Journal of Neonatal Research and Pediatrics Care

Chembio Publishers
Right Choice for Researchers

Research Article *Volume 2; Issue 1*

The Advent of A DSD Team at a Major Teaching Hospital in Nigeria: An Initial Experience in Patient Management

Jarrett OO*[1], Lawal TA[2], Esan OB[3], Adeniji-Sofoluwe AT[4] and Olayemi O[5]

[1]Department of Paediatrics, University College Hospital Ibadan, Nigeria

[2]Division of Paediatric Surgery, University College Hospital Ibadan, Nigeria

[3]Department of Psychiatry, University College Hospital Ibadan, Nigeria

[4]Department of Radiology, University College Hospital Ibadan, Nigeria

[5]Department of Obstetrics and Gynaecology, University College Hospital Ibadan, Nigeria

***Corresponding author:** Dr Olumide Olatokunbo Jarrett, Department of Paediatrics, University College Hospital and College of Medicine, University of Ibadan, Ibadan, Nigeria, Tel No: +2348037203717; Email: tokunbojarret@yahoo.com

Received Date: May 02, 2019; **Published Date:** May 18, 2019

Abstract

Background: Disorders of Sexual Development (DSD) constitute a great challenge in patient management as it involves critical reasoning, careful evaluation, multidisciplinary involvement and making difficult decisions such as sex of rearing and gender issues. The DSD team of the hospital was formed in the year 2010. Our aim is to highlight our initial experience as a DSD team managing these children with DSD in the Pediatric Endocrine/Genetic unit of UCH, Ibadan, Nigeria.

Method: Information was obtained from patients' records and from the Endocrinology Unit register. Details of bio data, clinical history, investigations, treatment and outcome were retrieved.

Results: The team saw 15 patients during the period, aged one day to nine years. Twelve patients (80 %) had pelvic ultrasound scan done to visualize internal organs while karyotype could only be evaluated in 8 (53 %) patients who could afford it. In two patients (13%) that had mutational analysis done while the commonest DSD in the series was XY DSD. There was one death, while one case of XY DSD defaulted.

Conclusion: Despite the intense limitations in terms of inability to get important hormonal and genetic tests done in every case because of financial constraints, the DSD team has continued to function.

Keywords: Gender; Multi-disciplinary team; Ambiguous genitalia

Abbreviations: DSD: Disorder of Sex Development; AG: Ambiguous Genitalia; PAIS: Partial Androgen Insensitivity Syndrome; HCG: Human Chorionic Gonadotropin

Introduction

A newborn infant, in whom development of chromosomal, gonadal and anatomical sex is atypical, is a complex

Citation: Jarrett OO, et al. The Advent of A DSD Team at a Major Teaching Hospital in Nigeria: An Initial Experience in Patient Management. J Neo Res Pedia Care 2019, 2(1): 180013.

problem [1]. A consensus statement recommends the term 'disorder of sex development (DSD)', a generic term encompassing any problem noted at birth where the genitalia are atypical in relation to the chromosome or gonad. The karyotype is used as a prefix to define the category of DSD, replacing the arcane terminology of male and female pseudohermaphroditism [1].

Genitalia are ambiguous whenever there is difficulty in attributing gender to a child based on the appearance of the external genitalia [2,3]. Most DSD associated with ambiguous genitalia (AG) are the result either of inappropriate virilization of girls or incomplete virilization of boys [4]. The birth of a child with AG is a challenging and distressing event for the family and physicians and also comes with life-long consequences [4].

The functional and cosmetic abnormalities as well as medical implications of DSDs confront families of affected newborns with decision issues such as gender of rearing or choice of genital surgery and other concerns such as uncertainty over the child's psychosexual development, fertility options and personal wishes of the child later in life [5]. In general, the assignment of gender of rearing must be guided by the etiology of the genital malformation, the anatomic condition and family considerations [6].

