

Y-chromosomal microdeletion in idiopathic azoospermic and severe oligozoospermic Indonesian men

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Abstrak

Aim: to detect Y-chromosomal microdeletion in Indonesian men with azoospermia or severe oligozoospermia using multiplex PCR. Methods: we performed 2 multiplex PCR amplifications of the Azoospermia Factor (AZF) region in 71 men. Criteria for including a patient were fulfilled if they presented with azoospermia or severe oligozoospermia, with or without additional abnormalities of sperm motility or of head morphology, raised or normal levels of FSH, normal levels of LH and testosterone, and with no evidence of testicular tumors or other abnormalities. Five men participated as control persons. Results: partial deletion of AZFa was found in 11 men (15.49%), complete deletion of AZFb in 1 man (1.4%), and complete deletion of AZFc in 1 man (1.4%). The unspecific type of deletion was also detected, including the DBY gene in 2 men (2.81%), and partial deletion of both AZFa and AZFb in 2 men (2.81%). No AZF deletion was observed in the control probands. Related to the type of deletion, the AZFa and AZFb deletion showed spermatogenesis arrest in most tubules, while deletion of the DBY gene is associated with the sertoli cell only (SCO) syndrome. Conclusion: the frequency of partial deletion of AZFa was found to be relatively high in our center. The type of deletion is associated with the testicular histology.

.....Tujuan: mendeteksi mikrolelesi kromosom Y pada pria dengan azoospermia atau oligozoospermia berat menggunakan multipleks PCR. Metode: kami menggunakan 2 multipleks PCR untuk mengamplifikasi regio AZF pada 71 pria. Kriteria inklusi adalah pria azoospermia atau oligozoospermia berat, dengan atau tanpa kelainan motilitas atau morfologi sperma, FSH meningkat atau normal, LH dan testosteron normal, dan tidak ada tumor testis atau kelainan lainnya. Lima pria normal berpartisipasi sebagai kontrol. Hasil: delesi parsial AZFa ditemukan pada 11 pria (15,49%), delesi komplit AZFb pada 1 pria (1,4%), dan delesi komplit AZFc pada 1 pria (1,4%). Tipe delesi tidak spesifik juga terdeteksi, yaitu delesi gen DBY pada 2 pria (2.81%), dan delesi parsial AZFa dan AZFb pada dua pria (2.81%). Pada pria kontrol tidak ditemukan delesi. Tipe delesi AZFa dan AZFb menunjukkan gangguan spermatogenesis pada sebagian tubulus, yaitu spermatogenesis berhenti pada fase spermatosit, sedangkan delesi gen DBY menunjukkan gambaran sel sertoli saja (SCO) pada semua tubulus. Kesimpulan: frekuensi delesi parsial AZFa relatif tinggi pada penelitian ini. Tipe delesi menunjukkan gambaran gangguan spermatogenesis yang berbeda pada histopatologi testis.