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Evaluating the feasibility of NGS-based germline and somatic genetic testing in triple-negative breast cancer: The PERSONA-breast trial



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Approximately 5-10% of all patients with breast cancer exhibit a genetic predisposition to the disease. Germline mutations of BRCA1/2 are recognized as the most common defect associated with inherited breast and ovarian cancers and mainly express the molecular phenotype of basal-like or triple negative breast cancers (TNBCs): in fact, 70% of BRCA1/2-associated cancers are triple-negative. However, only up to 25-30% of all hereditary breast cancer and only 20% of TNBCs can be ascribed to BRCA1 and BRCA2 mutations. Since the discovery of BRCA1 and BRCA2 genes, more than other 25 genes have been associated with familial breast and/or ovarian cancer, which are not currently assessed with genetic testing. Next generation sequencing now enables parallel testing of multiple genes. A growing body of data suggests that implementation of genetic testing, with the inclusion of somatic DNA alterations assessment, might impact management and treatment decisions in both early-stage and locally advanced/metastatic breast cancer. At the European Institute of Oncology, we initiated a clinical study which aims at conjugating both somatic and germline DNA sequencing of tissues from breast cancer patients to provide a more comprehensive picture of the mutational landscape and intrinsic risk of recurrence in patients with TNBC. We will use two different gene panels: the TruSight Cancer (Illumina), a commercially validated targeted enrichment panel which looks at 94 genes and 284 SNPs associated with a predisposition towards cancer, and a new in-house panel, developed at IEO. The latter platform analyses 349 genes with an established function in the biology of multiple solid tumors, including breast cancer. The purposes of our study are to evaluate the feasibility of introducing multiple genes sequencing in the clinical

setting, to establish the prevalence of inherited mutations in patients with TNBC, as well as to identify new important genes responsible for the predisposition to the disease. We believe that deeper knowledge of the molecular landscape of each tumor will inform medical and surgical treatment decisions, providing each patient with the best possible outcome.

Biography

Corona S P has completed her PhD from Melbourne University, Australia. She is currently working as a Medical Doctor/Researcher at the Breast Surgery Division of the European Institute of Oncology in Milan, a worldly renowned institute. She has published papers and served as Reviewer in international peer-reviewed journals.

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