CASE REPORT

Devil is in the detail

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SUMMARY

A 15-year-old girl of Asian origin, hailing from a rural agrarian background, presented with history of multiple episodes of dizziness for 3 years. The episodes were precipitated mostly by emotional and/or physical stress and relieved on lying down, with a few episodes culminating in transient loss of consciousness. As preliminary cardiac and neurological evaluation were normal, she was being treated by the primary physician as a case of probable psychogenic syncope, supported by the consistent association of the episodes with emotional stress. A detailed review of family history revealed that the premature demise of the patient's siblings which were attributed to snakebite and head trauma by the family could have been in reality sudden cardiac deaths. Treadmill test revealed exercise-induced polymorphic ventricular tachycardia confirmatory for the diagnosis of catecholaminergic polymorphic ventricular tachycardia. She was initiated on beta-blocker therapy to which she showed remarkable response.

BACKGROUND

This case is important as it highlights the significance of a detailed history in clinching the diagnosis of a rare but fatal genetic syndrome. Family history of sudden cardiac death, despite its heavyweight contributory value to the diagnosis and prognostication, is often missed as it is masqueraded by the lack of awareness of an unsuspecting patient/family member who provides the history.

The sudden premature deaths of the patient's siblings were perceived and reported as accidental deaths by the family to the primary care physician, resulting in a potentially dangerous delay in the evaluation and treatment of this fatal condition. The patient who was harbouring a symptomatic fatal genetic arrhythmia syndrome was being treated with a presumptive diagnosis of 'psychogenic syncope' corroborated by 'absence of family history of sudden cardiac death', normal ECG, echocardiogram, MRI brain and EEG.

This case opens our eyes to the fact that family history of sudden cardiac death may often be elusive in that sudden deaths may be attributed to presumed natural or extraneous causes by the family, unless associated with explicit chest pain, palpitations or dyspnoea. Hence finer details of family history need to be actively looked into if one were to identify sudden cardiac deaths in the family.

This case brings us back to the classic dictum in clinical medicine—detailed history remains the strongest diagnostic tool—which holds true even in this era of superlative technological advancements. No advanced investigative tool can supercede or replace a detailed history from its pedestals of clinical medicine.

Another learning point to be highlighted from the case is that 'not all symptoms precipitated by emotional stress are psychogenic in origin'. The consistent association of her symptoms with emotional stress prompted the primary physician to cling to the possibility of psychogenic syncope, of course with the deceptive reassurance rendered by advanced investigative tools.

CASE PRESENTATION

A 15-year-old girl of Asian origin, hailing from a rural agrarian background, presented with history of multiple episodes of dizziness for 3 years. The episodes were precipitated mostly by emotional and/or physical stress, and were relieved on lying down. She reported a feeling of fast pounding of the heart preceding some of the episodes. There were a few episodes which culminated in transient loss of consciousness lasting <1 min, not associated with convulsions. Family history revealed that her elder sister died following head trauma at the age of 12 years, and her younger brother at the age of 7 years following snakebite. A detailed evaluation by the primary physician, including resting 12-lead ECG, echocardiogram, EEG and brain imaging, was found to be normal. Hence she was being treated with a provisional diagnosis of psychogenic syncope, supported by the consistent association of the episodes with emotional stress.

Before proceeding with further investigations, the history was reviewed in finer detail focusing on the possibility of sudden cardiac death in any of the family members. A dedicated probing into the details surrounding the deaths of first-degree family members revealed that her 12-year-old sister, who had gone to draw water from a well, was found unresponsive adjacent to a rock beside the well. The family presumed that she must have slipped and sustained head trauma, resulting in her untimely demise. The 7-year-old brother was playing tag with his friends near the bushes when he suddenly collapsed and died. His death was assumed to be due to a possible snakebite as no other cause was found.

INVESTIGATIONS

Clinical examination, baseline ECG (figure 1) and echocardiogram were normal. Also, 24-hour Holter monitoring revealed multiple episodes of polymorphic ventricular tachycardia that coincided with physical exertion. Exercise stress test (figure 2) resulted in induction of polymorphic ventricular

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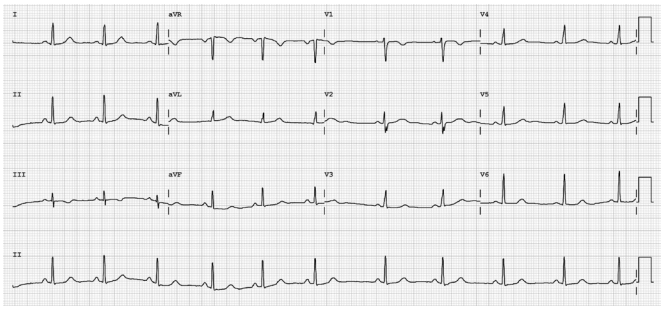

Figure 1 Baseline resting ECG showing sinus rhythm with normal QT interval.

tachycardia without haemodynamic decompensation at 1 min 27 s (4.6 METs) of Bruce protocol. The onset of tachycardia was preceded by increasing frequency of ventricular premature contractions culminating in ventricular bigeminy before progressing to polymorphic ventricular tachycardia. The tachycardia gradually reverted to sinus rhythm at 1 min of recovery without any active intervention. There was no prolongation of QT interval during exercise or recovery. Cardiac MRI and high resolution computed tomography (HRCT) of the thorax ruled out any structural disease involving the heart or mediastinum.

