well-being of both patients and their families.[28]

The management of disorders of sex development also requires ongoing, long-term psychosocial support. As a lifelong condition, DSD can make it challenging to ensure that patients continue to receive appropriate care beyond the initial diagnosis and surgical treatment.[29] The absence of a structured patient-tracking system and consistent counseling services can affect patient outcomes negatively.

The importance of early detection and education regarding DSD cannot be overstated. Routine health screenings by infancy through puberty can improve treatment outcomes and reduce long-term morbidity. These measures can help minimize diagnostic gaps and enhance the overall quality of care provided to affected patients and their families.[30]

This study has several important limitations that should be acknowledged. First, diagnoses were made based solely on clinical outcome devoid of confirmation by chromosomal testing or comprehensive genetic analysis, that limits the accuracy of classification according to current DSD nomenclature. Second, as a single-center retrospective study, the outcome may not be fully generalizable to other regions in Indonesia that differ in healthcare infrastructure and population characteristics. Third, the geographic concentration of cases in Banjarmasin likely reflects referral patterns rather than true epidemiological distribution, potentially obscuring the actual burden of DSD in peripheral areas. Fourth, information on psychosocial outcomes, family counseling, and long-term follow-up was not available in the medical records, limiting a comprehensive understanding of the broader impact of DSD on patients and their families. Finally, the four-year study period may not adequately capture long-term temporal trends or potential seasonal variations in case presentation.

Future studies should prioritize several key areas to advance DSD care in Indonesia. First, the establishment of a prospective multicenter registry by standardized data collection protocols would generate more robust epidemiological data and facilitate comparisons across regions. Second, expanding access to chromosomal and genetic testing at regional referral centers is essential for accurate diagnosis and appropriate management planning. Third, the development of community-based early detection programs, along with training for primary healthcare workers to recognize DSD presentations, could substantially reduce diagnostic delays. Fourth, longitudinal studies focusing on psychosocial outcomes, quality of life, and long-term health consequences among individuals with DSD are needed to inform comprehensive care protocols. Finally, health policy research evaluating the cost-effectiveness of various screening and management strategies would support evidence-based resource allocation for DSD services.

The findings of this study have several clinical implications. First, a national training program for primary healthcare providers on neonatal DSD screening is required. Second, a tiered referral system and telemedicine should be strengthened to overcome limited access to multidisciplinary teams and diagnostic testing at regional hospitals. Third, local governments should strive to provide chromosomal and hormonal testing services at tertiary referral hospitals to ensure accurate DSD diagnosis. Fourth, reproductive health education among adolescents needs to be intensified to reduce delayed diagnosis of primary amenorrhea. Finally, a community-based approach involving religious leaders and psychologists is important to address sociocultural stigma that hinders DSD case reporting.

CONCLUSION

The clinical presentation spectrum was dominated by severe hypospadias and primary amenorrhea, with most patients being male and diagnosed across a wide age range. A marked

geographical imbalance was observed, with cases predominantly concentrated in Banjarmasin, likely reflecting the central role of tertiary referral centers rather than true variations in disease prevalence. These outcomes point to several systemic issues, including delayed case identification, potential underdiagnosis in rural areas, and reliance on clinical assessment in the absence of genetic confirmation. Strengthening early detection, improving equitable access to multidisciplinary care, and expanding diagnostic capacity across the region are essential to enhance patient outcomes and support more comprehensive DSD management.

Conflict of Interest

The authors declare that there are no conflicts of interest related to this study. No financial or personal relationships exist that could have influenced the conduct of the research or the interpretation of its outcome.

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Authors' Contributions

MU contributed to conceptualization, methodology, formal analysis, investigation, data curation, writing of the original draft, project administration, and funding acquisition; IWH contributed to methodology, validation, investigation, resources, writing review and editing, and supervision; NAP contributed to data contribution, validation, formal analysis, visualization, and writing review and editing; NB contributed to data contribution, data curation, and writing review and editing; AM contributed to investigation, resources, and data curation; NRA contributed to investigation, resources, and writing review and editing; and NAM contributed to resources, software, and administration. All authors have read and approved the final version of the manuscript.

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Data Availability Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request and with permission from the Health Research Ethics Committee of Ulin General Hospital, Banjarmasin. The data are not publicly available due to privacy and ethical restrictions concerning patient confidentiality.

Declaration of AI Usage in Scientific Writing

During the preparation of this work, the authors used artificial intelligence (AI) – assisted tools for specific purposes. Scispace was employed to facilitate literature searching and initial screening of relevant articles. Deepseek and Grammarly were used to improve language clarity, grammar, and overall readability oe the manuscript. All AI-generated suggestions were critically reviewed and verified by the authors, who take full responsibility for the accuracy and integrity of the final content.

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