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http://grupsderecerca.uab.cat/cacereslab/ingenihus

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Inversion genotyping goes personal

A new genotyping tool for detecting inversions in the human genome potentially associated with disease is being developed by the comparative and functional genomics group of the Universitat Autònoma de Barcelona (UAB). Together with the UAB Technology Transfer Office, the team is now looking to commercialize the approach and expand its use to many indications.

The study of genetic variants in the human genome and their application to personalized medicine has exploded since the completion of the human genome sequence in 2001. In particular, single-nucleotide polymorphisms (SNPs) and copy-number variations have been characterized in great detail. To date, however, very few of these analyses have uncovered mechanisms involved in disease.

One type of genetic variant that has been studied very little, despite the fact that it was shown to be at the root of simple and complex diseases and other relevant traits, is genomic inversions. A number of technical limitations due to the difficulty of determining DNA-segment orientation through sequencing or the use of SNP microarrays have made such studies extremely rare.

Enter Mario Cáceres, head of UAB's comparative and functional genomics group and an Institució Catalana de Recerca i Estudis Avançats (ICREA) research professor. As part of an ambitious project funded by the European Research Council (ERC) with a €1.5 million ($1.68 million) starting grant, Cáceres's group is carrying out the complete characterization of polymorphic inversions in the human genome. The group recently published the first high-quality partial catalog of several hundred polymorphic inversions. In addition, the group obtained a €150,000 ($168,000) ERC Proof of Concept Grant to develop an efficient and high-throughput assay for genotyping inversions in hundreds of samples in a fast and easy way. The method, dubbed Inversion Genotyping in Humans (INGENIHUS) and for which a patent is pending, has shown high sensitivity and accuracy.

"We have found that many inversions are not associated to SNPs and have been missed by genome-wide association studies. Our new high-throughput genotyping assay focuses specifically on those inversions most difficult to detect with existing methods and makes it possible to study the potential functional consequences of these variants," said Cáceres.

The development of INGENIHUS has created a new opportunity for progress in the diagnosis and prognosis of genetic diseases that could be treated with personalized medicine. This tool, based on probe hybridization, allows for the simultaneous detection of multiple inversions in hundreds of individuals in a few days and at an affordable cost. To date, around 50 inversions have been genotyped in 550 individuals from six populations (European, African or Asian ancestry).

Next, the team plans to wrap up proof-of-concept studies and make the tool available in Q3 2015, and together with the UAB Technology Transfer Office, it is looking to commercialize INGENIHUS through a licensing deal with an interested industrial partner.

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