



# Clinical Presentation and Etiologic Diagnosis of Disorders of Sex Development in Children in Senegal

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## Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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## ABSTRACT

**Introduction:** Disorders of Sex Development (DSD) refer to situations where chromosomal, gonadal or anatomical sex is atypical. We aimed to describe the baseline characteristics on clinical and etiological diagnosis of DSD in Senegalese children.

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**Methods:** This retrospective cohort study over a period of 8 years (2015-2022) included all children aged 0 to 18 years followed for DSD at the pediatric endocrinology department of NCHAR in Dakar. Sociodemographic clinical, paraclinical (genetics, imaging, hormonal) parameters were collected and analyzed with Epi Info 7.2. The description was made using position and dispersion parameters and illustrations in the form of appropriate tables and graphs. Bivariate analysis was used for associations between variables, with an alpha error risk of 5% and a CI of 95%. All ethical rules have been respected.

**Results:** We included 102 DSD cases. Only 61 patients (59.80%) had a genetic diagnosis. Mean age at diagnosis was  $31.2 \pm 46.6$  months. Rearing sex of child was male in 35 cases (57.38%) and female in 22 cases (36.06%) and undetermined in 4 cases (6.56%). Based on karyotype analysis, 31(50.82%) of children had 46,XY DSD, 27 (44.26%) had 46,XX DSD and three (4.92%) children with sex chromosome DSD. Etiologies were dominated by congenital adrenal hyperplasia (81.48%) and androgen insensitivity (38.71%) in 46,XX and 46,XY DSD respectively. External masculinization score  $> 4$ , palpation of a gonad, a phallus length  $> 2.5$ , a single urogenital orifice as well as ultrasound sex were associated with the XY karyotype.

**Conclusion:** Health care professional and medical students should be trained for early diagnosis. A national guideline should be developed for diagnosis and management of DSD during and beyond the pediatric age group, and adapted to evidence and available resources. This can only be achieved if the management of DSD is supported by universal health insurance.

*Keywords: DSD; congenital adrenal hyperplasia; androgen insensitivity; karyotype; Senegal.*

## 1. INTRODUCTION

Disorders of Sex Development (DSD) refer to situations where chromosomal, gonadal or anatomical sex is atypical [1]. The terms 'pseudohermaphroditism', 'intersex', 'sex reversal', that previously described DSD are now considered as derogatory to the patients and the consensus on the management of intersex disorders recommended a new nomenclature [2]. The proposed changes in terminology aim to integrate upcoming advances in molecular genetics in the most recent DSD classification [3].

Due to the lack of clarity of the term, there is no certainty of the incidence of the conditions included. It has been estimated that its individual incidence is approximately 1 in 4,500–5,500 newborns [4] and when considering all genital congenital anomalies, including cryptorchidism and hypospadias, the incidence can be from 1:200 to 1:300 [5]. The incidence of DSD in 46,XY individuals has been estimated in 1 in 20,000 births and the global incidence of DSD in 46,XX individuals (mainly congenital adrenal hyperplasia) is 1 in 14,000–15,000 live births [6], which varies by region due to differences in the frequency of pathogenic variants. Congenital adrenal hyperplasia and mixed gonadal dysgenesis constitute half of all patients with DSD which clinically present with genital ambiguity [7].

Genital ambiguity associated with 46XY DSD is more complex. It can involve many genes

involved in the determination and differentiation of the bipotential gonad, making sometimes difficult the most appropriate choice of sex at birth. This situation remains without diagnosis in about half of the cases, despite the new genetic techniques (exome, Next-Generation sequencing (NGS)) [8]. The management of DSD and its long-term consequences is complex, particularly in terms of options for medical or surgical treatments, fertility and quality of life of patients. [2,3] These difficulties are exacerbated in our resource-limited context where new genetic techniques and several hormonal analyses are often not available. In addition, issues such as socio-cultural environment, the taboo associated with genital ambiguity, late referral to a specialized clinic and lack of training of health care professionals, especially midwives and pediatricians, make diagnosis and management of these patients even more challenging [9,10,11,12,13]. We aimed to describe the baseline characteristics, clinical presentation and etiological diagnosis of DSD in children followed in the endocrine unit of National Children's Hospital Albert Royer (NCHAR) of Dakar, in Senegal.

