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AB013. Distribution of azoospermia factor microdeletions in Indonesian infertile males

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Background: Infertility has been a critical clinical problem, which affects 8–12% of couples worldwide. Failure of reproductive system might be result of both genetic and environmental factor. Fifteen percent of male infertility cases are attributed to genetic factors. The most important part of spermatogenesis presents on specific region known as azoospermia factor region (AZF) in Yq chromosome. Microdeletion in the loci is known as one of the genetic causes in idiopathic infertile males. AZFa microdeletion associated with sertoli cell syndrome, while AZFb microdeletion lead to pre-meiotic spermatogenic arrest and AZFc microdeletion caused sperm maturation defect. We studied frequencies of microdeletions of AZFa, AZFb, and AZFc loci in infertile males admitted to CEBIOR, Semarang, Indonesia.

Methods: Total of 36 infertile patients who admitted to CEBIOR were analyzed in this study during period of 2008 to 2017. DNA samples were analyzed for microdeletions of Y chromosome by PCR-screening using several sequences-tagged-site (STS) markers from different region of the AZFa, AZFb, AZFc on Yq chromosome and SRY on Yp as internal control.

Results: The sites of deletion varied among patients. Out of 36 analyzed cases, 10 (27.78%) subjects showed microdeletion of AZF regions. The most common microdeletions were detected in AZFa region (50%), followed by AZFc and AZFb. One patient has microdeletion that include AZFa+b+c region and another patient has AZFa+c microdeletion.

Conclusions: The frequency of AZF microdeletions in this study is higher than other populations (1–15%). PCR based Y chromosome analysis for AZF microdeletion is necessary for genetic counseling prior to providing assisted reproduction technique.

Keywords: Azoospermia factor region (AZF); microdeletion; infertility; CEBIOR; Indonesia

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