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Disorders of Sex Development in South Kalimantan: First Case Reports of a Neonate with 46,XX Virilization and an Adolescent with 46,XY Undervirilization

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Abstract

Introduction: *Disorders of Sex Development (DSD) are rare congenital conditions with high diagnostic complexity.* ***Objective:*** *To describe the challenges in the diagnosis and management of DSD in South Kalimantan through two case reports.* ***Method:*** *Data were obtained from the medical records of two DSD patients in South Kalimantan.* ***Results and Discussion:*** *Case 1: A 10-day-old 46,XX neonate with ambiguous genitalia (Prader III), hyperkalemia (K 8.8), hyponatremia (Na 134), and hypoglycemia. Salt-wasting Congenital Adrenal Hyperplasia (CAH) was diagnosed (17-OHP 237 ng/mL). Case 2: A 17-year-old raised female with progressive virilization since age nine. Examination showed Prader IV, 7-cm phallus, bilateral testes (10–12 mL), absent Müllerian structures, and 46,XY karyotype, suggesting Partial Androgen Insensitivity Syndrome (PAIS) or 5α-reductase deficiency. Both cases reflect regional limitations: unavailable comprehensive hormonal testing, genetic molecular analysis, and no permanent multidisciplinary DSD team.* ***Conclusions:*** *Limitations in diagnostic resources at the regional level significantly impact the optimization of DSD patient management, necessitating the development of diagnostic capacity and multidisciplinary collaboration*

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Disorders of Sex Development in South Kalimantan: First Case Reports of a Neonate with 46,XX Virilization and an Adolescent with 46,XY Undervirilization

Introduction

Disorders of Sex Development (DSD) are congenital conditions in which discrepancy among chromosomal, gonadal and anatomical sexual development. Since the introduction of the karyotype-based classifications 46, XX DSD, 46, XY DSD and sex chromosome DSD in the Chicago Consensus in 2006, older terms such as "hermaphroditism," which have a profound social stigma attached to them, are no longer used. There is an estimated prevalence of DSD globally at 1 in 4,500–5,500 live births yet that figure likely reflects considerable underreporting especially from developing nations with no standardized neonatal screening systems (Markosyan, 2021).

Among 46,XX DSD, salt-wasting congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency is the most critical because it can cause life-threatening adrenal crisis in the first week of life. Affected neonates present with hyponatremia, hyperkalemia, and virilized genitalia, a picture easily mistaken for sepsis if 17-hydroxyprogesterone (17-OHP) testing is unavailable (Juniarto et al., 2018). In 46,XY DSD, undervirilization can result from Partial Androgen Insensitivity Syndrome (PAIS) or 5α-reductase deficiency. Newborns with a female or mildly ambiguous phenotype are frequently raised as girls in the absence of a definitive diagnosis, presenting with dramatic virilization during puberty. Diagnosis at a later age can have serious psychosocial ramifications, including gender dysphoria, social isolation, and debilitating clinical depression (Lee, Houk, & Faisal, 2016).

DSD management in Indonesia is hindered by considerable systemic factors. Fludrocortisone is not included in the JKN/BPJS formulary, karyotyping and 17-OHP testing are only available at Type A hospitals situated in major cities, while multidisciplinary teams for comprehensive DSD care exist only at a few national referral centres on Java (Listyasari et al., 2021). To date, no scientific publication regarding DSD cases in South Kalimantan had been published in an indexed journals. This report describes for the first time two contrasting DSD spectra: 46,XX neonate with salt-wasting adrenal crisis and accompanying genital virilization, as well as a 46,XY adolescent raised female who exhibited progressive virilization until the age of 17.

The aim of this paper is to report the clinical presentation, diagnostic method and management of two DSD cases in South Kalimantan and describe their structural diagnostic and therapeutic barriers at regional facilities.

Method

This study employs a descriptive *case report* design presenting two cases of DSD with contrasting clinical presentations from the same regional referral center. A descriptive approach was chosen because it allows for an in-depth description of the clinical, diagnostic, and management characteristics of a rare condition that has not previously been documented in the South Kalimantan region. The case report design is internationally recognized as a valid method for establishing an initial foundation of scientific evidence for conditions with low incidence and high clinical complexity, such as DSD (Juniarto et al., 2021).