## 2. METHODOLOGY

This was a retrospective, descriptive analysis of a cohort of patients seen over a period of 8 years (2015-2022). It primarily included all children aged 0 to 18 years followed for DSD at the pediatric endocrinology department of NCHAR in Dakar during the study period. We excluded DSD

with incomplete files, isolated cryptorchidism or isolated micropenis and simple clitoral hypertrophy. Secondly children without genetic analyze (karyotype or gene SRY) were not included.

Sociodemographic parameters (age, rearing sex, sex assignment...), antenatal (ultrasound sex, use of androgens, estrogens or progestins and perinatal history), clinical, (presence of gonads, number of urogenital orifices, phallus length and shape, scrotum appearance), paraclinical (genetics, imaging, hormonal), therapeutic and evolutionary data were collected.

Data were entered via Excel (Microsoft Office 2016) and analyzed with Epi Info software version 7.2. The description is made using position and dispersion parameters and illustrations in the form of tables and graphs according to the type of variables. For descriptive analysis, categorical variables were used to

describe frequency tables and bar charts. Quantitative variables were described in a histogram mean, median, mode, standard deviation and range. Chi-squared tests were used to compare categorical variables and proportions across groups. Continuous variables were compared using student's *t* test and Mann–Whitney *U* test. A *p* value of less than 0.05 was considered statistically significant.

### 3. RESULTS

Of the 31,935 patients seen at the NCHAR of Dakar over 8 years, 512 (0.37%) were seen in the pediatric endocrinology clinic including 120 for DSD as a primary reason for consultation. (23.43% of all patients seen in endocrinology). We excluded 18 DSD patients (5 for incomplete files, 8 for isolated cryptorchidism, 2 for isolated micropenis, 3 for simple clitoral hypertrophy). Among the remaining 102 cases only 61 patients (59.80%) had a genetic diagnosis.

