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Long read variant calling protocol using Sniffles

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The method was used for a bioinformatics analysis

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Abstract

Abbreviated description of variant calling pipeline for long read whole genome sequencing data.

Prerequisites

1 Long read whole genome sequencing of adenocarcinoma, polyp, and normal samples was conducted.

Bioinformatics analysis

2 SVs were called with Sniffles v2.2 using default parameters along with the --snf tag. Software version URL: https://github.com/fritzsedlazeck/Sniffles/releases/tag/v2.2

3 To compare SV calls across the adenocarcinoma, polyp, and normal samples, Sniffles2 VCF SV files were processed through a somatic SV calling workflow with a threshold of 2 supporting reads for SV detection. A variant had to be present in 11% of the reads to be considered a somatic variant.

4 The SNF files were merged.

5 SVs for the normal, polyp, and adenocarcinoma samples were extracted using the SUPP_VEC function for BCFtools.

6 In-depth analysis of specific SVs unique to the adenocarcinoma sample was conducted by visualizing loci with Integrative Genomics Viewer (IGV).

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