

**PRESS RELEASE. HOLD FOR RELEASE UNTIL SUNDAY, FEBRUARY 8<sup>TH</sup> AT 17:00**

*The results will be presented this month on the online version of the prestigious journal "Nature Genetics"*

## **Three new genetic traits are identified that are associated with the risk of suffering an acute myocardial infarction**

**Barcelona, 4 February 2009.** Within the framework of the **REGICOR** (Registre Gironí del Cor) study, an international consortium of North American and European researchers, including researchers from the **Municipal Institute of Medical Research** (IMIM-Hospital del Mar) and the **Hospital Josep Trueta** in Girona, has discovered three new genetic traits linked to acute myocardial infarction (on genes *WDR12*, *PHACTR1* and *KCNE2*) and has confirmed another six identified in previous studies on this disease (*LDLR*, *PCSK9*, *CXCL12*, *CDKN2A-2B*, *SORT1*, *MIA3*). Myocardial infarction is the leading cause of disability and death by disease in developed nations. **It is calculated that between 80,000 and 85,000 myocardial infarctions occur each year in Spain.**

In an early phase of the project, the researchers determined 2.5 million genetic traits (also called polymorphisms) in nearly 3,000 people that had suffered an acute myocardial infarction before the age of 60 (41 for men and 47 for women, on average). They then compared them with more than 3,000 healthy individuals as a control. The study participants came from the United States, Sweden, Finland, Italy and Spain. Comparing the DNA of these two groups, the 1,400 most significant polymorphisms were selected and analysed in more than 10,000 individuals who had suffered a myocardial infarction, comparing them to 10,000 healthy individuals, all of whom were different from the 6,000 individuals who had participated in the first phase of the project. Based on this process, a set of polymorphisms located on 9 genes was able to be confirmed, **each one of which increases the risk of suffering an acute myocardial infarction.**

Two of the genes identified affect the control of cholesterol levels in the blood (*LDLR* and *PCSK9*) and a third affects the mechanisms of inflammation (*CXCL12*), which affect the development of arteriosclerotic plaques in the arteries. However, surprisingly, the mechanism that explains the relation of the other six genes (*CDKN2A-2B*, *SORT1*, *MIA3*, *WDR12*, *PHACTR1*, *KCNE2*) with myocardial infarctions is completely unknown. According to **Roberto Elosua**, coordinator of the research group on cardiovascular epidemiology and genetics at IMIM-Hospital del Mar: *"These results confirm the importance of cholesterol levels and the mechanisms of inflammation, but more importantly, they open new lines of research to determine new mechanisms related to these six genes that influence the risk of suffering from this disease. In the long term, this knowledge may contribute to the development of new drugs that increase the existing therapeutic options for the prevention and treatment of this prevalent disease."*

The researchers affirm that the determination of these polymorphisms may soon make it possible to identify those individuals with an elevated risk of suffering an infarction. However, it must be remembered that the increase in risk associated with each one of these polymorphisms is relatively low (10-15%) and that their presence does not mean that the individual will necessarily develop the disease. It is important to note that the possible interaction with environmental factors is still crucial: diet, physical exercise, refraining from smoking and controlling risk factors such as hypertension, cholesterol and diabetes.

### **Reference articles:**

*Myocardial Infarction Genetics Consortium. Genome-wide association of early-onset myocardial infarction with common single nucleotide polymorphisms, common copy number variants, and rare copy number variants. Nat Genet Epub 2009 Feb 8.*

*Erdmaan J, et al. Novel susceptibility locus for coronary artery disease on chromosome 3q22.3. Nat Genet Epub 2009 Feb 8.*

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