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Prevalence of exon 7/exon 8 deletion in patients with hypotonia and spinal muscular atrophy

Aswathy C G[1], Sankar V H[1,2]*, Sherrin T Alex[1], Santhi S[1] & Haritha Dasaradh[1]

[1]Genetic and Metabolic unit, Child Development Centre, Medical College Campus, Thiruvananthapuram-695011, Kerala, India

[2]Department of Pediatrics, SAT Hospital, Government Medical College, Thiruvananthapuram-695011, Kerala, India

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Spinal Muscular Atrophy (SMA) is a neuromuscular disease due to degeneration of the anterior horn cells of the spinal cord. The estimated incidence of SMA is 1:6,000-1:10,000. The complete deletion of exon 7 of the *SMN1* gene is the hallmark of 95-98% of SMA patients in most population. The first line of investigation for a child or young adult patient suspected to have SMA should be Multiplex ligation-dependent probe amplification (MLPA) testing for homozygous deletion of exons 7 and exon 8 in the *SMN1* gene. In this paper, we report the results of *SMN 1* exon 7 deletion tests in children who attended the Genetic clinic of a tertiary care hospital in Kerala with one or more of the symptoms especially floppy infants, hypotonia, muscle weakness, tongue fasiculations *etc*. *SMN1* exon 7 and exon 8 deletion was confirmed in 58% cases (19) of the total 33 hypotonia patients. SMA Type I, Type II and Type III were 68.4% (13), 21% (4) and 10.5% (2) respectively among the SMA positive cases. Carrier testing of the non-consanguineous parents showed that all parents were heterozygous carriers. Until 2016, the treatment for this disease was supportive only. Recently Nusinersen, Zolgensma and Risdiplam have become available for SMA patients. The carrier testing in parents with previous SMA child history is essential for the implementation of prenatal diagnosis of this disorder in future pregnancies. The paper emphasizes the importance of this rare neuromuscular disease.

Keywords: Hypotonia, MLPA, SMA, *SMN1*, *SMN2*.