

Detection of DNA copy number alterations using penalized least squares regression. [1]

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MOTIVATION: Genomic DNA copy number alterations are characteristic of many human diseases including cancer. Various techniques and platforms have been proposed to allow researchers to partition the whole genome into segments where copy numbers change between contiguous segments, and subsequently to quantify DNA copy number alterations. In this paper, we incorporate the spatial dependence of DNA copy number data into a regression model and formalize the detection of DNA copy number alterations as a penalized least squares regression problem. In addition, we use a stationary bootstrap approach to estimate the statistical significance and false discovery rate.

Abstract

RESULTS: The proposed method is studied by simulations and illustrated by an application to an extensively analyzed dataset in the literature. The results show that the proposed method can correctly detect the numbers and locations of the true breakpoints while appropriately controlling the false positives.

AVAILABILITY: <http://bioinformatics.med.yale.edu/DNACopyNumber> [6]

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SUPPLEMENTARY INFORMATION:

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