ETIOLOGY OF DISORDERS OF SEX DEVELOPMENT IN KENYAN CHILDREN AND ADOLESCENTS

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Objectives

The purpose of this study was to describe baseline data on etiological diagnosis 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

•Thirty-nine (54.9%) children had karyotype testing done.

•The median age (IQR) of children with reported karyotypes and those without was 3.3 years (1.3-8.9) and 8.3 years (3.6-12.1), respectively (p = 0.021).

•Based on the new DSD nomenclature 19 (48.7%) of karyotyped children had 46,XY DSD and 18 (46.2%) had 46,XX DSD. There were 2 (5.1%) children with sex chromosome DSD.

•Among the 71 patients, 10 (14.1%) patients had a diagnosis of ovotesticular DSD based on histology results and 8 (11.3%) were diagnosed with Congenital Adrenal Hyperplasia (CAH) based on the presence of müllerian structures and elevated 17-hydroxyprogesterone levels. One of these patients had salt-wasting CAH.

•A diagnosis of 5α-reductase deficiency was made in 2 patients based on normal testosterone (T) levels, low or normal dihydrotestosterone (DHT) levels and a high T/DHT ratio after human Chorionic Gonadotropin (hCG) stimulation test.

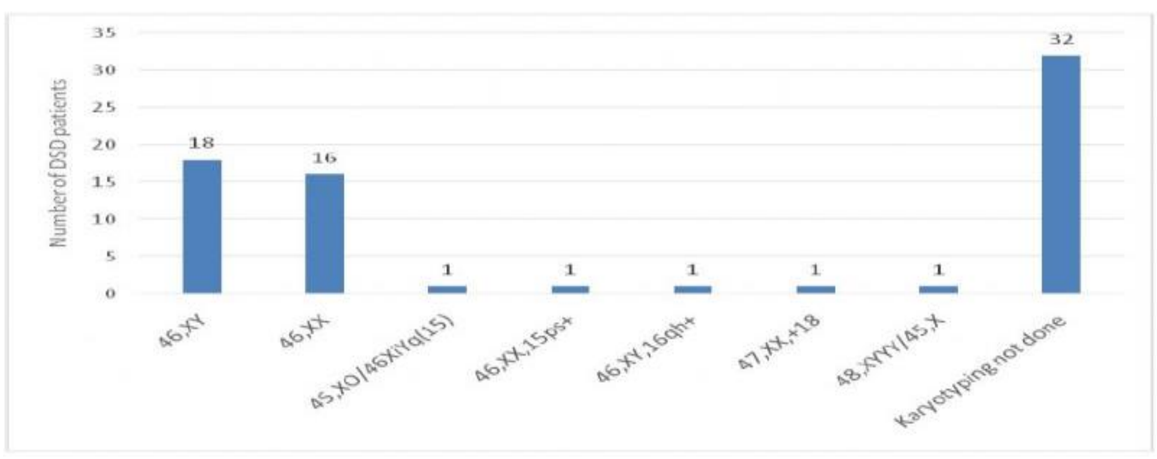
•One patient was diagnosed with Partial Androgen Insensitivity Syndrome based on a 46,XY karyotype, absence of műllerian structures and normal T and DHT response to hCG stimulation.

•Twenty-four patients underwent genitoplasty/ urethroplasty while 9 patients underwent orchidopexy.

•Two patients with ovotesticular DSD who were assigned male gender underwent oophorectomy while one with ovotesticular DSD assigned a female gender underwent bilateral gonadectomy.

•No patient was found to have a gonadal tumour.

Karyotypes



Etiological diagnosis

	Frequency (n)	Percent (%)
DSD etiology		
Ovotesticular DSD	10	14.1
Congenital adrenal hyperplasia	8	11.3
Disorder of androgen synthesis/action	6	8.5
Testicular DSD	5	7.1
Disorder of testosterone biosynthesis	4	5.6
Syndromic associations	3	4.2
5α reductase deficiency	2	2.8
Ovarian dysgenesis (right)	2	2.8
Gonadal regression	1	1.4
Iatrogenic (non-CAH androgen excess in syndromic baby)	1	1.4
Mixed gonadal dysgenesis (45,XO/46,XiYq(15)	1	1.4
Partial Androgen Insensitivity Syndrome	1	1.4
Turner variant (48,XYYY/45,X)	1	1.4
Not established	26	36.6
Total	71	100.0

Conclusions

•The commonest cause of DSD was ovotesticular DSD in contrast to western studies[1,2,3] which found CAH to be more common.

•Investigation of DSD cases is expensive and needs to be supported.

•A network for detailed diagnostics in resource limited countries would be highly desirable.

References

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Sex differentiation, gonads and gynaecology or sex endocrinology

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