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Epigenome-wide discovery of ovarian and breast cancer specific DNA methylation markers

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Breast and ovarian cancers pose huge and unsolved challenges to the medical profession. The EU FP7 consortium EpiFemCare develops blood tests based upon DNA methylation technology to facilitate early detection and prediction of therapeutic outcome of these cancers. In phase 1 of the project Infinium® HumanMethylation450 BeadChip technology is used to assess the methylation status of ~485'000 sites in cancer and control tissues. In parallel, Reduced Representation Bisulfite Sequencing (RRBS) is used to identify and confirm cancer specific methylated circulating DNA in matching serum samples. Using Genedata Expressionist® for Genomic Profiling, we have established an automated bioinformatics pipeline for the detection of cancer specific Differentially Methylated Regions (DMRs) that most likely fulfill the strict specificity criteria of a serum based test. The most promising DMRs are taken forward to clinical assay development (phase 2) and validation in thousands of serial samples from prospective clinical trials (phase 3).