Both cases were identified at the Pediatric Endocrinology Clinic and the Emergency Department of Ulin General Hospital in Banjarmasin, the regional referral center for South Kalimantan, between January and May 2025. The first case was a 10-day-old female-genotyped (46,XX) neonate referred from a secondary-level general hospital with complaints of recurrent vomiting, lethargy, severe hyperkalemia (K 8.8 mmol/L),

hyponatremia, and ambiguous genitalia, accompanied by a history of a sibling's death with similar symptoms. The second case was a 17-year-and-1-month-old adolescent with female *sex development* referred with an initial diagnosis of DSD, accompanied by progressive virilization since elementary school age, *Prader stage* IV, a 7 cm *phallus*, and palpable bilateral testes with a volume of 10–12 mL. Clinical data for both cases were collected retrospectively and prospectively through medical record reviews, laboratory results, imaging studies, and outpatient *follow-up.*

Diagnosis in both cases was established through an integrated approach that included a thorough medical history including family history and *a family tree* a standardized physical examination using the Prader scale to assess the degree of virilization in 46, XX individuals and *the External Masculinization Score* (EMS) for 46,XY individuals, as well as supportive tests. Chromosome analysis (*karyotyping*) was performed using the G-banding method with a band resolution of 400, counting 100 cells and analyzing 10 cells, in accordance with the standards *of the International System for Human Cytogenomic Nomenclature* (ISCN), which confirmed a 46,XX karyotype in the first case and a 46,XY karyotype in the second case. Electrolyte levels, 17-OHP levels, blood glucose, and hormonal profiles were measured in an accredited clinical laboratory using standard *immunoassay* methods. Urogenital ultrasound imaging was performed to assess the presence of Müllerian structures (uterus and ovaries) and gonadal morphology in both cases.

The management provided is based on the clinical guidelines from the Indonesian Pediatric Society (IDAI) Textbook of Pediatric Endocrinology, as well as those from *the European Association of Urology* (EAU) *guidelines on paediatric urology*, adapted to the availability of resources at regional facilities. (Batubara et al, 2018; EAU, 2025). In the first case, management included correction of acute hyperkalemia with 10% intravenous calcium gluconate, salbutamol nebulization, maintenance fluid rehydration, initiation of intravenous hydrocortisone at a *stress* dose subsequently switched to oral therapy, addition of fludrocortisone, and oral NaCl supplementation. In the second case, the approach included cytogenetic confirmation, intensive psychological counseling, and planning for surgical reconstruction in collaboration with the urology team.

This entire research procedure has received ethical approval from the Health Research Ethics Committee (KEPK) of Ulin General Hospital in Banjarmasin, under approval number 110/VIII-Reg Riset/RSUDU/25. Written *informed consent* was obtained from the patient's parents in the first case and from the patient and their family in the second case prior to publication. Patient confidentiality is fully guaranteed through the use of anonymous codes in all published data.

Research and Discussions

1. Results

Case 1: 46,XX Neonate with Adrenal Crisis in *Salt-Wasting* Type CAH

A 10-day-old female infant (46,XX) was referred to the Emergency Department of Ulin General Hospital in Banjarmasin from a secondary-level general hospital in critical condition. The main complaints were recurrent vomiting, lethargy, and refusal to breastfeed since the age of 6 days. During the medical history, the patient's mother revealed a history of the death of her first child at 19 days of age with similar complaints without a clear diagnosis, a strong indication of an autosomal recessive inheritance pattern in the family.

Physical examination revealed signs of severe dehydration, a weak pulse, *a capillary refill time* (CRT) >3 seconds, hyperpigmentation of the areola and genitalia, clitoromegaly (*phallus* 1.5 cm), fusion of the posterior labia, and no palpable gonads on either side. The virilization score was at *Prader stage* III. Laboratory tests revealed extreme hyperkalemia (K 8.8 mmol/L), hyponatremia (Na 134 mmol/L), and hypoglycemia (30 mg/dL). A 17-OHP level of 237 ng/mL (normal range <10 ng/mL) is a pathognomonic biochemical marker for 21-hydroxylase deficiency. Chromosome analysis using G-banding confirmed a 46, XX karyotype. Abdominal ultrasound revealed a normally positioned uterus and bilateral ovaries, along with bilateral adrenal hyperplasia. The diagnosis was established as acute adrenal crisis due to *salt-wasting* type CAH with 46,XX genital virilization.

