Mutations directly identifiable in active genes

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Researchers at Uppsala University, Sweden, have developed a new method for identifying genetic variation, including mutations, in active genes. Hopes are strong that the method represents an important research tool that will lead to the development of new diagnostic tests.

The new method, which is directly applicable to cell preparations and tissue sections, should enable studies of the effects of genetic variation in patient samples from a variety of diseases, including, particularly, cancer. The method was developed under the supervision of Mats Nilsson, Professor of Molecular Diagnostics at the Department of Genetics and Pathology, Rudbeck Laboratory, Uppsala University. The findings have just been published as an article in the well-regarded journal Nature Methods.

The method is an elaboration of a technique previously developed by the same research team, involving the use of molecular "padlock probes" to identify specific molecules in individual cells. The probes are able to to distinguish similar genetic sequences, which makes them highly suitable for mutation analysis. Due to the signal amplification associated with padlock probes it is, for the first time, possible to directly identify genetic variation at the mRNA level, that is to say, in molecules produced by active genes, in cells in microscopic preparations, .

"The method allows us to study biological processes in individual cells as opposed to the average states of large numbers of cells," says Mats Nilsson.

Processes specific to cells that represent a minority in the context of a given sample can thus be identified, since their associated signals can escape being drowned out by those generated by the majority. The method is thus of significant interest to the study of tumour tissue, which contains a mix of cancer and normal cells.

"Hitting the proverbial needle in the haystack should now be possible," says Mats Nilsson. "This should entail significantly more sensitive and precise diagnostic methods, improving the prospects that patients will receive the treatment they need."

The method should also make possible various ways of studying the effects of genetic variants on different types of cells and tissues, something that is difficult with methods that rely on preparations comprising a multitude of tissue cells.

The researchers aim to extend the method to allow for parallel identification of multiple molecules and for analysis of biobank material.

More information: Paper: www.nature.com/nmeth/journal/v … full/nmeth.1448.html

Provided by Uppsala University

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