عنوان مقاله:

Dealing with Pseudogenes/Paralogs in NGS data

محل انتشار:

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خلاصه مقاله:

Pseudogene is a poor facsimile of an original protein-coding gene that has lost the ability to produce a functional protein. Paralogous genes (or paralogs) are a particular class of homologous genes, result of gene duplication, with the same or different functions. These genes are characterized by high sequence similarity with their corresponding functional genes. Due to presence of pseudogenes and paralogs in genome, ambivalent mapping in the analysis of NGS data cannot always be avoided. This fact can complicate the detection of genuine mutations residing in functional genes. Mis-mapping of reads to locations in the genome other than the specific target, lowers the variant quality score, resulting in false positive variants (erroneous variant calls) or missed causative variants. This generally makes the molecular analysis unreliable. Specific care must be taken for analysis of genes with pseudogene or paralogs in NGS data. In addition, NGS findings must be validated with another reliable technique. Here we report some cases with pathogenic / likely pathogenic variants detected in genes with pseudogenes or paralogs. The logic used in each case to detect the possible variant in NGS data will be reviewed. Further, determining true-or false-positive variants by a second technique will be presented.

کلمات کلیدی:

Next-generation sequencing (NGS), Pseudogenes, Paralogs, Mapping