

Abstract to 'Genome Disorder'

Detection of some new statistical biases in next generation sequencing, Illumina platform.

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A rare mutation detection is one of the most powerful applications of next generation sequencing (NGS). Therefore the fidelity of NGS laboratory, machine and post-processing informatics becomes increasingly critical.

Here we describe a new statistical biases in high quality mismatch detection of NGS, Illumina platform. Some of these biases are already known to have dramatic effect on downstream SNP analysis. Some arrived recently, and related to PCR and library preparation errors.

We introduce novel informatics methods to confidently filter these high quality artefacts from sequencing data. We also demonstrate several new sequence quality measures of biases' detection. These artefacts are difficult (if possible) to see with conventional quality measures.