Spanish researchers develop new method to diagnose hereditary breast and ovarian cancer

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Researchers of the Catalan Institute of Oncology (ICO) at the Bellvitge Biomedical Research Institute (IDIBELL) have developed and validated a new method to diagnose hereditary breast and ovarian cancer syndrome based on mass sequencing of BRCA1 and BRCA2 genes. The model is based on a genetic and bioinformatic analysis which has been proved very effective. The new protocol has been described in an article published in the *European Journal of Human Genetics*.

In recent years, new advances in sequencing techniques have involved the development of new platforms for nucleic acid sequencing, called mass sequencing platforms or next-generation sequencing. These technological improvements have brought a revolution in biomedical research, in the field of genetics and genomics. The emergence of next-generation sequencers and the possibility of combining samples from different patients using identifiers have allowed to adapt these new technologies in the field of the genetic diagnosis.

Using a platform of last generation mass sequencing, the team led by the researcher Conxi Lázaro, from the Hereditary Cancer Program at the ICO and IDIBELL, has developed a comprehensive protocol that allows to sequence all coding and adjacent regions of BRCA1 and BRCA2 genes, responsible for hereditary breast and ovarian cancer syndrome.

Mass sequencing algorithm

"This approach allows to identify all point mutations and small deletions and insertions analyzed, even in regions of high technical difficulty, such as homopolymeric regions", explains the ICO-IDIBELL researcher. The protocol developed is an own algorithm of mass sequencing and bioinformatics analysis that has been shown to be very efficient to detect all existing mutations and to eliminate false positives.

The validation of this algorithm to diagnose hereditary breast and ovarian cancer syndrome has shown a sensitivity and specificity of 100% in the analyzed samples, while reducing costs and time for obtaining the results.

Furthermore, the research team led by Lázaro has implemented the use of this approach for the responsible genes for hereditary colorectal cancer, such as familial polyposis and Lynch syndrome.

Up to ten percent of cancers are hereditary, which means that the genetic mutations predisposing to various types of tumors are transmitted from parents to offspring. The identification of these mutations is very important to prevent the occurrence of tumors in people who have familial predisposition.

The hereditary breast and ovarian cancer syndrome is one of the hereditary cancer types that affects more people. The disease is caused by mutations in the BRCA1 and BRCA2 genes. These mutations are also associated with other kind of cancers.

More information: Feliubadaló L, Lopez-Doriga A, Castellsagué E, del Valle J, Menéndez M, Tornero E, Montes E, Cuesta R, Gómez C, Campos O, Pineda M, González S, Moreno V, Brunet J, Blanco I, Serra E, Capellá G, Lázaro C. Next-generation sequencing meets genetic diagnostics: development of a comprehensive workflow for the analysis of BRCA1 and BRCA2 genes. European Journal of Human Genetics.

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