DIFFERENTIAL DIAGNOSIS

A diagnosis of catecholaminergic polymorphic ventricular tachycardia (CPVT) was made, based on the demonstration of exercise-induced polymorphic ventricular tachycardia with no accompanying structural heart disease.

TREATMENT

She was started on oral beta-blocker therapy, following which her exertional palpitations subsided.

OUTCOME AND FOLLOW-UP

Twenty-four-hour Holter monitoring done 2 months later, while on medication, was devoid of premature ventricular activity.

DISCUSSION

CPVT is an inherited arrhythmia syndrome, characterised by polymorphic ventricular tachycardia precipitated by adrenergic stress.[1] Patients with CPVT commonly present with exercise-induced or emotion-induced palpitations, dizziness or syncope. Sudden cardiac death may also be the initial presentation of the syndrome. CPVT has been cited as a cause of sudden infant death.[2] The mean age of presentation is around 6–10 years. In their series of 101 patients with CPVT, Hayashi *et al* reported that about 35% of affected individuals become symptomatic before the age of 10 years and 72% before 20 years.[3] The exact prevalence of this condition is unknown although estimates of 1:10 000 have been reported.[4] The syndrome is characterised by a structurally normal heart and a normal baseline ECG, occasionally with a lower-than-normal heart rate.

Family history of sudden cardiac death is a vital clue suggesting the possibility of the diagnosis of genetic arrhythmia syndromes. However, family history may often be inconspicuous as it is masqueraded by the lack of awareness of an unsuspecting patient or family member who provides the history, as was in our case. The premature deaths of the patient's siblings were perceived and reported by the family as accidental (head trauma and presumed snakebite, respectively) until a detailed in-depth discussion revealed the possibility of sudden cardiac death induced by catecholaminergic stress. This fact emphasises the importance of leaving no stone unturned while obtaining minute details of the history, especially with patients from a rural background, where unexplained death is often attributed to extraneous causes like snakebite.

Exercise or emotional stress induces ventricular tachycardia classically at a heart rate threshold above 100–120 beats per minute. The onset of tachycardia is typically preceded by premature ventricular contractions and short runs of non-sustained VT. The other possibilities to be considered in the setting of exercise-induced VT in a young patient are congenital long QT syndrome type 1, arrhythmogenic right ventricular dysplasia, hypertrophic cardiomyopathy and abnormal coronary artery coursing between pulmonary artery and aorta. The development of bidirectional ventricular tachycardia during exercise is classical of CPVT.[5]

Beta-blockers and lifestyle modification remain the mainstay of therapy.[5 6] Therapy may be guided by exercise stress testing or Holter monitoring. Missing doses of beta-blocker can provoke lethal arrhythmias. Drug compliance needs to be stressed on particularly to the uneducated patient subset. Flecainide can be used as an add-on therapy in those who remain symptomatic while on beta-blockers.

Implantable cardioverter defibrillator (ICD) implantation and left sympathetic denervation are the other modalities of therapy, recommended in patients who experience recurrent syncope or polymorphic/bidirectional ventricular tachycardia despite

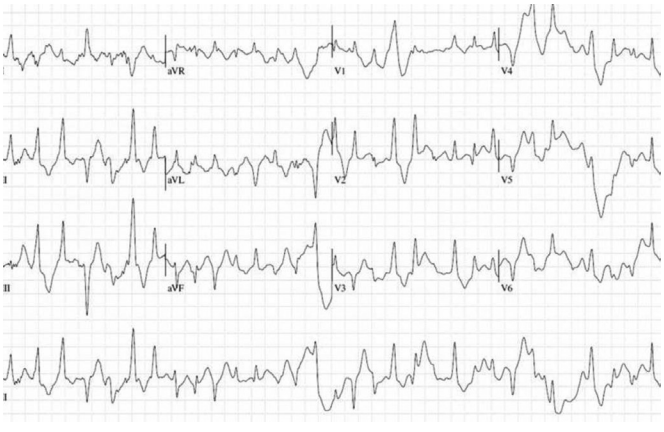

Figure 2 Polymorphic ventricular tachycardia induced at 2 min of exercise stress test with Bruce protocol.

Learning points

- catecholaminergic polymorphic ventricular tachycardia is a rare and potentially fatal genetic arrhythmia syndrome, characterised by stress-induced bidirectional ventricular tachycardia.
- Family history of sudden cardiac death may remain elusive unless actively sought for.
- Awareness of this condition among primary care physicians would avoid potentially fatal delay in prompt initiation of treatment, as well as screening of family members.
- Oral beta-blocker therapy is a readily available, cheap and effective treatment for this life-threatening condition.
- Even in this era of superlative technological advancements, detailed history taking remains the strongest diagnostic tool.

optimal medical management with beta-blockers with or without flecainide. Screening of siblings and parents by clinical evaluation and genetic testing (when a mutation has been detected) is mandatory to identify undiagnosed patients and asymptomatic carriers who are at risk of arrhythmic events. It is suggested that genetically positive family members should receive beta-blockers even after a negative exercise test.[6]

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