Recurrent unexplained syncope in an adolescent girl

Stefano Di Bernado, Tatiana Boulos-Ksontini, Yvan Mivelaz, Nicole Sekarski, Etienne Pruvot

a Paediatric cardiology unit, Department of pediatrics, University Hospital, Lausanne, Switzerland
b Arrhythmia unit, Service of cardiology, University Hospital, Lausanne, Switzerland

Case presentation

This fourteen year-old girl was referred to our outpatient clinic because of recurrent episodes of unexplained syncope. The episodes were of similar presentation. During endurance evaluation at school (the so-called “12-minute running test”), she repetitively developed dizziness, blurred vision and weakness after 8–10 minutes, before passing out for an unusual duration of about 6–8 minutes. After spontaneous recovery,
What are the most likely diagnosis and optimal therapy?

Comments

Based on the observation of runs of nonsustained VT and premature beats of two morphologies, the diagnosis of catecholaminergic polymorphic ventricular tachycardia (CPVT) or LQTS with normal QT interval [1] was highly suspected. Sport was prohibited and beta blocker therapy was initiated first with tapering of propranolol, followed by bisoprolol therapy because of fainting, dizziness and Raynaud phenomenon. Based on the severity of the recurrent syncopal events and as well as persistence of runs of nonsustained VT despite prescription of beta blockers, an implantable cardioverter-defibrillator (ICD) was implanted. The patient remained asymptomatic for months under treatment. Thirteen months after the ICD implantation, a first presyncopal episode occurred during a stressful mathematical test at school. The first nonsustained polymorphic VT episode lasting 24 beats (coupling interval as short as 150 ms) was recorded by the ICD, that was suggestive of an under dosage of the beta blockers. Medication was changed for metoprolol 25 mg bid. Thirty-three months after the implantation, the patient forgot her medication one night and the next morning, while standing quietly, she presented a near death experience suggestive of severe brain hypoperfusion. Apart from these symptoms, no other signs e.g., palpitations or chest pain were noted. Witnesses described the patient as pale and sweaty during the episodes.

Figure 1 shows her ECG at rest. A normal sinus rhythm is observed at 67/min with normal PR interval (138 msec), QRS duration (85 msec), QRS axis (90), and QTc interval (420 msec), and no sign suggestive of ventricular hypertrophy or repolarisation disorder. The echocardiography demonstrated a normal heart as well with left ventricle and right ventricle dimensions and functions within limit. A cardiac MRI with late enhancement allowed us to exclude a congenital coronary anomaly, myocardial scars and signs of arrhythmogenic right ventricular dysplasia.

The 24-hour Holter recording revealed several episodes of nonsustained arrhythmia without any symptoms (fig. 2 and 3). Figure 2 shows a tree-derivation Holter recording with short-coupled single ventricular premature beats, doublets and triplets interspaced with sinus beats during daily activity. Figure 3 and 4 also shows a ventricular premature beat of a different morphology (arrow) that occasionally occurred during sinus tachycardia. Importantly, these episodes occurred only when the adolescent was involved in some physical activity or during stressful tests at school, while her rhythm was normal at night or at rest.
syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.

CPVT occurs in patients without any evidence of heart disease. This disorder typically begins in childhood or adolescence. Affected patients present with syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.

CPVT occurs in patients without any evidence of heart disease. This disorder typically begins in childhood or adolescence. Affected patients present with syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.

CPVT occurs in patients without any evidence of heart disease. This disorder typically begins in childhood or adolescence. Affected patients present with syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.

CPVT occurs in patients without any evidence of heart disease. This disorder typically begins in childhood or adolescence. Affected patients present with syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.

CPVT occurs in patients without any evidence of heart disease. This disorder typically begins in childhood or adolescence. Affected patients present with syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.

CPVT occurs in patients without any evidence of heart disease. This disorder typically begins in childhood or adolescence. Affected patients present with syncope that spontaneously resolved within seconds. Nonetheless, she was transferred to the hospital. Figure 5 shows the interrogation of the ICD at time of presyncope. A nonsustained polymorphic VT of 29 beats compatible with Torsades-de-Pointes is visible. Genetic analyses did not find the typical mutations associated with LQTS1-3 or CPVT.
life-threatening VT or ventricular fibrillation occurring during emotional or physical stress. The VT morphology may vary continuously, from beat to beat, or may appear as a bidirectional VT [2]. Two mutations have been identified in patients with CPVT: the cardiac ryanodine receptor gene (autosomal dominant form) and the calsequestrin 2 gene (autosomal recessive inheritance) [3]. Both proteins play a major role in the regulation of cardiomyocyte’s intrasarcoplasmic Ca2+. Treatments associate an ICD to terminate sustained arrhythmias and to prevent syncope and/or sudden cardiac death, and antiarrhythmic medication in order to prevent arrhythmias and minimise ICD shocks [4]. Beta blockers form the cornerstone of treatment, in patients with ongoing arrhythmias despite therapy with a beta blocker, the addition of verapamil or flecainide may be effective. In refractory cases, left sympathetic denervation can be an alternative therapeutic option [4, 5].

Syncopal episodes are usually benign in nature, in children. The evaluation of syncopal children or adolescents relies on a thorough, detailed history and physical examination. Syncopal episodes that are associated with exercise or sport have to be thoroughly evaluated for their potential danger [6, 7].

References


