Homozygous Point Mutation in a Patient with Spinal Muscular Atrophy Type 1

Isa Abdi Rad[1], Ali Vahabi[1] and Ahad Ghazavi[2]

Abstract
Spinal muscular atrophy (SMA) is an autosomal recessive disease with progressive hypotonia and muscle atrophy resulting from degeneration of the anterior horn cells in the spinal cord and the brain stem nuclei. The molecular diagnosis of SMA is based on the determination of SMN1 (survival motor neuron 1) gene copy number which shows homozygous deletion in approximately 96%
of the patients. Approximately 4% of patients with SMA has no homozygous deletion and retain at least one copy of the SMN1 gene with pathogenic insertions, deletions, or point mutations. And also, very rarely, both alleles are affected with point mutations.
We report SMA type 1 patient with homozygous point mutation [c.549 del C (p.Lys184ser fs 29)] resulting in frame-shift in SMN1
gene. The patient was the second offspring of a consanguineous family whose first child also died due to SMA1. To the best of our knowledge, this homozygous point mutation on the SMN1 gene has not been reported yet.

Keywords
Spinal muscular atrophy; SMN1 gene; Homozygous point mutation