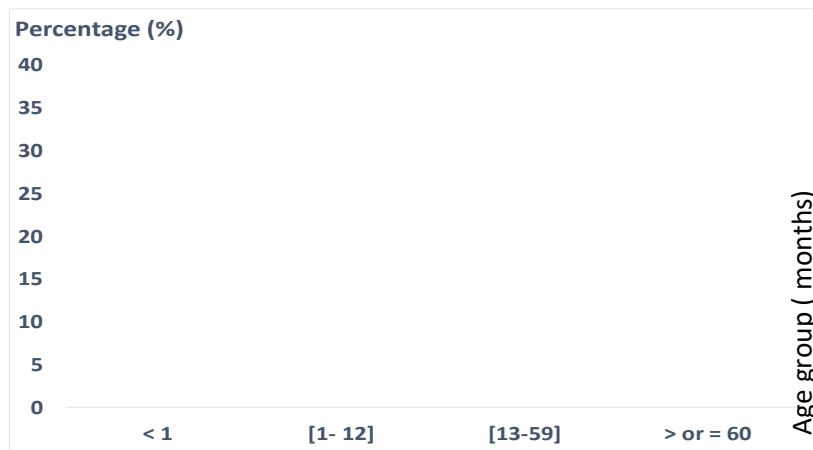


Fig. 1. Age at diagnosis

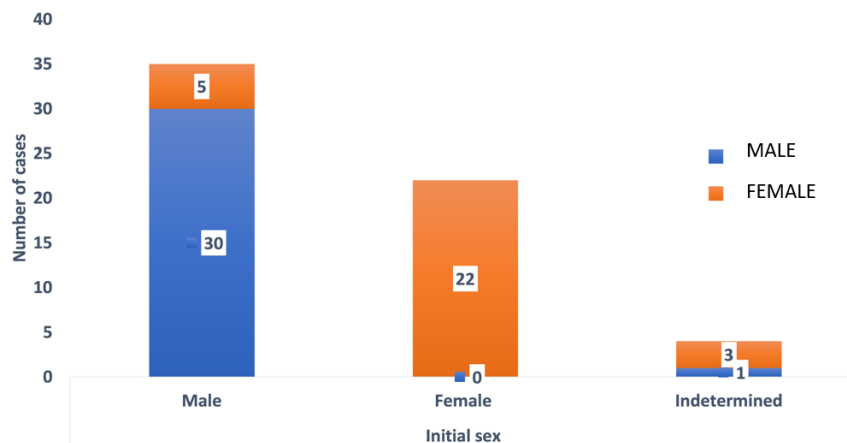


Fig. 2. Distribution of initial and final sex assignments

**Table 1. Clinical findings in 46XY DSD patients (n=31)**

Clinical findings	Number (n)	Percentage (%)
<b>Phallus length (cm)</b>		
< 2.5	14	45.16
>=2.5	17	54.84
<b>Curved phallus</b>		41.93
Yes	13	
<b>Labioscrotal folds</b>		
Rugated	28	90.32
Smooth	3	9.68
<b>Palpated Gonad</b>		
Yes	29	93.54
<b>Single urogenital orifice</b>		
Yes	31	100
<b>Position of urethral orifice</b>		
Hypospadias	16	51.62
Epispadias	1	3.22
Normal	14	45.16
<b>External masculinization score (EMS)</b>		
4	7	22.58
5	2	6.45
5,5	2	6.45
6	16	51.62
8	2	6.45
9	2	6.45

### 3.1 Sociodemographic Characteristics

The mean age at diagnosis was 31.2 ± 46.6 months [range: 2 days-156 months]. Only 18.5% were diagnosed in the neonatal period (Fig. 1), including only 1 case of antenatal diagnosis.

Parental consanguinity was found in 45.5% of cases and taking estrogen-progestins medications during the first trimester in 10% of mothers.

Rearing sex of the child was male in 35 cases (57.38% and female in 22 cases (36.06%). A rearing sex had not been assigned in 4 patients (6.56%). Following clinical, laboratory investigations and a pelvic ultrasound, male sex (n=1) and female sex (n=3) could be assigned to the 4 patients who came to the first consultation without sex assignment. Investigations also led to a change in sex assignment from male to female in 5/35 patients who presented as males at the first clinic (Fig. 2).

### 3.2 Clinical Findings

#### 3.2.1 Patients with 46XY DSD

The urogenital orifice was unique in all cases and at least one gonad was palpated in 93.54% of

cases. The mean external masculinization score (EMS) was 6 ± 2.5 [4–9]. A majority of patients (64.28%) had a score greater than or equal to 6. Other clinical data are summarized in Table 1.

#### 3.2.2 Patients with 46XX DSD

Clitoromegaly (86.67%) and posterior fusion of the labia majora (70%) were the main signs. Prader stages 3 and 4 predominated (Table 2).

### 3.3 Etiologies of DSD

Based on karyotype analysis, 31(50.82%) of karyotyped children had 46,XY DSD, 27 (44.26%) had 46,XX DSD and three (4.92%) children with sex chromosome DSD. In 46,XY DSD cases, etiologies were dominated by androgen insensitivity (38.71%), testicular dysgenesis (19.36%). In 46,XX DSD, congenital adrenal hyperplasia constituted the main etiology (81.48% of cases). Table 3 shows the distribution according to the cause of the genital anomaly.

### 3.4 Predictive Factors of Genetic Sex

External masculinization score > 4, palpation of a gonad, a penile length > 2.5, a single urogenital orifice as well as ultrasound sex were associated with the XY karyotype (Table 4).

**Table 2. Clinical findings in 46 XX DSD patients (n=30)**

Clinical findings	Number (n)	Percentage (%)
<b>Clitoral length</b>		
> = 1.5cm	26	86.67
< 1.5 cm	4	13.33
<b>Labial folds</b>		
Rugated	3	10.00
Smooth	27	90.00
<b>Number of urogenital orifices</b>		
Single	9	30.00
Double	21	70.00
<b>Palpated gonad (n=27)</b>		
Yes	2	6.66
<b>Posterior fusion of labia majora</b>		
Yes	21	70.00
<b>Prader stage (n=27)</b>		
1	8	26.67
2	1	3.33
3	5	16.67
4	16	53.33

**Table 3. Etiologies of DSD (N=62)**

Diagnostic group	Etiologic diagnosis	Number	Relative frequency/ diagnostic group	Relative frequency/ total number
<b>46 XY DSD (n=31)</b>	Androgen insensitivity	12	38.71	19.67
	Testicular dysgenesis	6	19.36	9.84
	5-alpha Reductase deficiency	4	12.90	6.56
	SRY gene mutation	3	9.68	4.92
	CAH	1	3.22	1.63
	Syndromic	1	3.22	1.63
	Unknown	4	12.90	6.56
<b>46 XX DSD (n=27)</b>	CAH	22	81.48	36.06
	Iatrogenic	2	7.41	3.27
	Syndromic	1	3.70	1.63
	Unknown	2	7.41	3.27
<b>Sex chromosome DSD (n=3)</b>	45,XO/46,XY mixed gonadal dysgenesis	1	33.33	1.63
	45, X0 Turner and variants	2	66.67	3.27