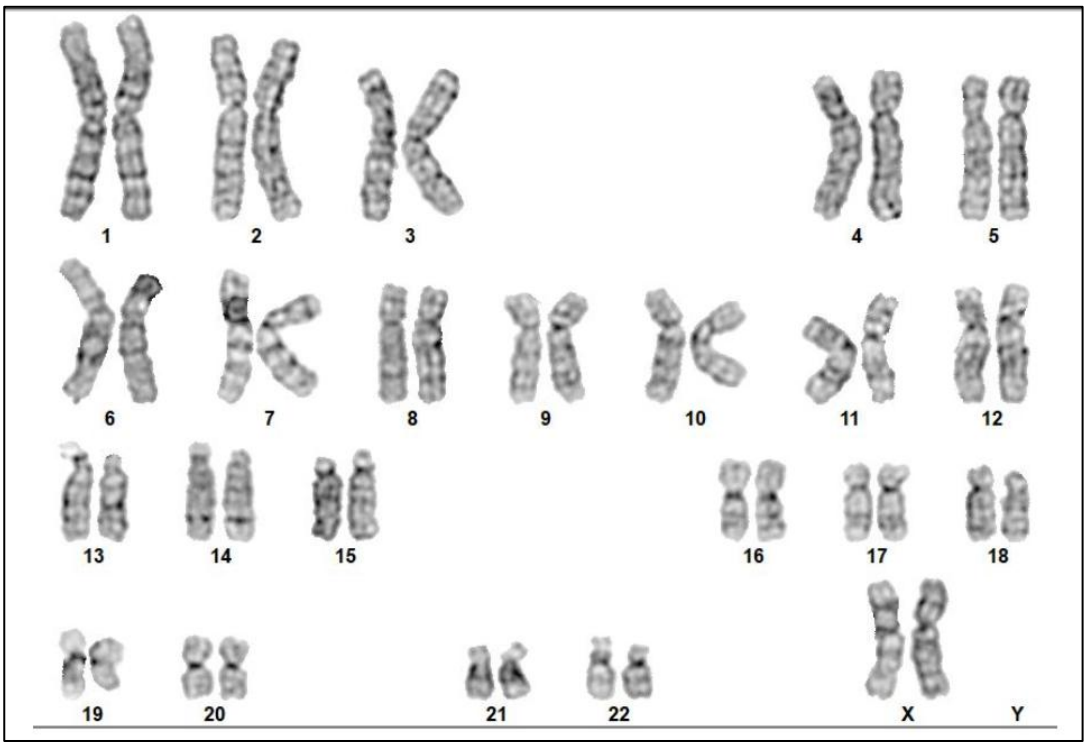

Figure 1. Chromosome results showed no structural chromosomal anomalies in a neonate with 46,XX CAH.

Case 2: 46,XY Adolescent with Progressive Virilization Suspected of PAIS/5α-Reductase Deficiency

A 17-year-and-1-month-old adolescent *raised as* female was referred from Doris Sylvanus General Hospital with a preliminary diagnosis of DSD. Changes in her voice were first noticed by her teacher in the third grade of elementary school (around age 9), but the family only brought the patient for treatment after the physical changes became more pronounced. The medical history revealed no history of menstruation (primary amenorrhea).

Physical examination revealed a physique with masculine muscle mass, facial hair growth, a deep voice, and undeveloped breasts (Tanner stage I). The external genitalia were at *Prader stage* IV, with a 7-cm-long *phallus*, rugated labioscrotal structures and

hyperpigmentation, and bilateral testicles palpable within the labioscrotal fold, each with a volume of 10–12 mL. *The External Masculinization Score* (EMS) was estimated at 4–6 out of a maximum of 12, reflecting significant intrauterine masculinization failure despite functional testes. A pelvic ultrasound confirmed the absence of a uterus, fallopian tubes, or ovaries. Chromosome analysis using G-banding confirmed a 46,XY karyotype. The working diagnosis is 46,XY DSD suspected of *Partial Androgen Insensitivity Syndrome* (PAIS) or type 2 5α-reductase deficiency.

