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

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INTRODUCTION

- 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.

CASE HISTORY

- 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 genetic assessment and cardiac screening.
- The deceased’s mother and brother were well.
- However, his father was concurrently being assessed for dysphagia.
- On direct questioning there was a history of myotonia in the deceased.
- There was a paternal family history of sudden collapse (fig 5.)

EXAMINATION

- The deceased’s father had frontal balding, facial muscle weakness and myotonia consistent with a clinical diagnosis of Myotonic Dystrophy.

CARDDIAL SCREENING

- The deceased’s father’s echo showed some septal hypokinesia with a normal MRI. During an ajmaline test, there was marked QRS widening (fig 2) immediately before commencement of an asymptomatic sustained ventricular tachycardia (fig 3).
- The deceased’s mother had a corrected QT interval of 450ms associated with hypoparathyroidism which resolved with treatment (fig 4). She had a normal echo, MRI and ajmaline test.
- Initially, the deceased’s brother had a normal resting ECG but subsequently was found to have a corrected QT interval of 500ms. His echo was normal.

GENE TESTING

- Gene testing identified a DM1 gene expansion from leucocyte DNA in the father and brother and in DNA from pathological blocks from the deceased, confirming Myotonic Dystrophy.
- Long QT and Brugada syndrome gene testing (KCNQ1, KCNH2, KCNE1, KCNE2, SCN5A) in the father and brother was normal.

TREATMENT

- The deceased’s father was treated with an ICD.
- The deceased’s brother commenced beta-blockers and is considering an ICD.
- Other first degree relatives of the deceased’s father have been recommended to consider DM1 gene testing and cardiac screening.

REFERENCES


SUMMARY OF PEDIGREE

- The incidence of ECG abnormalities in Myotonic Dystrophy is 37-80%, predominantly conduction disturbances (ref 1.)
- There are a number of studies of Myotonic Dystrophy documenting sudden death, a long QT interval or Brugada-type ECG changes as follows:
  - In 406 DM1 patients followed for an average 5.7 years there were 27 sudden deaths, 9 with documented ventricular tachyarrhythmia (ref 2.)
  - 7 of 500 DM1 patient showed a Brugada-type ECG pattern and developed severe ventricular arrhythmias (ref 3.)
  - In a study of 27 Myotonic Dystrophy patients, 33% had a QTc interval of 460ms or over (ref 4.)
- In this family the tachyarrhythmias did not necessarily correlate with the severity of other features associated with DM1.

CONCLUSION

- This family highlights the variability of cardiac dysrhythmias associated with Myotonic Dystrophy.
- Myotonic Dystrophy should be considered in families with an otherwise unexplained sudden arrhythmic death.

Fig 1. Father’s resting ECG
Fig 2. Father’s ECG during ajmaline testing just prior to VT
Fig 3. Father’s ECG post-ajmaline
Fig 4. Mother’s resting ECG