

# “ New high-throughput method to genotype human inversions for personalized medicine purposes

## THE INVENTION

Novel high-throughput assay to genotype those human inversions in human genome that can not be detected by current techniques. This method provides key information highly useful in the field of personalized medicine (diagnostic, genetic test, drug development and clinical trials). We are seeking a company partner to commercialize this licensing opportunity.

### Innovative aspects and advantages

- > High-throughput method allowing us to genotype simultaneously multiple inversions in hundreds of individuals.
- > Method with high sensitivity, reproducibility and accuracy method.
- > To date, 24 inversions have been genotyped in 540 human individuals from 6 populations (European, African or Asian origin).
- > Genotyping success of 98%.

### IP Rights

Priority European patent application, 23rd July 2013

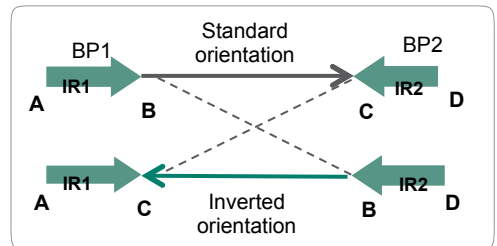
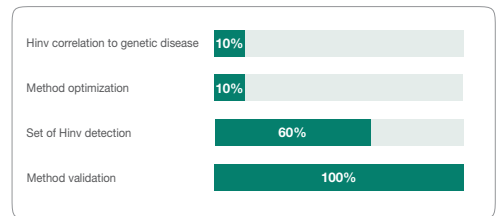
### Scientific Team

Mario Cáceres, leads the Comparative and Functional Genomics research group of the Biotechnology&Biomedical Institute of the UAB. He has the European Research Council Starting Grant of Investigation for the research done on: Evolutionary and functional analysis of polymorphic inversions in the human genome.

### Summary

In the last few years, changes that affect bigger regions of the DNA, known as structural variants, have been discovered. Human inversions (HInv) are a common class of them that changes the orientation of one segment of the genome. They have a role in phenotypic characteristics, including susceptibility to genetic diseases and genomic disorders however they cannot be detected by massive sequencing techniques. Current methods available to detect HInv don't allow the detection of all the classes of HInv and there is not any method to genotype multiple HInv at a population level. The method is based on Multiplex Ligation-dependent Probe Amplification (MLPA) and presents a very high sensitivity, reproducibility and accuracy in comparison to other techniques like inverse PCR. This method is the fastest method to determine HInv in big sets of DNA samples, being able to produce 12769 genotypes in a short period of time. This is especially useful for HInv flanked by large repetitive sequences (<70 kb), which are precisely the ones most difficult to study by other methods

### State of development



► Fig. 1 Structure of Human Inversion