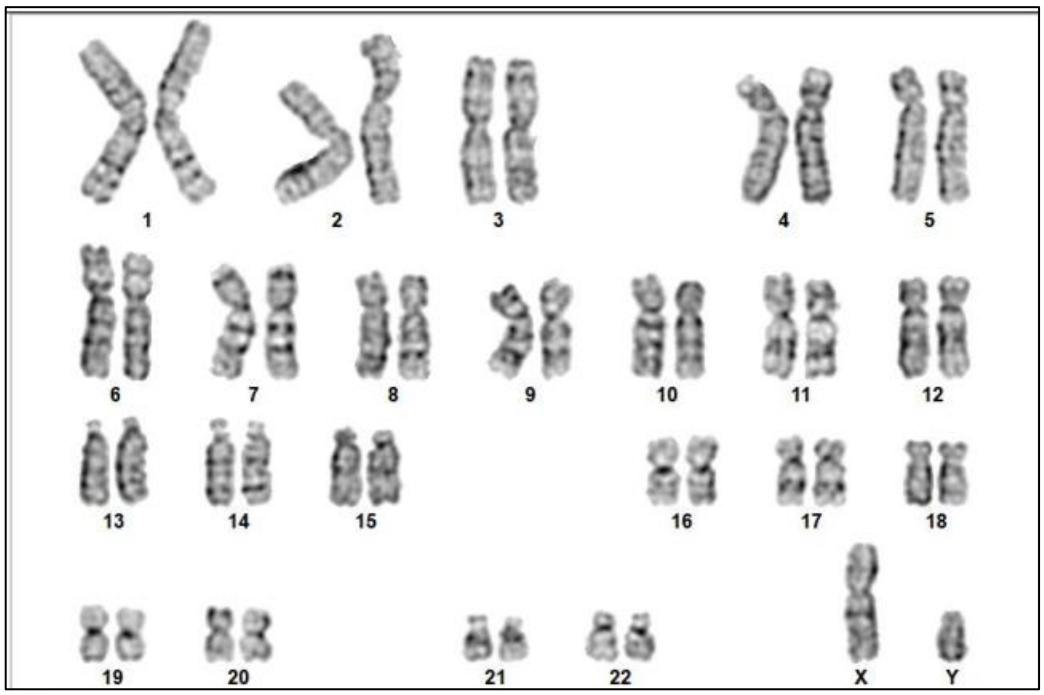

Figure 2. Chromosome analysis of the patient demonstrates a normal 46,XY karyotype without numerical or structural abnormalities, suspected of *Partial Androgen Insensitivity Syndrome* (PAIS) or type 2 5α-reductase deficiency

Case Presentation and Clinical Management

A comparison of the clinical characteristics of the two cases is presented in Table 1

Table 1
Comparison of Clinical, Laboratory, and Supporting Characteristics of the Two Cases

Parameter	Case 1 (46,XX CAH Salt-Wasting)	Case 2 (46,XY Suspected PAIS/5ARD)
Age at diagnosis	10 days	17 years 1 month
Chief complaint	Vomiting, lethargy, poor feeding	Progressive virilization, primary amenorrhea
Degree of virilization	Prader stage III, phallus 1.5 cm	Prader stage IV, phallus 7 cm
Palpable gonads	Non-palpable bilaterally	Bilateral testes 10–12 mL
Sodium (mmol/L)	134 (hyponatremia)	Normal (140)
Potassium (mmol/L)	8.8 (extreme hyperkalemia)	Normal (4.2)
Hormonal testing	17-OHP 237 ng/mL (high)	Not available
Uterus/ovaries	Present (normal ultrasound)	Not visualized (ultrasound)
Karyotype	46,XX	46,XY
Clinical urgency	Acute medical, life-threatening	Long-term psychosocial
Management	Resuscitation + hormone replacement therapy	Counseling + multidisciplinary surgical plan
Outcome	Significant clinical improvement (age 43 days)	Undergoing multidisciplinary evaluation

Source: Processed Primary Data, 2026

In the first case, management began immediately upon the patient's arrival in the emergency department using a structured approach. Severe hyperkalemia (8.8 mmol/L) was corrected with intravenous 10% calcium gluconate for myocardial protection, followed by salbutamol nebulization to promote intracellular potassium redistribution and rehydration with 0.9% NaCl maintenance fluids. Hormone replacement therapy was initiated with an intravenous *stress* dose of hydrocortisone (50 mg/m²), which was gradually switched to oral therapy (25 mg/m²/day in 3 divided doses), plus fludrocortisone 0.1 mg/day and oral NaCl supplementation. Education *on "sick day rules"* was provided to the parents. Clinical outcomes at the 43-day follow-up were very encouraging: potassium decreased to 5.4 mmol/L, sodium was corrected (138 mmol/L), weight increased to 3.8 kg, and the infant appeared active with no complaints of vomiting.

