APPLIED RESEARCH

STATISTICAL PHYSICS MODELS FOR STUDYING GENETIC MUTATIONS IN CANCER

On 20 May, the journal Nature Genetics published important results obtained by a team of Italian researchers who studied the causes of the genetic alterations that most frequently occur in the development of cancer: "chromosomal translocations". The team discovered that the damage to the DNA tends to happen within specific genes, and at precise moments in their activity, which can be identified with a high degree of accuracy. Not all the genes subject to breaking, however, induce mutations linked to cancer, like the translocations. Typically it is those that most frequently come into physical contact within the 3D structure of the chromosomes.

The study, which was led by a team of researchers from the European Institute of Oncology and the University of Milan, also involved physicists from INFN who developed innovative statistical models based on the data of new technologies, such as the so-called "Hi-C" techniques. These allow researchers to measure the probability of physical contact between pairs of genomic loci, for all the possible pairs. These models were employed in analysing the data to understand the molecular mechanisms that link the occurrence of translocations to the three-dimensional architecture of our genome, that is, to the way in which our DNA is organised in space.

The chromosomes, in fact, have a complex 3D organisation in the cell nucleus, which is necessary for the correct fulfilment of their various functions. The way in which chromosomes fold in 3D remains, however, largely unknown; in particular, it is not clear in which way the mutations linked to disease (for example, rearrangements of DNA such as inversions or translocations) modify the chromosomal architecture and thus influence gene regulation. ■