Case Report: A Late Diagnosis of Catecholaminergic Polymorphic Ventricular Tachycardia

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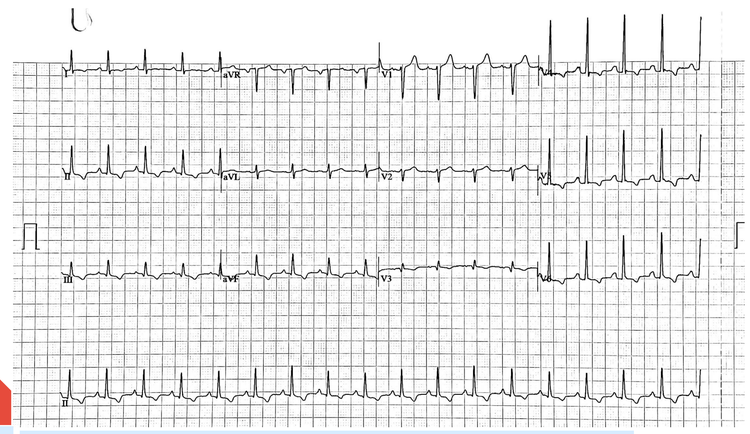
Introduction

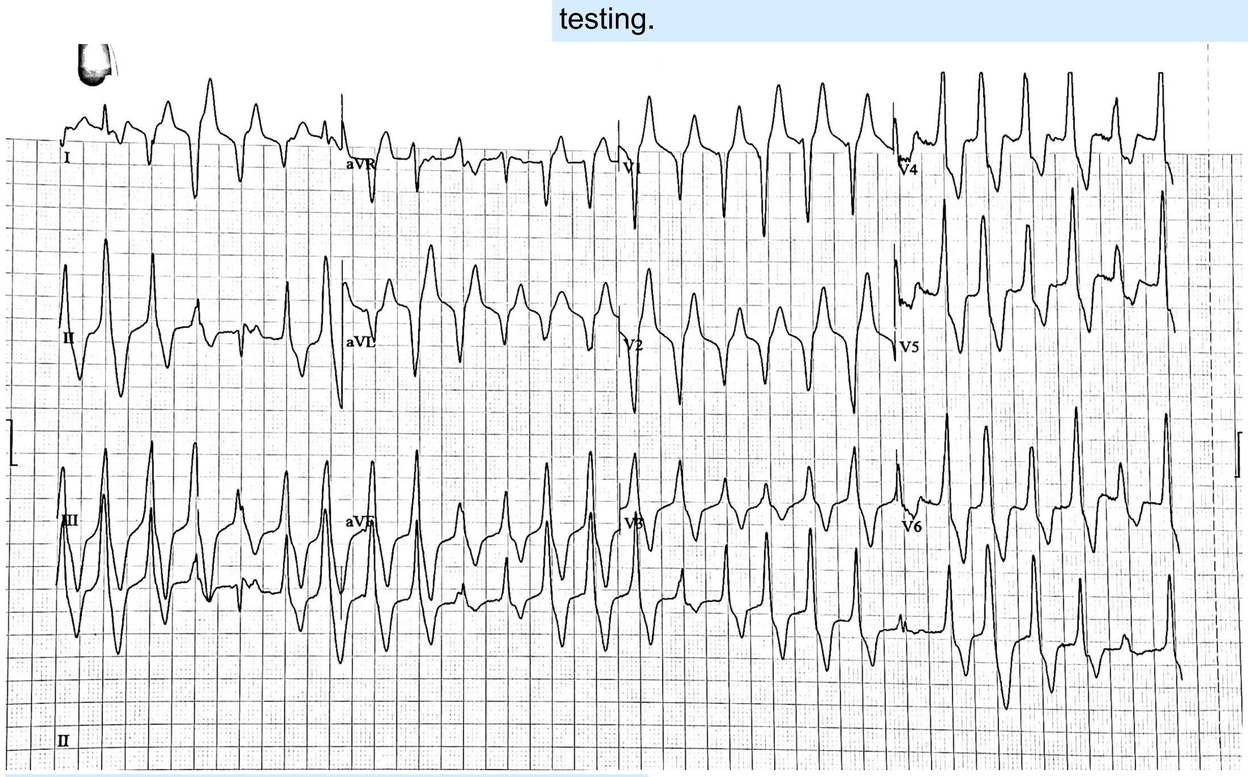
- Catecholaminergic polymorphic ventricular tachycardia (CPVT) is an inheritable mutation of cardiac Ca^{2+} receptors leading to a potentially fatal arrythmia if not properly treated. (3)
- CPVT's exact prevalence is unknown, however it is estimated at 0.010%. (1) Mortality ranges from 30-35% if left untreated. (4)
- This bidirectional or polymorphic arrythmia is most often precipitated by physical stress or emotional/adrenergic stress in the absence of CAD or any other underlying cardiac defects. (2,4,5)
- RYR2 and CASQ2 are the two most common genetic mutations to induce CVPT. (2)
- In most cases, CPVT is diagnosed between 7-12 years of age with rare cases being diagnosed in the fourth decade. (1)
- We present the case of a 48-year-old male who presented with pre-syncopal episodes who, during exercise stress testing, was noted to have CPVT.

