Researchers complete the first epigenome in Europe

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A study led by Manel Esteller, director of the Epigenetics and Cancer Biology Program at the Bellvitge Biomedical Research Institute (IDIBELL), professor of genetics at the University of Barcelona and ICREA researcher, has completed the first epigenome in Europe. The finding is published in the latest issue of the international scientific journal *Epigenetics*.

The genome of all cells in the human body is the same for all of them, regardless their aspect and functions. Therefore, genome cannot fully explain the activity of tissues and organs and their disorders in complex diseases like cancer. It takes a further explanation. Part of this explanation is provided by epigenetics, a field of biology that studies the heredity activity of DNA that does not involve changes in its sequence. That is, if genetics is the alphabet, epigenetics is the spelling that guides the activity of our cells.

Methylation

Epigenetics refers to chemical changes in our genetic material and proteins that regulate it. The best-known epigenetic mark is the methylation, the addition of a methyl chemical group (-CH3) in our DNA. The epigenome consists of all the epigenetic marks of a living being.

The authors of the study have completed the epigenomes for all brands

of methylation of DNA from white blood cells of two girls: a healthy one and a patient suffering from a rare genetic disease called Immunodeficiency, Centromere instability and Facial anomalies syndrome (ICF). This disease is caused by a mutation in a gene that causes the addition of a methyl chemical group in its DNA.

The analysis performed by the researchers reveals that the patient has an epigenomic defect that causes fragility of chromosomes, which thus can easily be broken. In addition, the study shows that the patient has a wrong epigenetic control of many genes related to the response against infection, which causes a severe immune deficiency.

The study coordinator, Manel Esteller, emphasizes that due to this study, "we now know what happens in this type of rare diseases and we can start thinking about strategies for new treatments based on this knowledge."

Dr. Esteller is an international leader in the field of epigenetics. His work has been crucial to show that all human tumours have in common a specific chemical alteration: the hypermethylation of tumour suppressor genes. Since 2008 is the director of the Epigenetics and Cancer Biology Program at IDIBELL.

More information: Heyn H, Vidal E, Sayols S, Sanchez-Mut JV, Moran S, Medina I, Sandoval J, Simó-Riudalbas L, Szczesna K, Huertas D, Gatto S, Matarazzo MR, Dopazo J, Esteller M. Whole-genome bisulfite DNA sequencing of a DNMT3B mutant patient. *Epigenetics*, June 1, 2012.

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