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Biomarker of breast cancer using SVM algorithm based on WXS data

Gyu-Bum Han and **Dong Ho Cho**
Korea Advanced Institute for Science and Technology, Korea

Many researches have been done to confirm the relationship between genetic information and disease. However, since there is too much genetic information, only partial information of genome has been analyzed, which is not enough to be applied to clinical application. Also, it was difficult to identify the relationship between whole WXS data and disease. Therefore, we extract genetic mutations from WXS data and define the significance of each mutation. Then, we determine the set of mutations according to defined significance. Also, the disease and normal samples are learned through SVM algorithm, and blind samples are tested to verify the accuracy of the classification criteria which is the biomarker. We downloaded 550 WXS data related to breast cancer provided by NIH's TCGA and applied proposed method. First, we extracted mutations (SNP, insertion and deletion) from 500 learning samples (250 primary tumors and 250 blood derived normals) by using GATK. After that, we calculated significance of each mutation which is defined as the difference between disease and normal. We generated the set of mutations by varying significance values, and 500 samples were learned using SVM algorithms for each set of mutations. Finally, classification accuracy was confirmed by applying the biomarker derived from SVM learning to 50 (25 primary tumors and 25 blood derived normals) test samples. The maximum classification probability is 80%, which is obtained when significance value is 0.56 and the size of mutation set is 848. This value means relatively high accuracy compared to existing biomarker.

Biography

Gyu-Bum Han has completed his Master's degree at School of Electrical Engineering in KAIST in 2015. Currently, he is a PhD student at School of Electrical Engineering in KAIST. His research area of interest is Bioinformatics.

hgb1012@hanmail.net

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