New computational method allows the identification of genetic alterations in cancer patients within hours

- *Today the prestigious journal* Nature Biotechnology *is publishing an article on SMUFIN, a new computational method capable of identifying genetic alterations responsible for the formation and progression of tumors simply, quickly and precisely*
- *SMUFIN identifies accurately practically all types of genetic changes involved in the onset and progression of cancer, including large-scale chromosome rearrangements which, until now, have been hard to detect*
- *This new method is a realistic step towards personalized medicine, in which the genome analysis of each patient will aid diagnosis and allow the selection of treatments which are more effective and less invasive*
- *SMUFIN represents a new way of analyzing genomes which could also be applied to the study of the genetics underlying many other illnesses prevalent in our society*

(Barcelona, 26 October 2014) – A new computational method has made it possible to detect genetic changes responsible for the onset and progression of tumors in a simple, quick and precise way. The SMUFIN (*Somatic Mutations Finder*) method is capable of analyzing the complete genome of a tumor and identifying its mutations in a few hours. In addition, it is able to identify alterations which had previously not been revealed, even using methods which require the use of supercomputers over several weeks.

Today's issue of the prestigious journal *Nature Biotechnology* features an article describing the characteristics of SMUFIN, which has been developed by the computational genomics group at Barcelona Supercomputing Center (BSC). The team is led by David Torrents, ICREA Research Professor ICREA at the BSC, in collaboration with research groups at Barcelona's Hospital Clínic and the August Pi i Sunyer Biomedical Research Institute (IDIBAPS), the University Institute of Oncology of Asturias at the University of Oviedo (IUOPA), the European Molecular Biology Laboratory (EMBL, Heidelberg) and the Spanish National Genome Analysis Centre (CNAG, Barcelona).

A new way to analyze genomes

One of SMUFIN's main innovations is that it represents a radical change in the method by which genomes are analyzed. To date, identifying mutations responsible for the appearance of tumors has involved comparing genomes taken from the tumor with genomes obtained from healthy cells from the same patient via a reference human genome, which is used as a guide. This lengthy and complex process results in the loss of a considerable amount of information and makes it difficult to identify many mutations which have an impact on the tumor. This analysis is also executed on different computer programs in succession, each one of which is only capable of detecting certain types of variations.

SMUFIN, meanwhile, undertakes a direct comparison between the genome of healthy cells and cells from a tumor in the same patient and determines the location of almost all types of mutations at once, without requiring the use of several programs. This results in a much quicker and more complete analysis.

Advances in the research of aggressive tumors

The article published in *Nature Biotechnology* explains how SMUFIN, in addition to making the analysis faster and more cost-effective, is able to reveal hard-to-detect genetic alterations in aggressive tumors. Using SMUFIN to analyze two types of aggressive cancer samples, a blood tumor (mantle cell lymphoma) and one of the nervous system (pediatric medulloblastoma), has allowed the discovery of almost all the types of mutations occurring in their genomes for the first time and with over 90% accuracy. This includes alterations in the organization of chromosomes, which have not been revealed by methods used to date. This represents the first step necessary to understanding how these chromosome alterations affect the evolution and aggressiveness of the tumor.

Boost for biomedical research

SMUFIN makes it possible for a large number of research groups to study their patients' genomes in a way previously unavailable to them. In addition, when used by supercomputing centers, SMUFIN allows mutations to be identified in hundreds or thousands of cancer genomes in just a few days. In this regard, BSC is already participating in the largest global cancer genome initiative through the International Cancer Genome Consortium (ICGC), which aims to analyze the genomes of thousands of patients in order to study the genetic bases of the onset and evolution of a large number of tumor types.

Advance towards personalized medicine

SMUFIN represents a decisive step towards personalized medicine, where the genome analysis of each patient will enable faster and more accurate diagnosis. It will also allow the development and application of personalized treatments which are less invasive than those currently used. While methods which have existed up until now are complex, limited and require days or even weeks to

undertake the full analysis of a tumor genome, SMUFIN is a practical way to incorporate genome analysis into the health system, as it is capable of analyzing a tumor genome in a few hours accurately and simply.

A development within the framework of CLL and the Severo Ochoa programme

Development work on SMUFIN began at Barcelona Supercomputing Center in 2011 by the genomics team, which is part of the BSC-CRG-IRB (Barcelona Supercomputing Center, Genomic Regulation Centre and Institute for Research in Biomedicine Barcelona) Joint Programme in Computational Biology.

SMUFIN was developed in two research environments in which the center participates. One is the Chronic Lymphocytic Leukemia Genome Project, of which the scientific directors are Elías Campo (Hospital Clínic, IDIBAPS) and Carlos López Otín (University of Oviedo), and which aims to research leukemia by analyzing the genome of more than 500 patients. The development also forms part of the National Severo Ochoa Programme, with which Barcelona Supercomputing Center is driving forward the creation of bioinformatics tools capable of managing and analyzing big amounts of biomedical data which are necessary to make personalized medicine possible, among other tools.

About Barcelona Supercomputing Center

Barcelona Supercomputing Center (BSC) is the national supercomputing centre in Spain. BSC specialises in high performance computing (HPC) and its mission is two-fold: to provide infrastructure and supercomputing services to European scientists, and to generate knowledge and technology to transfer to business and society.

BSC is a Severo Ochoa Center of Excellence and a first level hosting member of the European research infrastructure PRACE (Partnership for Advanced Computing in Europe). BSC also manages the Spanish Supercomputing Network (RES).

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