Follow-up testing indicated for inherited cardiac syndrome that can cause sudden death

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Giulio Conte, M.D., of the Heart Rhythm Management Centre, UZ Brussel-VUB, Brussels, Belgium and colleagues investigated the clinical significance of repeat testing after puberty in asymptomatic children with a family history of Brugada syndrome who had an initial negative test earlier in childhood. Brugada syndrome is a genetic disease that is characterized by abnormal electrocardiogram findings without structural heart disease and an increased risk of sudden cardiac death. The study appears in the November 19 issue of *JAMA*, a cardiovascular disease theme issue.

Brugada syndrome can present within the first months of life, although more typically in the fourth or fifth decade. Testing using ajmaline (an alkaloid) challenge is recommended, as it is an antiarrhythmic agent that can bring out the diagnostic electrocardiogram (ECG) pattern typical in patients suspected of having Brugada syndrome. Although screening of first-degree relatives is common, no evidence-based guidelines exist, particularly for children with normal ECGs, according to background information in the article.

The study included 53 asymptomatic individuals with a first-degree relative with Brugada syndrome and negative ajmaline test performed before 16 years of age between 1992 and 2010 who were seen at the university hospital of Brussels, Belgium (UZ Brussel VUB) and had an ECG repeated annually and were scheduled to repeat the test after

puberty. Nine individuals were younger than 16 years and 1 presented with spontaneous Brugada type 1 ECG at age 16 years. The remaining 43 individuals repeated the ajmaline test, which unmasked type 1 ECG in 10 patients (23 percent).

"The ECG phenotype does not appear during childhood in most cases, but may develop later in response to hormonal, autonomic, or genetic factors," the authors write.

"Screening of asymptomatic first-degree relatives of patients with Brugada syndrome is advisable, although the ideal timing is unknown. Relatives developing symptoms should always be investigated with ajmaline challenge even if they had a negative drug test performed before puberty. These findings support the need for repeat monitoring of family members of patients with Brugada syndrome, including those initially considered at low risk because of young age."

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