There is paucity of data on DSDs in Nigeria and indeed most parts of Africa. A study in Egypt by Shawkyet et al. involving over 600,000 patients, found a prevalence of genito-gonadal anomalies of 1.34/1000 [7]. Sowande et al. [6] reported on nine cases they had operated on within a 14 year period, while a retrospective study by Ekenze SO et al. [8] in Enugu, south eastern part of Nigeria highlighted the outcome of management of 39 cases at their center. In most of the publications from the continent on DSDs, multidisciplinary team approach to care was not emphasized. Furthermore, there has been no report on the existence of a DSD team in Nigeria or on the impact of such on patient management.

The aim is to highlight our initial experience as the DSD team managing these children at the Pediatric Endocrinology/Genetic Unit of the University College Hospital, Ibadan Nigeria.

Methods

This is a retrospective study of patients seen on account of DSD over a seven-year period (January 2000 to December 2016) by a team formed for that purpose at a tertiary hospital in Nigeria.

The DSD team of the University College Hospital, Ibadan

The DSD team of the hospital was formed in the year 2010 with members from Pediatric Endocrinology, Pediatric Surgery, Psychiatry, Radiology and Obstetrics & Gynaecology. The team is made up of six specialists led by the pediatric endocrinologist who also doubles as the geneticist. The pediatric surgeon and pediatric urologist are fully involved in the decision-making and management especially in patients requiring correction surgery. The team includes a radiologist with expertise in pelvic ultrasonography; and in families requiring psychological support: the psychiatrist/child psychiatrist offers appropriate psychotherapy.

A consultant gynaecologist is also part of the DSD team of the hospital, playing a major role in cases of Ovotestis DSD. The pediatric endocrinologist and pediatric surgeon hold joint DSD clinics bimonthly, reviewing cases together, thus providing expert specialist care and making critical management decisions. This group of experts meets when it is required at case conferences when there are difficult management decisions to be made.

Data collection

Information for the study was collected using the patients' charts and Endocrinology Unit register. For this study, details of bio data, clinical history, investigations, treatment and outcome were retrieved. Results were presented using descriptive summary statistics.

Ethical considerations

The Helsinki principles were adhered to in this retrospective case report study. Caregiver's consent were obtained before data was collected.

Results

A total of 15 patients were seen with DSD between January 2010 and December 2016. The earliest age at presentation to the hospital was on the 1st day of life while the oldest patient at presentation was nine years. Nine of the patients were being raised as boys of which three had no palpable gonads on clinical examination. Six patients were being raised as girls, one of which had a palpable gonad. Pelvic ultrasound scan was the most frequently carried out investigation in these patients and was carried out in 12 (80%) of the 15 patients (Table 1).

Characteristics

Age at presentation	
0 – 28days	4
1 – 12months	5
1 – 5 years	4
>5years	2
Presence of **palpable gonads**	6 (40%)
Pelvic ultrasound scan	12 (80%)
Karyotype	8 (53%)
Mutational analysis syndrome	2 (13%) partial androgen insensitivity syndrome, 5 alpha reductase deficiency
Interventional **surgery**	10 (67%)
Commonest type of DSD	XY DSD
Number of **deaths**	1 (7%) CAH
Number of **defaults**	1 (7%)

Table 1: Characteristics of the 15 patients with DSD

Only two patients had mutational analysis done; one had a genetic mutation involving the androgen receptor, thus a diagnosis of partial androgen insensitivity syndrome (PAIS) was made, while the second had 5 alpha reductase deficiencies. The child with PAIS was born in the labour ward of UCH, hence ambiguous genitalia were picked at birth and the DSD team was notified immediately. The child had ambiguous genitalia; Prader stage 3 with microphallus, the presence of chordae, bifid inguino-scrotal folds, a single urogenital sinus and gonads in the inguinal canals bilaterally (descended into the scrotal folds at age 2 months). Testosterone levels were elevated on the first day of life, while pelvic ultrasound showed male internal genitalia. The child's karyotype was 46XY.

