E-ISSN: 2616-3470
P-ISSN: 2616-3462

www.surgeryscience.com
2021; 5(2): 139-140
Received: 10-02-2021
Accepted: 23-03-2021

Vita Indriasari
Department of Surgery, Division of Pediatric Surgery, Padjadjaran University, Bandung, Indonesia

Melian Anita
Department of Surgery, Division of Pediatric Surgery, Padjadjaran University, Bandung, Indonesia

Rizki Diposarosa
Department of Surgery, Division of Pediatric Surgery, Padjadjaran University, Bandung, Indonesia

Corresponding Author:
Vita Indriasari
Department of Surgery, Division of Pediatric Surgery, Padjadjaran University, Bandung, Indonesia

How are the characteristics of patients of disorders of sex development in Indonesia?

Vita Indriasari, Melian Anita and Rizki Diposarosa

DOI: https://doi.org/10.33545/surgery.2021.v5.i2c.675

Abstract
Background: Disorders of sex development (DSD) is a congenital condition in which there are disorders of sex development on chromosomes, gonads, and anatomy. The spectrum of these disorders is very broad, so that its incidence and characteristics are difficult to identify. The aim was to describe the characteristics of DSD patients at our institution.
Methods: This is a retrospective descriptive of patients diagnosed with DSD in the period January 2017 - December 2019. Data were taken from medical records including ages, type of DSD and their management.
Results: There were 70 patients with DSD, with a mean age of 6,74 years (range 0-19 years). Sixty four patients (91,42%) were raised as boys, with genotype 46 XY 90,62%, 46 XX 4,68%, 45 XO-46 XY 3,12%, and 47 XXY 1,56%. A total of 6 patients with genotype 46 XX (8,57%) were raised as girls. Four cases were DSD 46 XX (CAH 2,8%), DSD 46 XY was present in 54 cases, while chromosomal DSD was present in 4 cases including ovotesticular type, testicular type, Klinefelter Syndrome, embryogenic testis regression, and persistent Mullerian Tract Syndrome. A total of 2,8% experienced gender reassignment to become girls and 2,8% become boys. There were 78,57% who had undergone masculinized genitoplasty, 7,14% had undergone feminization genitoplasty, 14,28% had not undergone surgery, and 2,8% had not yet determined their gender.
Summary: DSD patients at our institution presented in preschool, with a predominantly 46 XY genotype, most of whom did not undergo gender reassignment, and surgical management was gender-adjusted.

Keywords: DSD, genotype, phenotype, mosaic

Introduction
Disorders of sex development (disorders of the development of the genital organs) is defined as a state of development of the male or female genital organs that is different from normal. This condition can result in abnormalities in the development of sex chromosomes, gonads, or the anatomy of the genital organs [1]. The worldwide incidence of CAH and mixed gonad dysgenesis is 1: 15,000 and 1: 10,000, but varies widely between different populations [2-4]. Cytogenetic Central Registry, the prevalence of XY females was 6.4 per 100,000 births, the prevalence of androgen insensitivity was 4.1 per 100,000 births with the median age at diagnosis was 7.5 years, and the prevalence of XY gonadal dysgenesis was 1.5 per year. 100,000 births with the median age at diagnosis being 17 years. The incidence of DSD varies among ethnic groups with the highest incidence in the population of South Africa [5]. In a study conducted in Palembang where women were the most phenotype found 12 (54.5%) of 22 respondents [6]. In Indonesia there is no data that presents how many incidence rates of patients with Disorder of Sex Development. The spectrum of these disorders is very broad, so that their incidence and characteristics are difficult to identify. The aim of this study was to describe the characteristics of DSD patients at our institution.

Method
This study is a retrospective descriptive study of patients diagnosed with DSD in the period January 2017 - December 2019. Data were taken from medical records including age, type of DSD and management.

Result
There were 70 patients with DSD, with a mean age of 6.74 years (range 0-19 years).

