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A Case of Asymptomatic Siblings with Brugada Syndrome

Background: Brugada Syndrome is a rare, inherited cardiac channelopathy that predisposes individuals to life-threatening arrhythmias and sudden cardiac death (SCD), and often presents diagnostic challenges due to its subtle clinical manifestations. Case:We present a case of two asymptomatic African American siblings, both found to have identical rare pathogenic variants associated with Brugada syndrome through genetic testing in the outpatient setting. The siblings were referred for evaluation following the sudden cardiac death of their father. Initial electrocardiogram (ECG) showed sinus rhythm with a right bundle branch block and a saddle-shaped QRS pattern. A subsequent 15-lead ECG revealed sinus rhythm with right axis deviation, right ventricular conduction delay, and ST elevation in lead V2. A repeated ECG with Brugada leads showed an incomplete right bundle branch block but no ST elevation. Genetic testing using the GeneDx Brugada Syndrome Sequencing and Deletion/Duplication Panel for both siblings identified a heterozygous pathogenic variant p.(Arg367Cys)(CGC>TGC): c.1099 C>T in exon 9 of the SCN5A gene. Both siblings were diagnosed with type 3 Brugada syndrome and were referred to cardiogenetics and electrophysiology for further evaluation and management. Discussion:Genetic testing plays a crucial role in diagnosing and managing hereditary cardiac diseases, especially in asymptomatic patients with clinical suspicion. Recent studies emphasize its importance in risk stratification and determining the appropriate management strategies. The availability of genetic testing in the outpatient setting provides a valuable tool for early identification and intervention in patients at risk for arrhythmic events.Conclusion:This case highlights the importance of genetic testing in diagnosing Brugada syndrome, even in asymptomatic patients, and underscores the need for further evaluation and management in at-risk individuals, especially those with a family history of SCD.