ESC European Society of Cardiology

European Heart Journal - Case Reports (2021) **5**(11), 1–3
https://doi.org/10.1093/ehjcr/ytab458

Syncope in a young male

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Received 4 October 2021; first decision 27 October 2021; accepted 9 November 2021; online publish-ahead-of-print 23 November 2021

Clinical vignette

An 18-year-old man presented to the emergency department with a 2 day history of dizziness and palpitations. On further questioning, he reported two episodes of sudden loss of consciousness in the preceding 6 months, one while driving the car and one while walking. He remembers blacking out in secondary school. He was otherwise well. Initial vital signs were heart rate of 60 beats/min and blood pressure of 120/70 mmHg. His medical history was significant for polysubstance use: a bottle of vodka twice a week, cigarette smoking of 15/day, marijuana use (unspecified amount), cocaine 3 g/day, and a previous history of benzodiazepine use for 2 years. There was no intravenous drug use or other personal cardiac history. There was no family history of sudden cardiac death or any known cardiac genetic syndrome although the patient did not know his father. He was admitted to the inpatient Cardiology service. An echocardiogram revealed a septal thickness of 12 mm and mild global systolic dysfunction with an ejection fraction of 47%. There were no valvular abnormalities. On telemetry, he was noted to have paroxysmal episodes of ventricular standstill, the longest lasting 6 s resulting in a medical emergency response.

Question 1

What is the likely diagnosis?

A. Hypertrophic cardiomyopathy
B. Danon disease
C. Fabry disease
D. PRKAG2 syndrome
E. Cardiac sarcoidosis

Question 2

What is the likely cause of the rhythm seen?

A. Sinoatrial exit block
B. Vagal-mediated response
C. Atrioventricular block
D. Hyperkalaemia
E. Post-tachycardia reversion pause

Question 3

How would you manage this patient?

A. Tilt table testing
B. Administration of oral theophylline
C. Exercise stress testing
D. Permanent pacemaker implantation
E. Accessory pathway ablation

Explanation 1

The correct answer is D. This is a rare, autosomal dominant condition definitively diagnosed by identifying the causative mutation in the gene encoding the 5′ AMP-activated protein kinase (AMPK) which is involved in cellular ATP metabolic regulation. Basic research on mice postulates that AMPK dysfunction causes glycogen myocyte deposition which directly correlated with ventricular pre-excitation. However, the precise biological mechanisms between PRKAG2-related enzyme dysfunction and different phenotypic manifestations in human is yet to be elucidated.[1] The clinical presentation varies from no symptoms to sudden cardiac death. This patient's electrocardiogram is suggestive, showcasing the typical triad of findings:[1] (i) ventricular pre-excitation, (ii) unexplained left ventricular hypertrophy, and (iii) conduction disease, usually high grade atrioventricular block. Hypertrophic cardiomyopathy is typically associated with tachyarrhythmias, not bradyarrhythmias.[2] Danon disease is almost always associated with cognitive disabilities and commonly with skeletal myopathy.[3] Patients with Fabry disease have a severe functional alpha-galactosidse A enzyme deficiency resulting in other clinical presentations such as neuropathic pain, dermatological skin lesions, gastrointestinal symptoms, and renal failure.[4] While left ventricular hypertrophy and pre-excitation are seen, atrioventricular block is uncommon. Cardiac sarcoidosis is associated with atrioventricular block, which tends to be consistent rather than paroxysmal.

Explanation 2

The correct answer is C. Sinoatrial exit block is due to failure of the sinoatrial node impulses to propagate, resulting in dropped P-waves. In this case, there are many P-waves but an absence of associated QRS complexes. This leads to the diagnosis of atrioventricular block

