

Genetic Counseling and Education

AB004. Genetic diagnosis and experiences in management of disorders of sex development in Indonesia

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Background: Disorders of sex development (DSD) are congenital conditions that present with atypical development of chromosomal, gonadal, or anatomical sex, while in clinical practice this term means any abnormality of the external genitalia. Management of DSD is challenging especially if identified at later age, creating complexities of problems within the families.

Methods: The Sexual Adjustment Team have been established since 1989 that has provided a multidisciplinary approach to the diagnosis, medical and surgical treatment, genetic counseling, and psychosocial support of patients with DSD.

Results: Management DSD from year 2004 to 2015 was

reported here for 617 patients and grouped in 426 cases with 46,XY DSD, 117 cases with 46,XX DSD, and 74 cases with sex chromosome DSD. Most of the patients in the group of 46,XY DSD were unknown male undervirilization (60.09%) followed by congenital adrenal hyperplasia (69.23%) in 46,XX DSD. Genetic diagnoses that underlie DSD were rarely identified. The reasons of this genetic diagnostic disparity were likely because of inadequate availability of advanced molecular laboratories, inadequate knowledge of the pathogenesis of DSD and awareness of the value of a molecular genetics diagnosis for improving care of patients.

Conclusions: The barrier in DSD management was cultural/religion differences, uncommon use of genetic counselor as health care provider and minimal facilities of national health insurance. This multidisciplinary of DSD team is the only team in Indonesia that can be used as a model for other center in Indonesia as well as other developing countries with minimal diagnostic facilities.

Keywords: Disorder of sex development; genetic diagnosis; Indonesia

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