

O-22(기초) Y Chromosome Microdeletion analysis by Y-linked Specific Gene from Korean Infertile Males

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Background & Objectives: Deletions on the long arm of the Y chromosome (Yq) are frequently associated with male infertility. We developed an easy, rapid and accurate Yq microdeletion diagnosis kit with Y-linked genes and STS markers.

Method: Y-linked 15 STS kit (GYMY-003) was made by 15 Y-linked STS markers consist of 3 multiplex set (A: sY147, sY152, sY158, sY86, SPGY1/ B: sY255, sY138, sY127, sY84, sY14/ C: sY124, sY130, sY242, sY157, sY254). Y-linked 25 Gene kit (GYMY-002) was made by 25 Y-linked genes consist of 5 multiplex set (A: TMSB4Y, SRY, AMELY, RPS4Y2, VCY/ B: ZFY, HSFY2, TBL1Y, SMCY, XKRY/ C: CYorf15A, NLGN4Y, EIFIAY, UTY, RBM1/ D: CYorf15B, TTTY5, TTTY6, TTTY17, DDX3Y/ E: CDY2, BPY2, CDY1, PRY, DAZ). Twenty five genes were located through out Y chromosome. Each set was run as multiplex and reacted at one PCR tube. One hundred forty three patients with non-obstructive infertility and one hundred three fertile men as control were included in this study.

Results: By the Y-linked 15 STS kit analysis showed that 28% (40/143) of patients had Yq deletion. The Yq deletion sites are located at AZFc (70%; n=28), AZFb+AZFc (27.5%; n=11) and AZFa+AZFb+AZFc (2.5%; n=1). Both Y-linked 25 Gene kit and 15 STS kit analyses showed the similar Yq deletion pattern from each patient. Interestingly, one patient has no deletion by STS kit, but DAZ gene deletion by Y-linked 25 Gene kit. Another patient showed no deletion by Y-linked STS kit, but has two Y-linked genes (TTTY5, TTTY6) deletion at AZFc region by using Y-linked 25 Gene kit. There were no Y-linked gene deletion fertile men.

Conclusions: The gene specific deletion without long range of Yq deletion may be important in male infertility, and further analysis of Y-linked genes is required for male reproductive medicine.