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Cytogenetic Investigation of Patients with Disorder of Sexual Development Using G-Banding Karyotype and Fluorescence In situ Hybridization

Authors : Riksa Parikrama, Bremmy Laksono, Dadang S. H. Effendi

Abstract : Disorder of sexual development (DSD) covers various conditions with a specific term such as Klinefelter syndrome, Turner syndrome, androgen insensitivity syndrome, and many more. The techniques to accurately diagnose those conditions has developed extensively. However, conventional karyotype and fluorescence in situ hybridization (FISH) are still widely used in many genetic laboratories as the basic method to determine chromosomal condition of DSD patients. Cytogenetic study was conducted on 36 DSD patients in Cell Culture and Cytogenetics Laboratory, Faculty of Medicine Universitas Padjadjaran, Indonesia. Most of the patients referred to the laboratory diagnosed with primary amenorrhea, hypospadias, micropenis, genitalia ambiguity, or congenital adrenal hyperplasia. The study used G-banding technique to acquire complete karyotype and followed by FISH as either confirmation or comparison method. Among 36 patients, G-banding karyotype and FISH results showed that two were diagnosed with 45, X (Turner syndrome); three with 47, XXY (Klinefelter syndrome); five with 46, XX DSD; 22 with 46, XY DSD; and four with 46,XY complete androgen insensitivity syndrome. G-banding karyotype analysis were paired with FISH using X and Y chromosome probe produced similar results. The present analysis showed that FISH is a reliable method to attain a rapid and accurate chromosome analysis result of DSD patients. Nevertheless, conventional karyotype technique is still vital if other condition appeared in DSD patients in order to get more detailed karyotype result which FISH method cannot achieve.

Keywords : chromosome, DSD, FISH, karyotype
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