A family with Myotonic Dystrophy (DM1) associated with Sudden Death, Long QT and a Brugada-like ECG pattern in different affected relatives.

1Yorkshire Regional Genetics Service, Chapel Allerton Hospital, Leeds;
2Department of Paediatric Cardiology, Leeds General Infirmary;
3Department of Psychology, St James’ University Hospital, Leeds;
4Department of Adult Cardiology, Leeds General Infirmary, UK

Myotonic dystrophy is a multisystem condition inherited in an autosomal dominant manner. In addition to myotonia and muscular dystrophy, affected individuals are also at risk of cardiac dysrhythmias and conduction defects. A 17 year old male died suddenly whilst riding a bicycle having been previously well. Post mortem examination was normal and cause of death was considered to be due to a primary underlying arrhythmia. His parents and 13 year old brother were referred for cardiac screening. His father's resting ECG showed evidence of R waves starting from V1. His echo showed some septal hypokinesia with a normal MRI. Following an ajmaline test, there was asymptomatic sustained VT. His mother had a normal echo, MRI and ajmaline test but had a corrected QT interval of 450ms associated with hypoparathyroidism which resolved with treatment. Initially, his brother had a normal resting ECG and echo but subsequently was found to have a corrected QT interval of 500ms. At the time of cardiac screening his mother and brother were well with no other medical history. However, his father was concurrently being assessed for dysphagia. On further examination, he had frontal balding, facial muscle weakness and myotonia. On direct questioning there was also a history of myotonia in the deceased. Gene testing identified a DM1 gene expansion from leucocyte DNA in the father and brother and in DNA from pathological blocks from the deceased. Long QT and Brugada gene testing (KCNQ1, KCNH2, KCNE1, KCNE2, SCN5A) in the father and brother was normal. This family highlights the variability of cardiac dysrhythmias associated with Myotonic Dystrophy and that gene testing for this condition should be considered in all patients being investigated for Long QT syndrome or with abnormal ajmaline tests.