S/N	Sex of rearing	Palpable gonad	Prader stage	Karyotype	Age at presentation	Diagnosis	Genetic testing	Surgery	Pelvic USS
1	Male	Yes	3	46XY	1DOL	PAIS	Yes	Yes	Male
2	Male	No	4	46XY	4months	5α reductase deficiency	Yes	Yes	Male
3	Male	Yes	4	No	4months	Hypospadias	No	male	-
4	Male	Yes	5	46XY	11DOL	?Noonan Syndromic DSD	No	Yes	Male
5	Male	No	5	46XY	9 years	-	No	Yes	Male
6	Male	Yes	3	-	3DOL	Cryptochidism? Cause	No	Yes	Male
7	Female	No	2	-	1 year	Salt losing CAH	No	Yes	female
8	Female	No	2	-	3weeks	Not CAH	No	Yes	No uterus, no ovary
9	Male	Yes	5	-	14 months	Micropenis	No	Not yet	Male
10	Female	No	3	46XX	9 months	Not CAH	No	Yes	-
11	Male	No	3	46XX	9 years	Ovotestes DSD	No	Not yet	Right ovary, right testes,
12	Female	No	3	-	3 years	-	No	Not yet	Right testes, no left gonad, no uterus
13	Male	Yes	3	46XO/46XY	6 months	Ovotestes mosaic DSD	No	Not yet	Uterus, left testes, no right gonad
14	Female	No	3	46XX	1 year +	Not CAH	No	Not yet	Female
15	Female	No	2	-	5 years	-	No	Not yet	-

Table 2: Physical examination and investigation findings in 15 patients with DSD.

In spite of the results of the mutational analysis, which confirmed PAIS and the various counselling sessions, the parents chose to raise the child as a boy, especially since their other two children were girls and also because of infertility issues that would prevail if the child was brought up as a girl; since there would be no uterus. The issue of possible future coital problems due to the microphallus was however lightly considered by the parents.

The child with 5 alpha reductase deficiency is also being brought up as a boy despite having no palpable gonads and the flat response to hCG stimulation test (human chorionic gonadotropin). Table II shows the clinical features, investigation results and intervention in the 15 patients studied. Eight (53%) patients had karyotype done. Based on the karyotype results the commonest DSD in our series was XY DSD. Ten of the 15 patients have had corrective surgery done following parental well-informed decisions. Three patients are on hormonal replacement therapy. A patient defaulted following family dispute and separation, which was triggered by lack of consensus on the part of the parents as regards decisions on patient's management. A mortality was recorded in this series: the only patient with congenital adrenal hyperplasia, salt losing type, who died after a febrile illness, having been off her cortisol for a while and parents did not bring her to the hospital at that time for treatment.

Discussion

The presence of a DSD team at the hospital has engendered an integrated approach to the management of these children despite the intense limitations. The multidisciplinary team can play a critical role in creating a climate of commitment to the health and welfare of the children born with DSDs, as well as to their families [9]. It can make possible the provision of excellent care that has as its goal, the long-term physical and psychological well-being of individuals with DSDs and of their families [10]. Integrated team care allows focus on psychosocial concerns while providing continuity of care in fields that may at some point be needed [9]. It also allows substantial learning among team members and provides a critical mass of families useful for providing local peer support [9]. Meeting others who share similar challenges has been consistently identified by adults and families affected by DSDs as the single most powerful therapeutic experience [11,12]. Each member of the team has specific responsibilities and it is necessary to define these roles to ensure efficiency of the team.

The role of health care professionals in the initial gender assignment is to obtain and help interpret test results concerning the etiology and prognosis of the child's anatomy and physiology, so as to inform the parents' decision about gender assignment [9]. From our experience, in spite of the constraints in healthcare financing prevalent in our society, the minimum requirement for decision making in cases of DSD include: a good clinical assessment, which includes carrying out a proper examination of the perineum; a pelvic ultrasound scan to reveal the internal sex organs and a karyotype. DSD occurs when there is discordance between the chromosomal sex, the gonadal sex and the phenotypic sex. Hormonal assays may not always be readily possible, however, in children with no palpable gonads on physical examination, it is very important to rule out CAH especially a salt-losing type; hence the need to admit the child and ensure appropriate confirmatory tests are done. The birth of a baby with unclear sex is a traumatic enough event for the family as it is almost never expected [13]. There were two cases of ovotestis DSD amongst our patients; one had a karyotype 45XO/46XY. This is an interesting case for which we had a case conference. The child is presently being reared as a boy. The extended family is already blaming the child's mother for this condition, while the decision-making remains a dilemma.