Sixty four patients (91.42%) were raised as men, with genotypes of 46 XY 90.62%, 46 XX 4.68%, 45 XO-46 XY 3.12%, and 47 XXY 1.56%. A total of 6 patients with genotype 46 XX (8.57%) were raised as women. Of the type of DSD, four cases were DSD 46 XX (CAH 2.8%), DSD 46 XY found in 54 cases, while chromosomal DSD was present in 12 cases, including ovotesticular type, testicular type, Klinefelter Syndrome, embryogenic testicular regression, and persistent. Mullerian Duct Syndrome. As many as 2.8% experienced gender reassignment to become women and 2.8% become men. There were 78.57% who had undergone masculinization genitoplasty, 7.14% had undergone feminization genitoplasty, 14.28% had not undergone surgery, and 2.8% had not yet determined their gender.

Discussion

From the data, there were 70 DSD sufferers, with a mean age of 6.74 years (range 0-19 years). According to the Danish Cytogenetic Central Registry, the prevalence of XY female is 6.4 per 100,000 births, the prevalence of androgen insensitivity is 4.1 per 100,000 births with the median age at diagnosis is 7.5 years, and the prevalence of XY gonadal dysgenesis is 1, 5 per 100,000 births with the median age at diagnosis being 17 years [5]. Babies born with DSD should undergo a rapid and thorough evaluation to determine whether sex determination is feasible. It is important to minimize any emotional trauma from family and children. Referral to a structured team of experienced endocrinologists, geneticists, psychologists and pediatric surgeons or urologists with complex disorders is required. Two screening criteria can be used to diagnose an infant as having one of four abnormalities: symmetrical and the presence of a Y chromosome. The three main groups of developmental deviations are responsible for the most common form of DSD in newborns (Table 1). In the first category, there is masculinization of women due to the overproduction of androgenic steroids, causing genital abnormalities requiring highly specialized medical or surgical (or both) management. In the second category, DSD occurs due to a deficiency of androgen production or androgen activity in males. The third category is abnormalities caused by mutations that cause absent, incomplete gonads or asymmetric differentiation of the gonads. It is important to understand the underlying pathophysiology in order to make an accurate diagnosis and to plan optimal management strategies [1, 7-9].

Conclusion

From the results obtained, the DSD patients at our institution came in pre-school, with a genotype of mostly 46 XY, most of whom did not undergo gender reassignment, and surgical management was adjusted according to the child's gender status.

Suggestion

From the conclusions obtained, it can be made regarding further research on the types of measures performed on DSD patients. So that in the future, statistical data can be seen regarding the description of DSD handling in Indonesia.

References

1. Rafael V, Pieretti, Donahoe PK. Chapter 123: Disorders of Sexual Development. Corran Pediatric Surgery. Seventh Edition. Elsevier USA, 2012, 1565-90.
2. Pang SY, Wallace MA, Hofman L, Thuline HC, Dorche C, Lyon IC, *et al*. Worldwide experience in newborn screening for classical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Pediatrics 1988;81:866-74.
3. Skakkebaek NE. Testicular dysgenesis syndrome. Horm Res 2003;60(3):49.
4. Kim KS, Kim J. Disorders of Sex Development. Korean J Urol 2012;53:1-8.
5. Witchel SF. Disorders of Sex Development. Best Pract Res Clin Obstet Gynaecol 2018;48:90–102.
6. Maritska Z, Prananjaya BA, Parisa N, Quardeta RY. Profil Hormon Penderita Disorder of Sex Development (DSD) di RSUP. Dr. Mohammad Hoesin Palembang. Biomedical Journal of Indonesia: Jurnal Biomedik Fakultas Kedokteran Universitas Sriwijaya 2019;(5):2.
7. Gatti JM, Mcelroy TD, Willig L. Differences of Sexual Development. Ashcraft's Pediatric Surgery. Seventh Edition. Elsevier USA 2019, 953-63.
8. Wong YS, Tam YH, Pang KKY, Yau HC. Incidence and diagnoses of disorders of sex development in proximal hypospadias. J Pediatr Surg 2018;53(12):2498-2501.
9. Ameyaw E, Agyei SBA, Hughes IA, Zacharin M, Chanoine JP. Incidence of disorders of sexual development in neonates in Ghana: prospective study. BMJ 2019;104:636-38.