Multiplex PCR based SMA diagnosis using high resolution CE-SSCP for clinical application

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Spinal muscular atrophy (SMA) is a disease caused by the genetic defect in survival motor neuron protein. SMA resulting in death of neuronal cells in the spinal cord and muscle wasting which is incurable, thus the disease causes the infantile or juvenile death. The early diagnosis of SMA should be based on the genotyping of SMN1 and SMN2 gene those encoding survival motor neuron proteins. There are two major causes of SMA, first is the copy number difference between SMN1 and SMN2, and second is the C-to-T nucleotide transition in SMN1 exon 7 +6 position. In this research, the simple diagnostic method for SMA based on the capillary electrophoresis based single strand conformation polymorphisms (CE-SSCP), which can be applied to two major causes of SMA was developed. The PCR primers targeting exon 7 and exon 8 of SMN1 and SMN2 were designed for multiplex and quantitative amplification of each target, thus copy number determination of SMN1 and SMN2 were done. Also, by using high resolution CE-SSCP condition, the single nucleotide transition in exon 7 +6 position of SMN1 was determined successfully.