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"Chloride channel Myotonia: Study of five cases "

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Abstract:

Chloride channel Myotonia is a form of channelopathy, and Myotonia is its manifestation. Myotonia may be defined as delayed relaxation of skeletal muscle after its contraction. Decreased chloride conductance across the transverse tubular system, renders the muscle membrane hyper-excitable and leads to repetitive firing, creating Myotonia. Myotonia congenital is another name for chloride channel Myotonia. Myotonia congenital appears in autosomal dominant type called Thomson disease, autosomal recessive type called Becker disease, and a type with sporadic occurrence. Symptoms appear in the first or second decade of life. Repeated muscle contraction, the so called warm up, result in resolution of the Myotonia stiffness. Muscle stiffness and hypertrophy is another finding at physical examination. In this study we report on 5 patients, which had clinical and electrical signs of Myotonia. Muscle hypertrophy and warm up phenomena were present in all cases. CPK measurement of all cases were normal. 2 patients underwent muscle biopsy that showed only atrophy and increased central nuclei. In three cases autosomal recessive inheritance (Becker), in one case autosomal dominant inheritance (Thomsen) and in one case sporadic occurrence was suggested. With respect to successful results of carbamazepine therapy in 4 patients, and being excellent in one of them, we suggest carbamazepine for the first choice of Myotonia treatment.

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top