In the second case, management was designed to be gradual and multidisciplinary. Cytogenetic confirmation via G-banding *karyotyping* confirmed a 46,XY karyotype. Intensive psychological counseling was immediately provided to the patient and family to prepare for the complex process of gender identity exploration. Planning for genital reconstruction surgery was conducted in collaboration with the urology team, while awaiting a definitive decision regarding gender selection that actively involved the patient.

These two cases represent two diametrically opposed ends of the DSD spectrum in terms of pathophysiological mechanisms, clinical urgency, and long-term impact. The first case was an absolute medical emergency a delay in diagnosis of just a few days could have been fatal due to ventricular arrhythmia and hypovolemic shock. In contrast, the

second case is a psychosocial emergency that has accumulated over 17 years, in which the patient was physically viable but experienced progressive gender identity distress.

2. Discussion

Clinical Interpretation of Case 1: Adrenal Crisis in a 46,XX CAH Neonate

The first case describes the classic presentation of life-threatening *salt-wasting* CAH from the very first days of life. The clinical triad of persistent vomiting, extreme hyperkalemia (K 8.8 mmol/L), and ambiguous genitalia is a characteristic manifestation of 21-hydroxylase deficiency due to a *CYP21A2* mutation, which simultaneously inhibits the synthesis of cortisol and aldosterone. Steroid precursors accumulating beyond the site of enzymatic blockade are diverted to the adrenal androgen pathway, causing virilization of the external genitalia in female fetuses as early as the 7th week of gestation. This disorder of adrenal steroidogenesis is the most common cause of DSD in 46,XX individuals, and in its classic form is always associated with a risk of life-threatening salt crisis if not promptly recognized (Albayrak et al., 2025).

A 17-OHP level of 237 ng/mL far above the normal range of <10 ng/mL constitutes pathognomonic biochemical confirmation of this diagnosis. The history of the death of the patient's biological older sibling, at 19 days of age with similar symptoms but no clear diagnosis is not merely routine family information; rather, it is concrete evidence of a recurring pattern of autosomal recessive inheritance within a single family, leading to a death that was, in fact, preventable. This addresses the first research question: regional diagnostic barriers directly contribute to *preventable* deaths within the patient's family (Bergougnoux et al., 2023).

The management provided was in accordance with the international guidelines of *the Endocrine Society*. Correction of hyperkalemia with intravenous 10% calcium gluconate and salbutamol nebulization provided immediate myocardial protection, followed by intravenous hydrocortisone at a *stress* dose and fludrocortisone 0.1 mg/day. Follow-up results at 43 days of age potassium levels decreased from 8.8 to 5.4 mmol/L and body weight increased to 3.8 kg demonstrate that with accurate diagnosis and prompt therapy, mortality due to CAH can be completely prevented even in regional facilities with limited resources. A previous report on a 46,XX individual with *Prader stage* IV virilization also documented that an individualized reconstructive surgical approach yields satisfactory functional and cosmetic outcomes when diagnostic planning is systematically conducted from the outset (Peng, Chen, Hu, Zhao, & Qin, 2024).

Clinical Interpretation of Case 2: Delayed Virilization in a 46,XY Adolescent

The second case addresses the second research question: how do regional diagnostic barriers result in irreversible long-term psychosocial consequences. A 17-year-old adolescent raised as a female, with a 46,XY karyotype, functional bilateral testes, absence of a uterus and ovaries, and progressive virilization since age 9 reaching *Prader stage* IV, presents a picture highly consistent with PAIS or type 2 5α-reductase deficiency. A crucial distinguishing feature is overt pubertal virilization manifesting as a deepening voice, muscle mass growth, and *phallic* enlargement which definitively excludes *Complete Androgen Insensitivity Syndrome* (CAIS). In CAIS, total resistance to androgens makes secondary masculinization impossible (Bertelloni, Tyutyusheva, Valiani, & Alberton, 2021).