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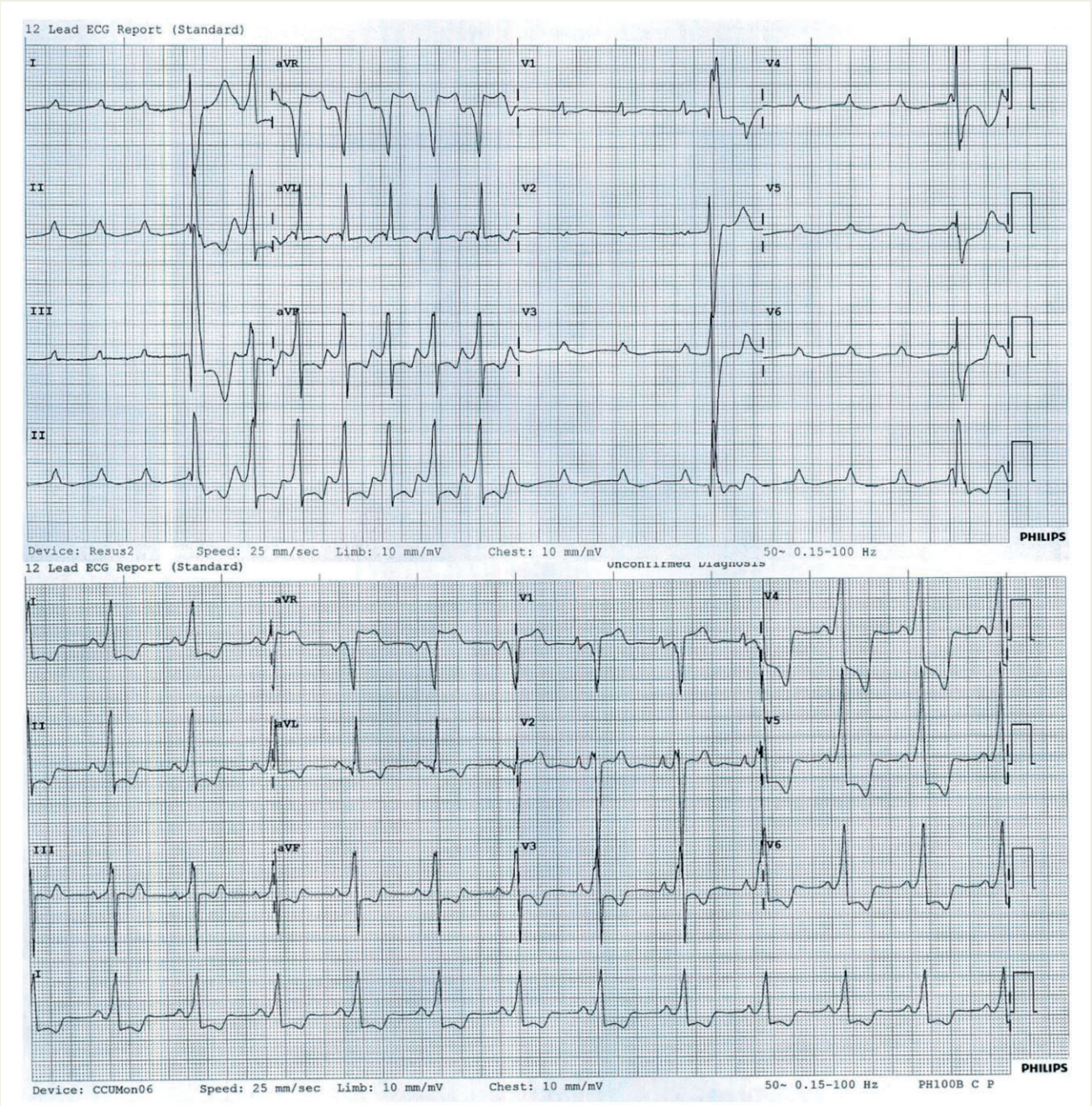
Handling Editor: Parag Ravindra Gajendragadkar



where each atrial depolarization does not produce a ventricular depolarization. Hyperkalaemia does not give *intermittent* blocks as demonstrated in this electrocardiogram. A post-tachycardia reversion pause is defined by *no* electrical activity during the pause due to suppressed sinus automaticity by the tachycardia (usually atrial fibrillation), as opposed to non-conducted P-waves.

Explanation 3

The correct answer is D. A pacemaker will prevent future syncopal episodes by being able to pace the ventricle when atrioventricular block occurs. Tilt table testing is indicated for the diagnosis of vasovagal syncope, but this patient's syncope sounds cardiac. Moreover, the non-conducted P-waves do not show sinus slowing which goes against vasovagal syncope. Theophylline may be effective in syncope related to low plasma adenosine levels; however, this syndrome is not associated with increased left ventricular wall thickness or pre-excitation. Exercise stress testing is indicated for investigating ischemia and is relatively contraindicated in this scenario as a period of atrioventricular block during exercise could lead to serious injury. Accessory pathway ablation would only be useful if the cause of his

syncope was a tachyarrhythmia that used the accessory pathway, precipitating rapid ventricular rates.

Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

Conflict of interest: None declared.

Funding: None declared.

References

1. Porto AG, Brun F, Severini GM, Losurdo P, Fabris E, Taylor MRG et al. Clinical spectrum of PRKAG2 syndrome. *Circ Arrhythm Electrophysiol* 2016;**9**:e003121.
2. Savage DD, Seides SF, Maron BJ, Myers DJ, Epstein SE. Prevalence of arrhythmias during 24-hour electrocardiographic monitoring and exercise testing in patients with obstructive and nonobstructive hypertrophic cardiomyopathy. *Circulation* 1979;**59**:866–875.
3. Boucek D, Jirikowic J, Taylor M. Natural history of Danon disease. *Genet Med* 2011;**13**:563–568.
4. Branton MH, Schiffmann R, Sabnis SG, Murray GJ, Quirk JM, Altarescu G et al. Natural history of Fabry renal disease: influence of alpha-galactosidase A activity and genetic mutations on clinical course. *Medicine (Baltimore)* 2002;**81**:122–138.