Managing Arrhythmogenic Right Ventricular Cardiomyopathy: Addressing the Challenges

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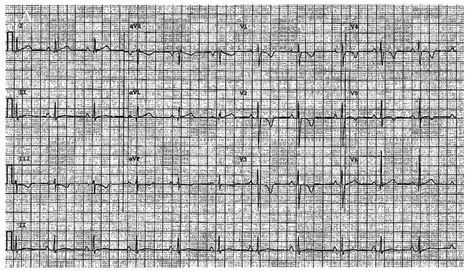
Background

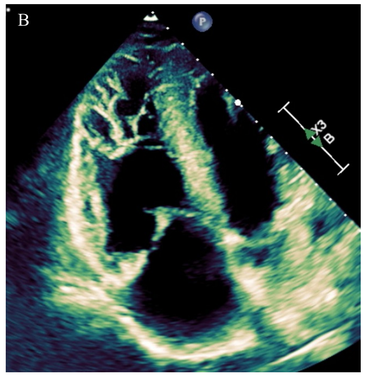
- Diagnosing arrhythmogenic right ventricular cardiomyopathy (ARVC) is a complex task
- We present a case of unclear ARVC, leading us to explore alternative possibilities

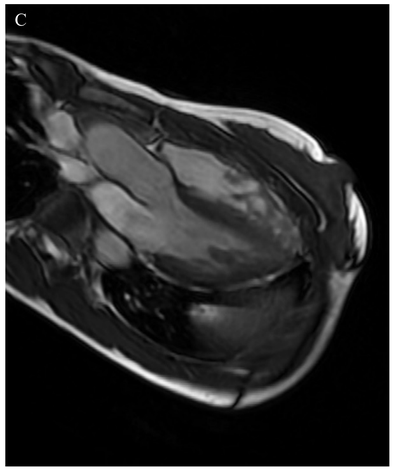
Case Presentation

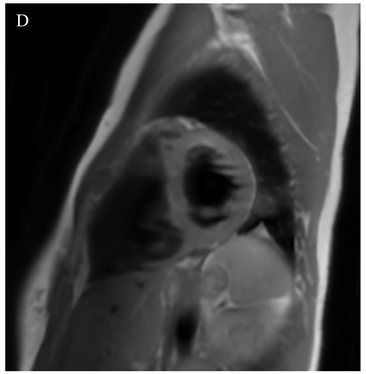
- A 19-year-old asymptomatic female cross-country athlete was referred to the cardiology clinic for abnormal EKG
- Transthoracic echocardiogram showed severe right ventricular (RV) enlargement, RV apical akinesis and severely reduced RV systolic function
- Stress exercise test no evidence of ischemia or arrhythmia
- Cardiac event monitoring revealed primary rhythm sinus with an average heart rate of 75 bpm with occasional premature ventricular complexes
- A cardiac magnetic resonance (CMR) imaging showed evidence of fatty infiltration of RV free wall. Cardiac phenotype was suggestive of arrhythmogenic right ventricular cardiomyopathy.
- Genetic testing revealed heterozygous for a variant (c.3785 G>A) of the MYH6 gene of uncertain clinical significance which typically has autosomal dominant inheritance.
- Ultimately, the patient was advised to abstain from strenuous exercise and avoid intense activities in the future, with a regular follow-up with the cardiology department and serial echocardiograms

Imaging









A: EKG showing anterior T-wave inversions; B: TTE showing enlarged trabeculated RV; C: MRI showing highly trabeculated RV; D: MRI showing dilated thin RV

Discussion

- Assessing our patient's perplexing cardiac condition posed a significant challenge for the cardiologist
- The dilemma: should we advise the patient to halt her physical activities and work, prioritizing her safety, or should we recommend she continue, risking potential life-threatening consequences?
- She met one major criterion, displaying repolarization abnormalities as evidenced by T-wave inversions in right precordial leads V1-V4
- However, genetic testing did not reveal any positive markers for genes associated with ARVC
- Intriguingly, our patient's clinical presentation does not align clearly with either ARVC or dilated or hypertrophic cardiomyopathies, creating a diagnostic and prognostic conundrum

Conclusion

- The diagnostic journey for our case had a challenging path
- Our objective is to raise important questions about the management of such cases and pave the way for future discussions on how to navigate similar situations

References

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- Disclosures: None