Genetic Risk in Early Repolarization Syndrome

Jean-Jacques Schott\textsuperscript{1,2,3}, Jean-Baptiste Gourraud\textsuperscript{1}, Vincent Portero\textsuperscript{1}, Solena Le Scouarnec\textsuperscript{1}, Pascal Chavernac\textsuperscript{2}, Philippe Mabo\textsuperscript{1}, Frederic Sacher\textsuperscript{2}, Michel Haïssaguerre\textsuperscript{3}, Herve Le Marec\textsuperscript{1}

\textsuperscript{1}Inserm U915, l’institut d thorax, France, \textsuperscript{2}Service de Cardiologie, CH Castres Mazamet, Castres, France, \textsuperscript{3}Service de cardiologie, Hopital cardiologique, Bordeaux and Hopital Pontchaillou, Rennes

Early Repolarization (ER) has been associated to idiopathic ventricular fibrillation and sudden cardiac death (SCD) in the general population. Recent population-based studies clearly demonstrate that ER is heritable ($H_2=0.49$). The identification of mutations in the KCNJ8 and CACNAB2b suggests a genetic substrate however the genetic transmission of ER is still poorly documented. We have screened relatives in 3 families affected by ER. In the first family the proband died during an electrical storm and presented an ER pattern. Eleven unexplained SCD occurred and familial screening identified 12 relatives with an ER aspect out of 30 relatives (prevalence 57%). In the second family the proband experienced electrical storms at 45 years old associated with an ER pattern. We identified 3 others unexplained SCD and 25 ER among 82 available ECGs (prevalence 34%). In the third family four patients experienced SCD, one of them presented an ER pattern. Sixteen other patients presented an ER pattern (17/29, prevalence 62%). Valsalva maneuver was performed for 80 relatives. It increased J wave amplitude in 17/20 affected patients and revealed an ER in 17 relatives. In conclusion our study suggests that ERS is transmitted as an autosomal dominant disease.

Keyword: idiopathic ventricular fibrillation, sudden death, genetics