Considerations of sex of rearing will depend on gonadal and genetic sex; social and cultural beliefs of the parents and medical opinion [13]. The cultural beliefs play a major role in the degree of stigmatization usually experienced by affected families and this leads to the tendency for secrecy and the urgency for definitive and corrective intervention. Surgery to provide an individual with a definite unambiguous gender is generally associated with a more satisfactory outcome [13]. Surgical correction, however, can result in scarring to the genital region, leading to complications such as: reduced sexual sensitivity and impaired functioning [13].

None of the children with karyotype 46XY, who had hypogonadism, was considered for female rearing. A male is considered a preferred gender; hence issues like micropenis and future sexual dissatisfaction were not of concern to the parents when compared with the future fertility prospects.

Conclusion

The presence of a DSD team on ground has engendered an integrated approach to the management of these children, despite the intense limitations in terms of inability to get

important hormonal and genetic tests done in every case because of financial constraints.

This is a preliminary report on DSD team activities in UCH, Ibadan. There will be follow up reports on the short and long term outcome of intervention in these children and their families.

Disclosure Statement

The authors have no conflicts of interest to disclose in relation to this paper.

References

1. Hughes IA (2008) Disorders of sex development: a new definition and classification. Best practise and research clinical endocrinology and metabolism 22(1): 119 -134.

2. Guerra-Junior G, Maciel-Guerra AT (2007) The role of the paediatrician in the management of children with genital ambiguities. J Pediatr (Rio J) 83: S184-191.

3. Byne W (2006) Developmental endocrine influences on gender identity: Implications for management of disorders of sex development. Mt Sinai J Med 73(7): 950–959.

4. Wilson JD, Rivarok MA, Mendonca BB, Warne GL, Josso N, et al. (2012) Advise on the management of ambiguous genitalia to a young endocrinologist from experienced clinicians. Semin Reprod Med 30(5): 339-50.

5. Arboleda VA, Sandberg DE, Eriv V (2014) DSDs: genetics, underlying pathologies and psychosexual differentiation. Nature Reviews Endocrinology 10: 603-615.

6. Sowande OO, Adejuyigbe O (2009) Management of ambiguous genitalia in Ile-Ife, Nigeria: challenges and outcome. Afr J Paediatr Surg 6: 14–18.

7. Shawky RM, Elsayed NS, Ibrahim DS, Seifelldin NS (2012) Profile of genetic disorders prevalent in Northeast region of Cairo, Egypt. Egyptian J Med Hum Genet 13(1): 45-62.

8. Ekenze SO, Nwangwu EI, Amal CC, Agugua- Obianyo NE, Onuh AC, et al. (2015) Disorders of sex development in a developing country: perspectives and outcome of surgical management of 39 cases. Pediatr Surg Int 31(1): 93–99.

9. Clinical guidelines to the management of disorders of sex development. A consortium on the management of disorders of sex development by the Intersex Society of North America.

10. Brown J, Warne G (2005) Practical management of the intersex infant. J Pediatr Endocrinol Metab 18(1): 3–23.

11. Kessler S (1998) Lessons from the Intersexed. New Brunswick, New Jersey: Rutgers University Press.

12. Preves S (2003) Intersex and Identity: The Contested Self. New Brunswick, New Jersey: Rutgers University Press.

13. Butler G. Growth hormone deficiency. In Butler G, Kirk J (eds) Paediatric Endocrinology and Diabetes. Oxford University Press Inc, New York (publisher) 2011: 76 – 81. ISBN 978-0-19-923222-2.