In PAIS, partial androgen receptor activity allows the body to respond to testosterone to a limited extent, resulting in incomplete intrauterine virilization but the occurrence of pubertal masculinization when testosterone levels surge. In type 2 5α-reductase deficiency, the failure to convert testosterone into DHT causes the external genitalia to appear female at birth; however, the pubertal surge in testosterone compensates for the DHT deficiency, leading to dramatic virilization known as the *Guevedoces* phenomenon. Elevated total testosterone levels with a T/DHT ratio >20:1 following *human chorionic gonadotropin* (hCG) stimulation serve as a key diagnostic marker to distinguish these two conditions; however, this test is not available in South Kalimantan (Stancampiano, Laura, Meroni, Bucolo, & Russo, 2024).

Similar cases of pubertal virilization in adolescents raised as females with a 46,XY karyotype have been previously reported in patients with variants of the *NR5A1*, *SRD5A2*, and *HSD17B3* genes, all of whom were initially not suspected of having DSD because their birth phenotype appeared female. These findings underscore that unexplained virilization in adolescent females should always raise the possibility of a 46,XY DSD diagnosis. Comprehensive evaluation should not be limited to anatomical and hormonal aspects alone, as recent research indicates that karyotype-phenotype mismatch may also carry clinically undetected neurocognitive consequences (Rey, Finkielstain, Vieites, & Bergada, 2021).

Gonadal Management, Cancer Risk, and Policy Implications

The presence of bilateral testes in the second case carries significant implications regarding the risk of gonadal malignancy. In 46,XY DSD with PAIS, the risk of gonadal germ cell tumors (gonadoblastoma or dysgerminoma) is estimated at approximately 15%, determined by the interaction between the genetic material of the Y chromosome in the GBY region, the type of DSD, and the anatomical location of the gonads. If the patient chooses a female gender identity, bilateral gonadectomy is recommended to eliminate the risk of malignancy while halting unwanted masculinization, followed by estrogen replacement therapy. If a male gender identity is chosen, the testes may be preserved via *orchidopexy* with close clinical monitoring and periodic ultrasound (Zhao, Chen, Wang, Zhang, & Li, 2024). This decision should not be rushed, as it concerns the patient's long-term quality of life both physically and psychologically and must actively involve the patient in the process.

Overall, both cases reveal recurring structural barriers that address the third research question: how do regional systemic barriers influence the diagnostic process and treatment outcomes. The lack of 17-OHP testing and genetic molecular analysis at the regional level, the unstable availability of fludrocortisone in the BPJS formulary, and the absence of a permanent multidisciplinary DSD team in South Kalimantan are structural issues that directly determine patients' lives and quality of life. These conditions align with the reality in developing countries, where comprehensive DSD management requires the integration of genetic information with well-defined clinical and endocrine phenotypes something that can only be achieved if diagnostic infrastructure and multidisciplinary teams are available at the regional level (Yavas & Guran, 2024).

Conclusion

This case report presents two contrasting DSD presentations from South Kalimantan. The first case: a 46,XX neonate with salt-wasting CAH survived an adrenal crisis through early electrolyte correction and hormone therapy. The second case: a 46,XY adolescent raised as female showed progressive virilization from age nine due to delayed diagnosis, resulting in a severe psychosocial burden. The preventable death of a sibling confirms that regional diagnostic limitations directly affect patient survival and quality of life.

Three urgent structural barriers must be addressed: (1) unavailability of 17-OHP testing and genetic analysis in regional hospitals; (2) unstable inclusion of fludrocortisone in the JKN formulary; and (3) the absence of a permanent multidisciplinary DSD team. Policy priorities include integrating 17-OHP measurement into the national newborn screening program, ensuring a continuous supply of fludrocortisone and hydrocortisone across all healthcare levels, and establishing regional DSD teams comprising pediatric endocrinology, urology, medical genetics, psychology, and social work. To close existing gaps, critical research steps are building a regional DSD registry to generate prevalence data from outside Java; piloting 17-OHP screening in South Kalimantan maternity units to assess effectiveness before national expansion; conducting qualitative studies on the long-term psychosocial impact of delayed DSD diagnosis in regional Indonesian settings; and characterizing the mutation spectrum of CYP21A2, AR, and SRD5A2 genes in the local population. Without these systematic measures, DSD cases will remain unrecognized or treated too late, leading to avoidable deaths.

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