A Novel Mutation Of The EMD Gene In A Family With Cardiac Conduction Abnormalities And A High Incidence Of Sudden Cardiac Death.

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Abstract

BACKGROUND: Emery-Dreifuss muscular dystrophy, caused by mutations in genes such as emerin (EMD) or lamin A/C (LMNA), is a disorder affecting the joints, muscles, and heart, with a wide spectrum of patient phenotypes including muscle wasting and cardiac conduction defects.

METHODS AND RESULTS: Here we report a multi-generation family from the Hunan Province of China. Affected family members displayed an uncommon clinical presentation of serious cardiac conduction abnormalities at an early age and a high incidence of sudden cardiac death along with mild skeletal muscular atrophy and joint contracture. Clinical analysis of affected members provided evidence of X-linked recessive inheritance. Consequently, using Sanger sequencing of X chromosome exomes, we identified a novel duplication mutation (c.405dup/p.Asp136X) in the EMD gene as the cause for the disease in this family. This variant is a novel mutation that has not been previously reported in Pubmed, Clinvar or other cases reported in the Human Gene Mutation Database.

CONCLUSION: Our finding expands the mutation spectrum of Emery-Dreifuss muscular dystrophy and provides a rationale for EMD mutation testing in cases of X-linked inherited cardiac conduction disorders.
disease and sudden cardiac death, even in those lacking pathognomonic neuromuscular features.

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KEYWORDS: Emery-Dreifuss muscular dystrophy; cardiac conduction abnormalities; emerin; mutation; sudden cardiac death

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