Migrating the SNP array-based homologous recombination deficiency measures to next generation sequencing data of breast cancer - DTU Orbit (17/02/2019)

Migrating the SNP array-based homologous recombination deficiency measures to next generation sequencing data of breast cancer

The first genomic scar-based homologous recombination deficiency (HRD) measures were produced using SNP arrays. As array-based technology has been largely replaced by next generation sequencing approaches, it has become important to develop algorithms that derive the same type of genomic scar scores from next generation sequencing (whole exome “WXS”; whole genome “WGS”) data. In order to perform this analysis, we introduce here the scarHRD R package and show that using this method the SNP array-based and next generation sequencing-based derivation of HRD scores show good correlation (Pearson correlation between 0.73 and 0.87 depending on the actual HRD measure) and that the NGS-based HRD scores distinguish similarly well between BRCA mutant and BRCA wild-type cases in a cohort of triple-negative breast cancer patients of the TCGA data set.

General information
State: Published
Organisations: Department of Bio and Health Informatics, Cancer Genomics, Semmelweis University, Eotvos Lorand University, University of Copenhagen, University College London, Danish National Life Science Supercomputing Center
Number of pages: 4
Publication date: 2018
Peer-reviewed: Yes

Publication information
Journal: npj Breast Cancer
Volume: 4
Article number: 16
ISSN (Print): 2374-4677
Ratings: Web of Science (2019): Indexed yes
Original language: English
Electronic versions: s41523_018_0066_6.pdf
DOIs: 10.1038/s41523-018-0066-6
Source: FindIt
Source-ID: 2436237128
Research output: Research - peer-review > Journal article – Annual report year: 2018