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Genes that control heartbeat could shed light on heart defects

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An international study involving researchers at the Medical Research Council (MRC) Human Genetics Unit has found several areas of the human genome responsible for controlling the rhythm of heartbeats. By analysing the genetic code of nearly 50,000 people, researchers are now one step closer to understanding more about those with irregular heartbeat and who may be at increased risk of heart problems.

The study, published today in *Nature Genetics* showed that there were 22 areas of the human genome associated with influencing the process of how the heart beats, known as cardiac ventricular conduction. The researchers were not only able to discover new genes associated with the heart, but were also able to pinpoint the precise role that some of the genes play in order to control the electrical pulses that regulate the heartbeat.

Each of the 50,000 participants had an electrocardiogram (ECG) to measure their heart's electrical activity. By analysing their genetic code, researchers were able to identify genes responsible for recorded variations in cardiac ventricular conduction.

The lead author on this international study was Dr Nona Sotoodehnia at the University of Washington. The data for the cardiac conduction genetics study came from a consortium of 15 European and American studies. The research involved more than 100 scientists from the United Kingdom, Europe and the United States.

Dr Caroline Hayward at the MRC Human Genetics Unit in Edinburgh who was involved in the analysis of the study says:

"By understanding more about the biological processes that control every heart beat, we can begin to have real insight into causes and potential treatments for people at increased risk of sudden death, heart failure and cardiac mortality."

Dr Jim Wilson, Royal Society University Research Fellow at the University of Edinburgh, said:

"This study demonstrates the great potential of modern genetics to help us better understand how our heart's electrical system works at the molecular level. This is very important as heart conduction abnormalities increase the risk of heart failure and sudden death: this knowledge will be used in the hunt for new treatments."

The research was funded by the National Institutes of Health (NIH), the Medical Research Council (MRC), the Chief Scientist Office and the Royal Society. UK universities taking part in the research include the University of Edinburgh, the University of Leicester and the University of Glasgow.

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2. For almost 100 years the Medical Research Council has improved the health of people in the UK and around the world by supporting the highest quality science. The MRC invests in world-class scientists. It has produced 29 Nobel Prize winners and sustains a flourishing environment for internationally recognised research. The MRC focuses on making an impact and provides the financial muscle and scientific expertise behind medical breakthroughs, including the first antibiotic penicillin, the structure of DNA and the lethal link between smoking and cancer. Today MRC funded scientists tackle research into the major health challenges of the 21st century. www.mrc.ac.uk

3. Genome-wide association studies examine hundreds of thousands of genetic variants in large numbers of people to try to find sequence variants and genes associated with particular diseases or conditions.