CAH= Congenital Adrenal Hyperplasia, SRY= Sex Determining Region on chromosome Y

#### 4. DISCUSSION

Differences in sexual development (DSDs) represent 23.43% of the activities at the NCHAR pediatric endocrinology department with approximately 15 new cases/year. However, this incidence, high compared to what has already been described in sub-Saharan Africa [9,13], does not reflect the reality in our countries. Most patients do not attend our clinics because of the fear of stigmatization. When they consult primary care structures, they are not systematically referred to our service. Diakité et al [13] highlighted these difficulties in his work and

reported that the low rate of referral makes it difficult to develop a national DSD registry in African countries. Furthermore, in our African societies, patients with DSD suffer from numerous taboos and myths that fuel their experience. In rural areas, the birth of a child with a congenital malformation is considered a curse and a shame for the family. The mother is considered primarily responsible for this outcome. These socio-cultural problems partly explain the difficulties in establishing the real incidence of DSD in our countries and many patients remain without diagnosis.

**Table 4. Predictive factors of genetic sex**

		Caryotype XY			
		Yes [n (%)]	No {n (%)}	IC 95%	p
<b>Male rearing sex</b>	Yes (n=35)	30 (85.71)	4 (11.43)	<b>3.34-157.39</b>	<b>0.00*</b>
	No (n= 26)	1 (3.84)	25 (96.16)		
<b>EMS</b>	> 4 (n=39)	24 (61,15)	15 (38,47)	<b>1.13-10.35</b>	<b>0.025*</b>
	<=4 (n=22)	7 (31.82)	15 (68.18)		
<b>Palpated Gonad</b>	Yes (n=30)	29 (96.67)	1 (3.33)	<b>3.91-57.34</b>	<b>0.00*</b>
	No (n=31)	2 (6.45)	29 (93.55)		
<b>Phallus length</b>	>2cm (n=22)	17 (72,27)	5 (22,73)	<b>1.84-20.00</b>	<b>0.001*</b>
	<= 2cm (39)	14 (35.90)	25 (64.10)		
<b>Urogenital orifice</b>	Single (n=37)	31 (83,78)	6 (16,22)	<b>8.13-160</b>	<b>0.00*</b>
	Double (n=24)	3 (12.5)	21 (87.5)		
<b>Ultrasound internal genitalia (n=52)</b>	Male (n=26)	25 (96,15)	1 (3,85)	-	<b>0.00*</b>
	Female (n=22)	0 (0.00)	22 (100)		
	Indetermined (n=4)	2 (50)	2 (50)		

EMS= External Masculinization Score

In 2005, the consensus group of the Lawson Wilkins Society of Pediatric Endocrinology (LWPES) and the European Society of Pediatric Endocrinology (ESPE) 2005 developed a new classification of DSD based on karyotype [14]. However, only around 2/3 of children have access to genetic diagnosis. This poor access to genetic tests is due to their unavailability, their high cost and the absence of health coverage in our populations. In our resource-limited setting, a careful clinical examination together with an abdominal and pelvic ultrasound become the most important factors in sex assignment. In our experience, palpation of a gonad, presence of a single urogenital orifice, penile length greater than 2.5 cm, as well as ultrasound sex were significantly associated with a XY karyotype.

Diagnosis was late at an average age of 31.2 months with only 18.5% of cases diagnosed in the neonatal period. Despite the fact that a large proportion of patients were delivered in hospital, genital anomaly was initially observed by the patient's mother in majority of the cases. This may be because midwives and primary healthcare physicians do not routinely perform a genital examination, are not trained to recognize atypical genitalia. In addition, there is no national guideline for diagnosis and management of DSD. However, compared to what has been described in sub-Saharan Africa, diagnosis has improved. In Ivory Coast Hue et al [9] reported an average age for diagnosis of 20 years, after pubertal development has occurred. Late diagnosis in a 24-year old patient was reported in Senegal by Cissé et al [15]. Diakit  et al in Mali found an average age at diagnosis of 19.5

years [13]. These various studies were not carried out exclusively in pediatric settings and had blamed suboptimal prenatal consultation and delivery care in the health structures to partly explain the absence of diagnosis during the antenatal and neonatal periods.

