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Centromeric SMN Deletions in Various Congenital Muscular Dystrophies

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Abstract: We studied centromeric SMN gene deletions in various forms of congenital muscular dystrophies. Our study cohort consisted of 48 patients (43 families): 24 with merosin-positive CMD, 18 with merosin-deficient CMD and 6 with muscle-eye-brain (MEB) disease. None of the patients showed deletions of the telomeric gene; however, the deletion frequency of the centromeric gene was 27%. In a multiplex family, the mildly affected sibling, who is ambulant, had a preserved centromeric copy of the SMN gene, whereas in the severely affected sibling the copy was missing. The centromeric copy of the SMN gene may have a contributing role in the phenotype of patients with CMD, as well as other modifying genes or environmental factors.

Key Words: Congenital muscular dystrophies, survival motor neurongene, SMN, deletion, SMA

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