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# Help-seeking Behavior of Disorders of Sex Development Patients in Palembang, Indonesia

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ABSTRACT

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#### 1. Introduction

Disorders of Sex Development is a disorder of chromosomal, gonadal, or anatomical sex development with a prevalence of 1:4500 to 1:5000 births.<sup>1,2</sup> DSD used to be known as intersex, pseudohermaphrodite, hermaphrodite, and sex reversal before it was finally agreed upon as DSD based on the Chicago consensus in 2006.<sup>3</sup> Etiology of DSD are often due to genetic abnormalities, still some are multifactorial such as an environmental cause or drugs use in pregnancy.<sup>4</sup>

DSD is known to have a variety of chief complaints and clinical manifestations ranging from mild to severe, such as mild hypospadias, cryptorchidism, primary amenorrhea, gynecomastia, and infertility.<sup>1,3,5</sup> Ambiguous genitalia is one of the common chief complaints brought patients to the hospital that should be recognized since the neonatal period.<sup>6</sup> Unfortunately, DSD is not always detected at birth and can be detected in various age groups. This is because there are various chief complaints and clinical manifestations that can be found in each age

**Introduction.** Disorders of Sex Development (DSD) is a congenital disorder in which there is atypical chromosomal, gonadal, or anatomical sex development. Children with DSD are often brought to the hospital too late for care due to limited knowledge about this condition among medical practitioners. Early diagnosis is important to improve prognosis and minimize complications and comorbidities of DSD patients. **Methods.** This study is a descriptive observational study with a cross-sectional approach through interviews and data collection from medical records of Dr. Mohammad Hoesin Palembang Hospital in 2020 - 2023. **Results.** In this study, it was found that the infancy age group (28 days - 1 year) was the most common age of arrival of patients to the hospital (29%) and the most common main complaint was clitoral enlargement (19.4%). **Conclusion.** This study found a wide variation in patients' chief complaints, which makes the age of arrival of DSD patients to the hospital still quite late.

group of DSD patients. The variety of chief complaints and clinical manifestations by age group makes early screening for DSD a challenge for each healthcare facility.

The second most common major complaint of DSD patients is vomiting which can cause life-threatening conditions caused by adrenal insufficiency.<sup>7</sup> This study found children with complaints of vomiting, diarrhea, and seizures were the common cause of salt-wasting CAH patients being rushed to the emergency room. Fluid loss in children without adequate replenishment of fluids may lead to hypovolemic shock.<sup>8</sup> Due to larger body surface area compared to adults, children also experience higher fluid losses.<sup>8</sup> Almost 90% of adrenal crisis patients will experience hypotension or hypovolemic shock if not treated properly and salt-wasting CAH patients have a high risk of experiencing repeated episodes of adrenal crisis.<sup>7</sup>

This study found the infancy age group (28 days – 1 year) to be the most common age group for treatment. In contrast to the previous study, the

majority of DSD patients came from the age of three to five.<sup>9</sup> This may be due to this age when children are starting to be enrolled in school and parents are filling out the child's gender form. At first, parents are not concerned about their child's condition. However, after they are asked about their child's gender, they start questioning whether their genitalia ambiguous children should be assigned as male or female. As mentioned earlier, most DSD patients have atypical external genitalia seen since the neonatal period, which makes DSD patients eligible for early screening and intervention for the condition. However, this study only found a few patients were seeking help during this period due to a lack of knowledge of midwives and doctors about this condition.

Lack of knowledge of midwives and doctors in primary care resulting in late DSD diagnosis is probably due to the absence of guidelines in the diagnosis and management of DSD. The higher knowledge of health workers and the community about this condition will accelerate the diagnosis of DSD so that comorbidities and complications of DSD patients can decrease. Parents tend to bring their genitalia ambiguous child to the hospital because of the complications following such as gastrointestinal symptoms (vomiting, diarrhea, or abdominal pain) caused by salt-wasting CAH.<sup>10</sup> This is followed a previous study stated that CAH is the most common DSD condition with various clinical manifestations.<sup>11</sup> DSD patients with milder degrees of ambiguous genitalia were referred after the age of 20.12

However, studies attempting to determine the incidence and prevalence of DSD are scarce. RSUP Dr. Mohammad Hoesin Palembang is located in South Sumatra, one of Indonesia's largest provinces. It is one of Indonesia's national referral hospitals. So this study tries to present collected data from DSD patients in RSUP Dr. Mohammad Hoesin Palembang.

