

## **Sexual Assignment of Children with Ambiguous Genitalia Based on Cytogenetics and SRY Gene Analysis**

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Ambiguous genitalia is a term used to describe a person whose sex chromosomes, genitalia, and/or secondary sex characteristics cannot be determined as exclusively male or female. Cytogenetics analysis has been used for routine examination for sex determination while SRY gene analysis used for confirmation the presence of Y chromosome. The aimed of this study is to determine sexual assignment of patients with ambiguous genitalia based on cytogenetic and SRY gene analysis.

Peripheral blood was obtained from 20 patients with ambiguous genitalia who referred to gender team of Faculty of Medicine Diponegoro University\_ (FMDU) Kariadi Hospital. Karyotype was established using a G-banding technique in the Molecular and Cytogenetics Laboratory of the Centre for Biomedical Research of FMDU. DNA was extracted from leucocytes of EDTA using the salting out method as described elsewhere. SRY gene was performed by using PCR and gel electrophoresis. Karyotype showed 46,XY in 13 patients, 46,XX in 5 patients and mosaic 45,X/46,XY in 2 patients. PCR analysis for SRY gene was confirmed in all of the patients with Y chromosome. In 3 patients containing Y chromosome had female phenotype while one patient with 46, XX had male phenotype.

Cytogenetics and SRY gene analysis is required for sex determination in patients with ambiguous genitalia. This information is important to provide an appropriate counseling for gender assignment.

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