

NEWS EMBARGOED UNTIL 7PM ON 11TH SEPTEMBER 2011

New Leads on the Genetic Origin of High Blood Pressure

Two international studies, to be published online on 11th September in the prestigious journals Nature and Nature Genetics, have identified 16 new genetic characteristics associated with blood pressure control

Barcelona, 9 September 2011.- The International Consortium for Blood Pressure genomewide association Studies (ICBP), an international project in which researchers from the IMIM (Hospital del Mar Research Institute) have participated as the only Spanish researchers, has identified 29 loci or genetic characteristics, 16 of which are new, associated with the risk of presenting with high blood pressure in a population of European origin. These 29 loci are also associated with the risk of presenting with ventricular hypertrophy (heart disease consisting in an increase in size of the heart muscle), ictus or cerebrovascular accident and myocardial infarction.

High blood pressure is a chronic disease characterised by continuously increasing measurements of blood pressure in the arteries, and is defined by the presence of systolic blood pressure figures ≥ 140mmHg or diastolic blood pressure ≥ 90mmHg. High blood pressure is the most common cardiovascular risk factor in Spain; recent studies have shown that 47% of men and 39% of women aged between 35 and 74 years present with high blood pressure. Genetic factors are known to explain a large part of the risk of the population presenting with high blood pressure. Therefore, a lot of effort is being invested in identifying the genetic characteristics associated with this disease, with 13 genetic characteristics associated with systolic and diastolic blood pressure levels being identified in previous studies, some of which were discovered by this same research group.

Over 200,000 people have taken part in the *ICBP* study. At first, about 2,500,000 genetic characteristics were determined and blood pressure was measured in an initial group of 69,000 people. A group of genetic characteristics of particular interest was identified, which was analysed again in a second phase in an additional group of 134,000 people.

According to Roberto Elosua and Gavin Lucas, researchers from the IMIM's cardiovascular genetics and epidemiology research group and co-authors of this study, "16 new areas of DNA associated with a higher probability of presenting with high blood pressure figures have been identified, so we currently know 29 loci associated with this disease". The effect of each of these "risk characteristics" is very small and ranges between 0.3 and 1.1 mmHg, yet the risk rises with the amount of risk characteristics a person has present in their DNA

from birth. Despite this major step forward, these genetic characteristics only explain 1% of the blood pressure differences which exist between people.

This study confirms the causation between high blood pressure levels and ictus and myocardial infarction, since the higher the number of genetic risk characteristics for high blood pressure, the greater the probability of presenting with these two diseases. Nevertheless, according to the researchers "one of the most surprising results is that these genetic characteristics are not associated with the presence of kidney failure, suggesting that high blood pressure is a consequence but not a cause of kidney disease".

Some of these genetic characteristics are in genes which could be related biologically with kidney function regulation and the production of proteins which control the level of contraction of muscle fibres in arteries, fibrosis in the heart and arteries, and the control of ion secretion in the kidneys. But there are many genetic characteristics which identify new mechanisms, still unknown, which open the door to research on new therapy targets for controlling high blood pressure.

The IMIM cardiovascular genetics and epidemiology research group was able to undertake this project thanks to a grant from the TV3 Fundación Marató awarded in 2008.

Reference Articles

"Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk". Ehret et al. The International Consortium for Blood Pressure genome-wide association studies. Nature . DOI 10.1038/nature10405

"Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure". Wain et al. The International Consortium for Blood Pressure genome-wide association studies. Nature Genetics.

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