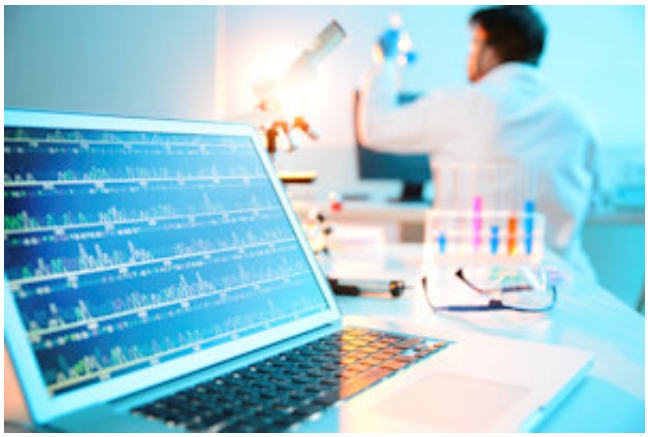
Researchers uncover how a faulty gene can trigger fatal heart condition

10 June 2015, by Jamie Brown



A faulty gene was identified in 2001l in heart cell

University of Manchester research presented today at the British Cardiovascular Society Conference has revealed how a faulty gene can cause fatal abnormal heart rhythms that are brought on by exercise.

Dangerous heart rhythms called arrhythmias, often caused by undiagnosed heart conditions, can cause sudden cardiac arrests that take the lives of seemingly healthy young men and women including sports people.

A healthy, functioning heart has a regular electrical impulse that causes the heart to beat and pump blood around the body. If this impulse is interrupted or becomes irregular, it causes an abnormal heartbeat, called an arrhythmia. Arrhythmias can be a long-term condition, treated with medication or surgery or they can occur unexpectedly, suddenly stopping the heart from beating. For some people, sudden arrhythmias can be brought on by exercise and are often fatal.

A faulty gene was identified in 2001, pointing to a calcium channel in heart cells not functioning as it should. The channel should open and shut to regularly let calcium into the heart cells, kicking off the required electrical impulse for the heart to beat. A research team from The University of Manchester, funded by the British Heart Foundation (BHF), has found that when someone has the faulty gene, this channel can stay open for too long, making it leaky. This causes a rare but potentially fatal heart condition called CPVT (catecholaminergic polymorphic ventricular tachycardia)

When someone exercises, adrenaline is released which increases the amount of calcium stored in the cells. If someone has CPVT and has these leaky channels, calcium can flood into the heart cells, causing a fatal arrhythmia. The leaky calcium channels might go completely unnoticed until this happens.

Professor Clifford Garratt and Dr Luigi Venetucci, clinicians in the team, are working with the families of those who have died from sudden arrhythmic death syndrome (SADS) to determine if they are also at risk. As the gene is inherited, the team want to know if everyone with the faulty gene develops an arrhythmia or if there are other genes involved.

BHF Professor David Eisner from The University of Manchester's Institute of Cardiovascular Sciences, who led the research, said: "People who are prone to sudden arrhythmias often die young. Survivors may have an internal defibrillator fitted at a young age, to shock their heart back into a regular heartbeat if needed but the device does not last forever and needs replacing as the child or young person grows.

"A better understanding of what goes wrong inside the heart during an arrhythmia is crucial to finding the genes that can cause abnormal heart rhythms, and developing better treatments for people at risk of the tragedy of sudden cardiac death."

Professor Jeremy Pearson, Associate Medical

Director at the BHF, which funds the Manchester research team, said: "Exercise is a vital part of maintaining a healthy heart and for the vast majority of people, it should be part of their daily routine but, for some people, exercise can trigger an underlying condition that they didn't know they had. We know that screening doesn't find everyone with the genes that can make them prone to sudden cardiac death and we urgently need more research to understand the causes of these rare, but potentially fatal, arrhythmias."

Provided by University of Manchester