Molecular characterization of ovarian carcinoma by the size of somatic copy number alteration

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Somatic copy number alterations (SCNAs) play a crucial role in cancer development by activating oncogenes and suppressing tumor suppressor genes. SCNAs can be divided into focal SCNAs (fSCNA), which are shorter than chromosome arm and chromosomal SCNAs (cSCNA), which that have the length of the chromosome arm. These two SCNAs are caused by different biological mechanisms. However, the characteristics of these two SCNAs are largely unknown in ovarian cancer. Therefore, we investigated the molecular characteristics of fSCNAs and cSCNAs in ovarian cancer.

In this study, we used 536 ovarian cancer patients from the cancer genome atlas (TCGA) data and 46 ovarian cancer patients from our data. We classified patients into 4 groups based on fSCNAs and cSCNAs in each data. Among the 4 groups, patients with high fSCNAs had higher mutation rates of germline BRCA1 gene and CDK12 gene than other groups. In contrast, patients with low fSCNAs and high cSCNAs had the lowest mutation rates of HRR related genes. As a result of differentially expressed gene (DEG) analysis between the 4 groups, patients with both low fSCNAs and cSCNAs had elevated expression of genes related immune response and epithelial–mesenchymal transition (EMT).

In conclusions, we identified that ovarian cancer patients could be classified into 3 distinct groups by fSCNAs and cSCNAs. Patients with high fSCNAs were characterized by HRD. In contrast, patients with low fSCNAs and high cSCNAs were characterized by homologous recombination proficiency. Patients with both low fSCNAs and cSCNAs were characterized by elevated expression of immune response and EMT.