#### 2. Methods

This was an observational descriptive study with a cross-sectional approach. The ethical exemption number of this study is No.DP.04.03/D.XVIII.06.08/ETIK/186/2024. The inclusion criteria of this study were (1) patients diagnosed both with the term DSD and/or other terms used previously such as intersex, pseudohermaphrodite, hermaphrodite, and others. Samples could also be (2) all patients who seek therapy and/or receive inpatient or outpatient care at RSUP Dr. Mohammad Hoesin Palembang who was diagnosed with DSD based on the 2006 Chicago Consensus classification by gualified medical professionals. The exclusion criteria of this study is medical record data of DSD patients with no contact number. The minimum sample size of this study was 30 patients and this study found 31 patients who had been diagnosed with DSD at Dr. Mohammad Hoesin Palembang Hospital for the period 2020 - 2023. From

medical records, the variables sought were the patient's chief complaints and sex at the time of admission.

## 3. Results

Among thirty-one patients included in the study, the majority of patients came during their infancy, which is between 28 days to 1 year of age (29%). Followed by early childhood, which is between 6 to 8 years of age. The least age group found was middle childhood and adult as can be seen in Table 1.

This study found thirteen variations of chief complaints. The most common chief complaint was clitoral enlargement with six patients (19.4%) complaining, followed by vomiting complained by five patients (16,1%), urination out of place complained by four patients (12.9%), and ambiguous genitalia complained by three patients (9.7%). Other chief complaints can be seen in Table 2.

## 4. Discussion

As can be seen in Table 1, most patients in the study came during their infancy (28 days – 1 year). In contrast to research conducted by Gürbüz F., the average age of DSD patients first seeking treatment is the age of three to five years.<sup>9</sup> Most DSD patients have atypical external genitalia that can be seen since the neonatal period which makes DSD patients able to get early screening and intervention for the condition. However, in this study, only a few patients were diagnosed during this period may be due to the fact that health workers are not well-trained to diagnose DSD early due to the lack of guidelines in the diagnosis and management of DSD.<sup>6</sup>

The second most common help-seeking age of DSD patients is during early childhood since this age range is the most common age range for circumcision and was detected by doctors after examination of external genitalia before performing circumcision.<sup>13,14</sup> This was proven by several patients in this study who were diagnosed and referred with hypospadias after precircumcision genital examination. This might also be due to children at this age beginning to enroll in school, requiring parents to complete forms including their child's gender. Initially, parents may not be concerned about their child's gender, they start to question whether their ambiguous genitalia children should be assigned as male or female.

The next age of arrival of DSD patients to the hospital is toddler period. In many countries including Indonesia, DSD is not widely known by families, communities, and even health workers themselves which causes some patients to be brought to the hospital when reaching toddler age (1 - 5years) so that public and health workers' knowledge of this condition needs to be improved.<sup>15</sup>

Table 1. Distribution of DSD patients based on help-seeking age (N=31)			
Help-seeking Age of DSD Patients	n	%	
Neonates (≤ 28 days)	5	16,1	
Infancy (28 days – 1 year)	9	29	
Toddler (1 year – 5 years)	5	16,1	
Early Childhood (6 years – 8 years)	6	19,4	
Middle Childhood (9 years – 11 years)	1	3,2	
Adolescence (12 years – 18 years)	4	12,9	
Adult (>18 years)	1	3,2	

Table 2. Distribution of DSD patients based on chief complaints (N=31)			
<b>Chief Complaints of DSD Patients</b>	n	%	
Clitoral enlargement	6	19,4	
Clitoral enlargement + Vomiting	1	3,2	
Vomiting	5	16,1	
Vomiting + Diarrhea	1	3,2	
Vomiting + Seizures	1	3,2	
Ambiguous genitalia	3	9,7	
Urination out of place	4	12,9	
Unpalpable both testicles	2	6,5	
Unpalpable right testicle	2	6,5	
Unpalpable left testicle	1	3,2	
Small penis	1	3,2	
Not menstruating yet	2	6,5	
Short stature + Not menstruating yet	2	6,5	

This study found a small proportion of patients presented during adolescence, which is in line with the previous study that found a small number of DSD patients presenting in adulthood with complaints of late puberty or primary amenorrhea.<sup>6</sup> DSD patients who present in adulthood usually present with a milder degree of ambiguous genital, but the child does not experience puberty or menstruation, which makes diagnosis at a later age.<sup>14</sup> DSD conditions can affect gonadal function so puberty in children with DSD can be delayed, atypical, or even no puberty.<sup>16</sup> This is the reason why the patient's parents brought their child for examination and diagnosis of their child's condition.

