Prasopchoke Niamrot 2008: Molecular Diagnosis of Down Syndrome and Sex Chromosome by Real-Time PCR Method. Master of Science (Genetics), Major Field: Genetics, Department of Genetics. Thesis Advisor: Associate Professor Surin Peyachoknagul, Dr.Agr. 76 pages.

In this study, the real-time PCR method was adapted to determine DS status in 3 particular regions of Down syndrome critical region (DSCR), i.e., *DSCR1*, *DSCR3* and *DSCAM*. Sex identification was performed using Sex-Determining Region (*SRY*) on Y chromosome.

The duplex and triplex real-time PCR assay were used to screen 50 DS patients and 50 normal controls by relative quantification using comparative Ct method, while singleplex real-time PCR assay was used to screen 10 DS patients and 10 normal controls by relative quantification using standard curve method. The singleplex, duplex and triplex real-time PCR assays for the detection of DS gave 100 %, 82 % and 74 % accurate results, repectively. As for sex identification, 100% accuracy was achieved. These data suggested that the real-time PCR method can not be totally used to replace the conventional cytogenetic analysis for DS detection. Therefore, further improvement for real-time PCR conditions is needed to get 100 % accurate results for clinical implementation.

However, real-time PCR method is still considered practical for rapid preliminary screening of DS but cytogenetic analysis is also needed for confirmation of a positive result and a double check is required for a negative result.

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