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Molecular analysis of the SMN gene mutations in spinal muscular atrophy patients in China

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ABSTRACT. Spinal muscular atrophy (SMA) is one of the most common autosomal recessive diseases. Survival motor neuron1 (*SMN*1) is the SMA disease-determining gene. We examined the molecular basis of SMA in 113 Chinese SMA patients. Homozygous exon 7 and 8 deletions in *SMN*1 were detected by PCR-RFLP. Heterozygous deletion of *SMN*1 was analyzed based on variation of the sequencing peak height of the two different base pairs of exons 7 and 8 between *SMN*1 and *SMN*2. Subtle mutation was detected by genomic sequencing in the patients with heterozygous deletion of *SMN*1. In our study, the rate of deletion of *SMN*1 exon 7 and/or 8 was 91.2%; the rate of subtle mutations was 1.8%. We detected the same subtle mutation (p.Leu228X) of *SMN* exon 5 in two patients (one type I, one type III). The p.Ser8LysfsX23 and p.Leu228X mutations accounted for 13 of the 23 families with subtle mutations reported in the *SMN*1 gene of Chinese SMA. This is the first report where the phenotype of SMA-type III is associated with p.Leu228X. We found two subtle mutation hotspots (p.Ser8LysfsX23 and p.Leu228X) of *SMN*1 exons 1 and 5 in Chinese SMA patients. These two mutations have not been reported from America or

Europe. It is proposed that the distribution of subtle mutations of *SMN*1 of SMA is associated with ethnicity or geographic origin.

Key words: Chinese SMA; *SMN* gene; Deletion mutation; Subtle mutation hotspot

