Incessant Bidirectional Ventricular Tachycardia as the Presenting Arrhythmia of Andersen-Tawil Syndrome in Two Families

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Introduction: Familial bidirectional VT (BDVT) is commonly associated with catecholaminergic polymorphic VT secondary to mutations in RyR2 and *CASO2* genes. We evaluated 2 families who presented with incessant BDVT and dysmorphic features.

Methods: Clinical and genetic evaluation of family members.

Results: In the first family (n=30), 3 females presented with incessant BDVT and dysmorphic features. 3 males presented with PVCs on Holter monitoring.
All displayed normal QT intervals. There were no symptoms related to arrhythmia, no family history of sudden death or evidence of structural heart disease (SHD). Screening for RyR2, *CASO2*, *triadin-1 (Trd)* and *junctin (Jn)* gene defects were negative. Screening of all exons and intron borders of *KCNJ2* revealed a missense mutation M307V in affected family members only, corresponding with Andersen Tawil syndrome (ATS).
In the second family (n=13), 2 females presented with incessant BDVT. Males presented with few PVCs on Holter monitoring. No cases of SCD were reported. The female proband presented with recurrent syncope in her fifth decade, intermittent prolonged QT interval as well as dysmorphic features, and no evidence of SHD. Screening for the genes of LQTS revealed mutations in the *KCNJ2* gene (R67W, associated with ATS) and *CACNA1C* gene (N2091S) in 3 affected family members, but also a G490R mutation in *CACNA1C* in the proband only.

Conclusion: We describe two families with BDVT as the main presenting arrhythmia of ATS, secondary to mutations in *KCNJ2*. In addition to BDVT there were also dysmorphic features. Interactions with other mutant ion channel defects and / or female gender likely underlie the variability in phenotypes.