CLINICAL PRESENTATION AND CHARACTERISTICS OF DISORDERS OF SEX DEVELOPMENT IN KENYAN CHILDREN AND ADOLESCENTS

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Objectives

•To describe the clinical presentation and characteristics of Disorders of Sex Development (DSD) in Kenyan children and adolescents.

Methods

•This retrospective study included 71 patients diagnosed with DSD who presented at ages 0-19 years from January 2008 to December 2015 at the Kenyatta National (KNH) and Gertrude's Children's (GCH) Hospitals.

•The study was carried out in January and February 2016 after written approval from the research and ethics committee of each hospital.

•Patients with non-congenital (acquired) problems of late puberty were excluded.

Results

•The mean age at the time of diagnosis was 2.7 years with a median of 0.4 years (range birth to 17 years).

•Physical measurements showed that 25 participants (39.7%) had weights below -2 SDs, and 20 participants (44.4%) had height below -2 SDs. Sixteen percent (16%) of these children were born prematurely while 53% were from the capital city of Nairobi. One child was managed for severe malnutrition. There was no evidence of abandonment or neglect.

•Out of 16 children, 12 (75%) had normal blood pressure and 4 (25%) were hypertensive. Among the hypertensive patients, one had anorectal malformation and congenital talipes equinovarus while 2 other patients had sex chromosome DSD (Turner variant 48,XYYY/45,X) and disorder of testosterone biosynthesis. Among the 7 patients with CAH, 4 were normotensive.

•Ambiguous genitalia was initially observed by the mother in 32 (51.6%) and by healthcare provider in 28 (45.1%) patients.

•Majority of the patients (41.2%) had an External Masculinization Score (EMS) ranging from 1 to 3, while another 39.7% had an EMS ranging from 4 to 6.

•Thirty-six (50.7%) patients were initially assigned a male gender while 31 (43.7%) reared as females. Twenty-three (23.9%) percent of patients had gender reassignment at final diagnosis.

Clinical presentation of DSD patients

	Frequency	Percent
Presentation with DSD symptoms at birth (n=70)	67	95.7%
Symptoms at diagnosis		
Ambiguous genitalia alone	55	77.5%
Ambiguous genitalia with other symptoms	11	15.5%
Hypospadias with undescended testes	3	4.2%
Male external genitalia with non-palpable gonads	2	2.8%
Total	**71**	**100.0%**

Family history and pregnancy related factors among DSD patients

	Frequency	Percent
Family history of genital ambiguity (n = 58)	6	10.3
Family history of infertility (n = 33)	5	15.2
Family history of unexpected spontaneous changes at puberty (n = 59)	1	1.7
Family history of fetal/ infant deaths (n = 30)	9	30
Maternal drug ingestion during pregnancy (n = 60)	2	3.3
No history of maternal virilization during pregnancy (n = 61)	61	100
Duration of pregnancy (n = 57)		
Term	51	89.5
Preterm	6	10.5
Place of delivery (n = 59)		
Home	9	15.3
Hospital	50	84.7
Birth weight (n = 57)		
Normal	43	75.4
Low<2.5kg	13	22.8
High>4.0kg	1	1.8
Very low<1.5kg	0	0
History of parental consanguinity (n = 24)	3	12.5

Conclusions

•Patients with DSD may present at a wide age range varying from the first day of life to late adolescence.[1,2]

•Ambiguous genitalia was initially observed by the patient's mother in majority of cases despite a high rate of delivery in hospital. This may be due to the fact that midwives and primary healthcare doctors are not trained on this and there is no national guideline for diagnosis and management of DSD. There is a need of intensifying training on DSD at primary and secondary level of healthcare centres.

References

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Sex differentiation, gonads and gynaecology or sex endocrinology

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