Based on karyotype analysis, 31 (50.82%) of children had 46XY DSD and 27 (44.26%) had 46XX DSD. There were 3 children (4.92%) with gonadal dysgenesis or Turner syndrome variants. These results are quite close to those found by Amolo et al [16] in Kenya, who also found a predominance of 46XY DSD. In Brazil, De Paula et al [17] reported 408 patients with genital ambiguity: 61.3% with 46XY, 30.4% with 46XX, and 8.3% with numerical or structural abnormalities of sex chromosomes over a 23-year period. In an international register analysis of 649 cases, Cox et al. [18] found that 71% were 46XY, 19% 46XX and 10% had sex chromosomes abnormalities, confirming that DSD with genital ambiguity are more frequent in patients with 46,XY karyotype, due to the complexity of male sexual differentiation [19].

Congenital adrenal hyperplasia (CAH) (81.48% of XX DSD and 37.70% of all cases) was the primary etiology in our study. This is similar to findings in other studies [20, 21,22]. This high frequency of CAH, in a context of absence of systematic neonatal screening and late diagnosis, requires particular attention because of the potentially serious and life-threatening consequences during the first days of life. Therefore, examination of external genitalia must be systematic in the delivery room. The

presence of ambiguous genitalia with absence of a clinically palpable gonad strongly suggest the diagnosis of a 46,XX DSD and CAH. Health professionals must be trained and national guidelines put in place for earlier recognition and management of patients with ambiguous genitalia. This significant proportion of CAH could be linked to strong parental consanguinity. Indeed, consanguinity was found in 45.5% of our patients and was significantly more frequent in CAH. It has been demonstrated that there is a significant link between the existence of parental consanguinity and the occurrence of congenital adrenal hyperplasia[23]. For instance, in contrast, in a study in Kenya, where the rate of consanguinity is low, the commonest presumed cause of ambiguity in patients with 46XX DSD was ovotesticular DSD followed by testicular DSD [16]. Two out of the 3 cases that had a history of parental consanguinity had presumed 46,XX testicular DSD while 1 had 46,XX ovotesticular DSD, suggesting a familial/genetic rather than sporadic occurrence in these cases [16].

In XY DSD, the most common etiology were androgen insensitivity (38.71%) and testicular dysgenesis (19.36%). These findings were similar than that reported by Amolo et al [16] in Kenya who found 31.5% of androgen insensitivity. In general literature, the most common diagnosis of DSD was congenital adrenal hyperplasia, followed by androgen insensitivity syndrome and mixed gonadal dysgenesis [7].

In forty-one (40.20%) of the 102 patients in our study, karyotype could not be performed, while 6 of 61 karyotyped patients (9.83%) did not have an etiological diagnosis established. This finding is lower than that reported in other series [16, 17, 24, 25, 26]. This was due to financial constraints and loss to follow-up of patients. Investigation of DSD is very expensive and out of reach for many patients in the absence of universal health coverage. In addition, molecular genetic testing for DSD is currently not available in Senegal, and sending samples out of the country would still require funding from individual patients since it is currently not financially supported by the government. Four boys had a genotype that was discordant with the rearing gender, leading to sex reassignment at age 3, 7, 15 and 36 months. This reassignment had important psychosocial issues, in a Senegalese context where families are very large. In addition, there is no legal provision facilitating a change of civil status sex

in Senegal. This further highlights the importance of an accurate diagnosis in order to avoid situations requiring a change of sex.

## 5. CONCLUSION

CAH and androgen sensitivity were the commonest cause of DSD in children in Senegal. Financial limitations make investigation and diagnosis difficult. It is therefore necessary for the health authorities to support the management of DSD through universal health care. Health care professionals and medical students should be trained to recognize unusual genitalia, paving the way for earlier diagnosis. A national guideline should be developed for diagnosis and management of DSD during and beyond the paediatric age group and adapted to our context and available resources. The management should be multidisciplinary, focussing on psychological evaluation throughout the care process to improve understanding of diagnosis and avoid stigma.

## CONSENT

Written consent was obtained from the parent or carer prior to inclusion for patients meeting the inclusion criteria.

## ETHICAL APPROVAL

As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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