Clinical Genetics

AB031. Disorders of sex development: a clinical profile from Palembang, Indonesia

Ziske Maritska, Siti Rokoyah Rezkylia Sakti, Nyayu Fauziah Zen, Lusia Hayati, Aditiawati Aditiawati

Department of Medical Biology, University of Sriwijaya, Kota Palembang, Sumatera Selatan, Indonesia

Background: Disorders of sex development (DSD) is a state of atypical genitals which raises doubts in assigning gender to the newborns. To date, there are no studies regarding DSD cases in Palembang. This study was aimed to describe the clinical profile of DSD patients at Dr. Mohammad Hoesin Hospital (RSMH), a public general hospital in the center of Palembang.

Methods: This was an observational descriptive study using medical records of 151 DSD patients at the RSMH. Age, sex, clinical findings, family history, and consanguineous marriage were investigated.

Results: Among the study patients, 61% were first evaluated during their childhood year. Most (84.4%) patients were male while 13.2% were female and 2% were undetermined. Majority (52.9%) of the patients had hypospadias followed by undescended testis (29%), vaginal agenesis (9.3%), congenital adrenal hyperplasia (1.98%), pseudohermaphrodite (1.98%), micropenis (1.33%), labial fusion (1.33%), hermaphrodite (1.33%), and buried penis (0.66%). There were 3 (1.99%) patients with a family history of second degree relatives affected with the same condition. History of consanguinity was found in 2 (1.33%) patients.

Conclusions: Most patients were initially assessed during childhood, addressing a challenge to raise awareness towards DSD among the community and health care providers. The patients' phenotypes were varied, assuming due to different causes. However, the absence of chromosomal analysis in Palembang hindered the establishment of DSD diagnosis based on Chicago consensus.

Keywords: Disorders of sex development (DSD); clinical profile

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