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Schwartz-Jampel syndrome associated with sensorimotor polyneuropathy: Report of three siblings

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Abstract:

Schwartz-Jampel syndrome, (SJS) is a rare disorder characterized by myotonia, joint contracture, facial dysmorphism and growth retardation, we present three siblings (two sisters and one brother) 19,24 and 27 years old from consanguineous healthy parents with SJS. Their clinical features were similar to those previously described. Motor and sensory nerve conduction study (NCS) were compatible with a sensorimotor polyneuro pathy. Myotonic discharges, complex repetitive discharges, myokymic discharges, positive sharp waves and fibrillation potentials were seen on EMG needle examination and MUPs were prominently neurogenic. One of the sisters had mental retardation and hypothyroidism from infancy. Thus, this is the first known report of sensorimotor polyneuropath and hypothyroidism in SJS and the first reported family with SJS from Iran.

Keywords:

Schwartz-Jampel syndrome ، Polyneuropathy ، Hypothyroidism

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