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Masonic Medical Research Laboratory
2150 Bleecker Street
315-735-2217, ext. 127
www.mmrl.edu

PRESS RELEASE

Contact: Ronald P. Kamp
Director of Development
and Communications
kamp@mmrl.edu

SIDS Linked to Genetic Variations Inherited from Both Parents

A genetic variation that occurs in more that 1% of the population is generally referred to as polymorphism. When a genetic variation occurs more commonly it is generally dismissed as playing a prominent role in the development of disease. This rule of thumb was recently shown to be faulty by a team led by Dr. Charles Antzelevitch, Executive Director and Director of Research at the Cardiac Research Institute of the Masonic Medical Research Laboratory (MMRL).

MMRL scientists working with colleagues worldwide co-authored a study showing that a common polymorphism in a gene encoding a potassium channel in the heart inherited from one parent when combined with a genetic mutation in the same gene inherited from the other parent resulted in two cases of early infant death. The parents were both asymptomatic, although each carried a mild form of the disease. The study, published in *Circulation Cardiovascular Genetics*, a Journal of the American Heart Association in early May, suggests that a common polymorphism, found in nearly 30% of the population, can markedly accentuate the malfunction of a mildly defective potassium channel, leading to lethal long QT syndrome-mediated arrhythmias and sudden infant death.

Long QT is an abnormality of the heart's electrical system caused by defects in heart muscle cell structures known as ion channels. These electrical defects predispose affected infants to a very fast heart rhythm called Torsade de Pointes, which can lead to sudden cardiac death.

Identification of infants at risk for sudden infant death syndrome (SIDS) is a major focus of scientists and clinicians. Recent studies suggest that 10-15% of SIDS cases are attributed to lethal cardiac arrhythmias. The first study linking an abnormal rhythm of the heart with SIDS was performed at the MMRL and published in the New England Journal of Medicine in 2000.

The Cardiac Research Institute at the MMRL is one of the leading centers of study of lethal cardiac arrhythmias in the world. Their scientists have developed experimental models of life-threatening cardiac arrhythmias that have aided in the development of new therapeutic modalities to combat sudden death in infants afflicted with congenital syndromes such as Long QT, Short QT and Brugada syndromes.

Please visit www.mmrl.edu to learn more about MMRL research.