A novel emerin gene mutation in Emery Dreifuss muscular dystrophy patient with spontaneous chordae tendinae rupture.

Pancheri E¹, Bozzetti S¹, Rimessi P², Macchione F¹, Barillari M³, Venturoli A², Guglielmi V¹, Fortunato F², Tonin P¹, Vattemi G⁴.

Author information

1. Department of Neurosciences, Biomedicine and Movement Sciences, Section of Clinical Neurology, University of Verona, Verona, Italy.
2. UOL of Medical Genetics, Department of Reproduction and Growth and Department of Medical Science, University Hospital S. Anna, Ferrara, Italy.
3. Department of Diagnostics and Public Heath, Section of Radiology, University of Verona, Verona, Italy.
4. Department of Neurosciences, Biomedicine and Movement Sciences, Section of Clinical Neurology, University of Verona, Verona, Italy. Electronic address: gaetano.vattemi@univr.it.

Abstract

Emery Dreifuss muscular dystrophy (EDMD) is an inherited myopathy characterized by early contractures, slow progressive muscle weakness and cardiac involvement. To date at least seven genes have been associated to EDMD with different inheritance patterns, being emerin gene responsible for the X-linked form of the disease. We report a 40-year-old man who was referred for severe gait difficulty. At age 6 years the patient presented with a waddling gate, lumbar lordosis and heel contractures. Both electrophysiology and muscle biopsy were consistent with a neurogenic disorder and he received a diagnosis of spinal muscular atrophy type 3. At the age of 30 the patient developed heart involvement with junctional escape rhythm and, eight years later, had a spontaneous chordae tendinae rupture. A new clinical examination showed severe muscular weakness and atrophy in scapulohumeroperoneal pattern with significant involvement of the lower facial and intrinsic hand muscles and on a second muscle biopsy emerin was absent by immunohistochemistry and by immunoblot analysis. Sequence analysis of EMD gene revealed the presence of a novel mutation represented by an out-of-frame deletion spanning from the beginning of exon 1 to the half of intron 2 (p.Asp6Glyfs*27). Our study expands the clinical and molecular spectrum of X-linked EDMD.