Algorithms for structural variant detection

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Stellingen

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Algorithms for Structural Variant Detection

1. The model-based detection strategy proposed for short-read based structural variants detection is not applicable to complex ones because they are largely unexplored. [This thesis, Chapter 2]

2. New variations are being revealed via next-generation sequencing in low costs, but it will not be able to find all variations hidden in the genome. [This thesis, Chapter 2]

3. The graph is a biologically interpretable data structure for detecting and representing structural variants. [This thesis, Chapter 3]

4. Reproducibility is critical for applying sequencing technology in both research and clinical settings. [This thesis, Chapter 5]

5. Combining BioTech and InfoTech is the future for developing high-throughput variants analysis systems. [This thesis, Chapter 6]

6. Long-read sequencing has emerged as superior to short-read sequencing and other methods for the identification of structural variations. [De Coster et al., 2021; DOI: 10.1038/s41576-021-00367-3]

7. Future applications of routine screening of complex and balanced SVs in the clinic represent a potential and often neglected genetic disease source, a true "iceberg under water". [Zepeda-Mendoza et al., 2019; DOI: 10.1016/j.ajhg.2019.02.024]

8. Diploid genome computing and analysis are definitely the next big algorithmic problem. [Anton Bankevich et al., 2022; DOI: 10.1038/s41587-022-01220-6]

9. Rapid reduction in sequencing costs has enabled the genomic profiling of cancer-associated genes as a component of routine cancer care.

10. Trying may not succeed, but giving up is certain to fail.