As can be seen in Table 2, clitoral enlargement was found to be the main complaint causing patients to seek help. This is guite different from other studies that found 24.2% of patients came with chief complaints in the form of ambiguous genitalia, short stature 20%, isolated perineal hypospadias 8.42%, primary amenorrhea as much as 8.42%, late puberty 8.42%, micropenis 6.3%, and clitoromegaly 5.26%.<sup>17</sup> Another study also found that the most common cause of referral to pediatricians was ambiguous genitalia which occurred in 82.9% of patients and usually occurred in the first year of life.<sup>12</sup> This is due to the appearance of atypical genitals and there are still many people who consider these things taboo. The condition of ambiguous genitalia can be micropenis or clitoromegaly which is usually first noticed by the patient's mother which makes the patient to seek help earlier. It is different from

primary amenorrhea with normal external genitalia that makes the patient come later, which is in adolescence entering adulthood. The criteria of ambiguous genitalia and clitoromegaly in this study were separated because the medical record's anamnesis data did not mention the type of the patient's ambiguous genitalia.

The next major complaint was vomiting, which is a life-threatening condition caused by adrenal insufficiency.<sup>7</sup> In this study, it was found that children with complaints of vomiting, diarrhea, and seizures were the most common causes of salt-wasting CAH patients being rushed to the emergency room. The mortality rate of patients with adrenal crisis varies because almost 90% of adrenal crisis cases will experience hypotension or hypovolemic shock if not treated properly and salt-wasting CAH patients have a high risk of experiencing repeated episodes of adrenal crisis.<sup>7</sup>

Penoscrotal hypospadias which is classified as 46, XY DSD is the third most common complaint after vomiting. This is slightly different from what was found in another study which hypospadias was the second most common cause of consultation after ambiguous genitalia.<sup>18</sup> Patients with hypospadias are often diagnosed during genital examination before circumcision.19 Hypospadias itself is the most common congenital disorder found in men.20 Hypospadias can also be a clinical manifestation of patients with CAH, especially penoscrotal hypospadias.<sup>21</sup> However, at Dr. Mohammad Hoesin Hospital, patients with penoscrotal hypospadias were

not genetically examined so they could not be assigned as CAH.

In another study on Turner syndrome, it was mentioned that the main complaint in 41% of patients with Turner syndrome was delayed growth.<sup>22</sup> This was also found in this study where short stature due to delayed growth was found in two patients in which both patients were diagnosed with Turner syndrome.

Limitations in this study are many patients could not be contacted and were not willing to be interviewed which caused the distribution of patients' data to be poorly distributed and some of the results of this study are slightly different from previous studies that have been conducted. Furthermore, patients with suspicion of DSD such as patients with penoscrotal hypospadias did not undergo karyotyping or further genetic testing so the definitive diagnosis of DSD was not established, but patients were only diagnosed based on clinical manifestation. Further research on the relationship between help-seeking behavior and the outcome of DSD patients can be conducted.

## 5. Conclusion

Help-seeking age of DSD patients is still late so knowledge about the condition of DSD both among health workers and the community has to be improved. If health workers' knowledge about this condition increases, the complications can be minimized and patient prognosis improves. Research on the relationship between the knowledge of medical staff and the help-seeking behavior of DSD patients is needed to elucidate the reasons behind the delayed help-seeking age of DSD patients to the hospital.

## 6. Author Contribution

T.C.K. and Z.M carried out the study. T.C.K. wrote the manuscript with support from Z.M., S.P., E.F.Z., and R.I. fabricated the samples. E.F.Z. and R.I. helped supervise the project. E.F.Z. and R.I. conceived the original idea. Z.M. supervised the project.

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