The first epigenetic test to diagnose tumors of unknown origin

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An EPICUP(R) Genetic Test. Credit: IDIBELL

In patients with cancer, initial diagnosis most often includes the

detection of the primary or original tumor and the presence or absence of metastases, ie cells from the original tumor that have escaped from their original location and are growing into other tissues of the patient. However, in between 5% and 10% of human tumors this process is done otherwise: metastasis is diagnosed, but the primary tumor is not detected despite various diagnostic testing. This situation is called Cancer of Unknown Primary (CUP). As the type of tumor is not known, the survival of these patients it is very limited.

Today, an article published in *The Lancet Oncology*, the most prestigious journal in the Medical Oncology field, by Dr. Manel Esteller, director of the Epigenetics and Cancer Biology Program (PEBC) of of Bellvitge Biomedical Research Institute (IDIBELL), ICREA researcher and Professor of Genetics at the University of Barcelona, shows that it is possible to use a newly-developed epigenetic test - called EPICUP- to find out what type of primary tumor is responsible for the metastasis in the patient, which will allow doctors to develop more specific treatments against it.

"A few years ago, we became aware that the chemical patterns that regulate the activity of genes (the epigenome) are specific to each tissue. For example, they are different in a pancreatic cell compared to a lung cell" says Dr. Manel Esteller. "We have analyzed these particular epigenetic signatures for each type of cancer in more than 10,000 human tumors. When we now study the DNA of the metastasis of a patient with a tumor of unknown origin, the photograph of the epigenome that we get will tell us that it belongs to the family of pancreatic cancer, lung, colon, breast, etc. in other words, we will give a diagnosis of the origin of the tumor.

Identification of the type of cancer by epigenetic test will have a significant impact on the choice of treatment. "From now on, the patient will not be treated blindly, since we will be able to provide a much more

specific therapy for this tumor type; actually, initial data shows that survival is doubled", explains Dr. Esteller, and he concludes on research in *The Lancet Oncology*: "Something very important to keep in mind is that this is not a discovery to be developed in the coming years; our collaboration with Ferrer laboratories made it possible for this test to be applied from this very moment."

More information: Moran S, Martínez-Cardús A, Sayols S, Musulén E, Balana C, Estival-Gonzalez A, Moutinho C, Heyn H, Diaz-Lagares A, Castro de Moura M, Stella GM, Comoglio PM, Ruiz-Miró M, Matias-Guiu X, Pazo-Cid R, Antón A, Lopez-Lopez R, Soler G, Longo F, Guerra I, Fernandez S, Assenov Y, Plass C, Morales R, Carles J, Bowtell D, Mileshkin L, Sia D, Tothill R, Tabernero J, Llovet JM, Esteller M. Epigenetic profiling to classify cancer of unknown primary: a multicentre, retrospective analysis. *The Lancet Oncology*, 16tlo0734, 2016.

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