

## Sex chromosomal mosaicism among DSD patients in Indonesia

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### Abstract

Chromosomal Disorder of Sex Development (DSD) is an atypical condition of external genitalia which inappropriate with sex chromosome result. The cytogenetic testing for DSD patients often revealed normal male and female karyotype. In Chromosomal DSD, some have chromosomal abnormalities, such as sex chromosome aneuploidy and mosaicism. Mosaicism is a condition when individual has more than one chromosomally different cell line due to mitotic non-disjunction during post zygotic period. Mosaicism involving different sex chromosome results broad spectrum of DSD phenotype. The aim of this study was to describe external genitalia characteristic and gender of DSD patients with chromosomal mosaicism. This research was a cross-sectional study among DSD patients who were referred to Center for Biomedical Research (CEBIOR), Faculty of Medicine Diponegoro University, Semarang, Indonesia since 2004 until 2017. Disorders of Sex Development patients were undergone physical examination and chromosomal analysis, and 22 patients who had chromosomal mosaicism were recruited. Gender assignment was done in all patients. There were nine males and three females gender of DSD patients with mosaic karyotype involving different sex chromosome (46,XY/45,X) with various percentage of cells affected. All males had severe hypospadias with six of them had bilateral cryptorchidism and two had unilateral cryptorchidism while a female patient had bilateral scrotal gonad. In addition, three males were 46,XY/46,XX had severe hypospadias and one of them had bilateral cryptorchidism. Furthermore, two males with 46,XX/47,XXY had unilateral cryptorchidism, one of them had severe hypospadias, while the other had mild hypospadias. The other results were a male with 47,XYY/46,XY/45,X had bilateral cryptorchidism; a male patient with 46,XY/46,XX/45,X hypospadias and micropenis; a female patient with 47,XXY/46,XY/45,X had labia majora hypoplastic; a male with karyotype 48,XYYY/47,XYY/45,X had severe hypospadias and unilateral cryptorchidism; and a male with 46,XYr/45,X had severe hypospadias. There were various phenotypic external genitalia among male and female DSD patients with sex chromosomal mosaicism. Gender assignment is important for these patients in the first childhood period.

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