Case Presentation

Our patient was a 48 y/o male with a past medical history of type two diabetes mellitus and hypertension who presented with episodic "forceful heartbeats" of two-weeks duration with associated presyncope and a 5-10 second period of syncope while driving. He experienced shortness of breath and left axillary discomfort that resolved on its own after 2 minutes. An echocardiogram revealed no significant wall motion abnormalities with preserved left ventricular systolic function. Exercise stress test was performed. Resting EKG revealed normal sinus rhythm and QTc 415 msec. At 5 minutes into exercise, the patient began to experience lightheadedness and reported that he is going to pass out. Stress EKG showed polymorphic ventricular tachycardia. At this time, exercise was stopped. His symptoms improved and arrhythmia resolved 3-4 minutes after cessation of exercise. Post stress EKG revealed a normal QTc. The patient was started on metoprolol tartrate and left heart catheterization was performed the following day showing no obstructive CAD. He was transitioned to nadolol for potential CPVT with no further episodes of VT reported by the patient.



Pre-exercise EKG



Exercise EKG

Discussion

The first case of CPVT was documented in 1975. (1) Since that time, two inheritance patterns for the Ca2+ transporters/binders responsible for CPVT have been idenitified. The most common mutation in the RYR2 receptor (up to 60% of cases) is autosomal dominant, while the autosomal recessive mutation of CASQ2 is less frequent (2-5% of cases). (3) The cardiac myocyte contraction is reliant on Ca released from the sarcoplasmic reticulum. (5) The RYR2 receptor plays an integral part in this process. The pathogenesis of the RYR2 receptor's role in CPVT is debated as there are many different mutations, however a top hypothesis is it allows Ca to release prematurely during catecholaminergic stimulation. (1,4) This generates an aberrant depolarization current which begins this arrythmia. (4,5) CASQ2 is available in the sarcoplasmic reticulum to bind Ca. (5) A CASQ2 mutation allows for increasing Ca release during normal cardiac diastole. (2) The treatment for CPVT is beta-blockers. Propranolol, nadolol, and metoprolol are the most studied. (4) For RYR2 receptor mutations only, class Ic antiarrhythmic drugs (flecainide and propafenone) have shown reduction in patients with CPVT, in the absence of structural heart disease. (2) ICD implantation is recommended to thwart arrythmias for patients still having arrythmias while on optimal medical therapy. (4)

Conclusion

Catecholaminergic polymorphic ventricular tachycardia can lead to sudden cardiac death if not promptly diagnosed and treated appropriately with proper medications. To this date this patient has had no episodes of CPVT since addition of nadolol, which is confirmed by a 2-week cardiac event monitor. Follow up to come as our patient is scheduled for cardiac MRI as well as genetic testing.

References

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