SComatic: Detection of somatic mutations in high-throughput single-cell profiling data sets

Challenge

- Charecterization of somatic mutations at single-cell resolution is essential to understand cancer evolution.[1]
- Single-cell genomics often suffer from drop-outs and are not easily scalable.[2]
- Ultra-sensitive sequencing methods allow for detection of mutations at single-molecule level, however cell type information is lost.[3]
- High-throughput single-cell assays such as sc-RNA-seq and scATC-seq offer the possibility to directly detect somatic mutations without the need for complex experimental protocols, however existing algorithms rely on previously matched bulk or single-cell DNA sequencing data, which are rarely available for existing high-throughput single-cell data sets.[4]

Technology

SComatic enables the identification of somatic single-nucleotide variants (SNVs) from single-cell datasets without the need for a matched reference sample, providing a novel approach to studying somatic mutagenesis. This capability is especially valuable for analyzing cell types and samples with inherent complexities, such as differentiated cells and polyclonal tissues exhibiting significant genetic heterogeneity, such as cancer cells. Notably, SComatic demonstrates superior performance compared to existing pipelines in detecting somatic SNVs in single-cell data, facilitating the exploration of mutational processes in both cancerous and non-cancerous cells.

References

[1] https://doi.org/10.1016/j.cell.2019.06.024

[2] https://doi.org/10.1038/nrg.2015.16

[3] https://doi.org/10.1038/s41586-021-03477-4

[4] https://doi.org/10.1186/s13059-019-1863-4

Internal EMBLEM Reference

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Applications

- **Precision Medicine:** Understanding somatic mutations in individual cells can guide personalized treatment strategies and identify genetic markers for disease prognosis and treatment response.
- **Drug Discovery:** Identifying somatic mutations associated with drug resistance or sensitivity can inform drug discovery efforts and personalized treatment approaches.
- **Cancer Genomics:** High resolution characterisation of somatic mutations in cancer cells can reveal the genetic basis of cancer progression.
- **Neuroscience:** Detailed analysis of neurons can provide insights in the genetic basis of neurological disorders

Commercial Opportunity

SComatic's ability to accurately detect somatic mutations in single-cell datasets without requiring matched reference samples opens up possibilities for advancing research efforts in various sectors, such as oncology, drug development and personalized medicine.
Contact us for a quote. Our services are available to everyone, and we offer special rates for start-ups and SMEs.

Seeking

☒ Licensing

Further Reading

https://doi.org/10.1038/s41587-023-01863-z

https://github.com/cortes-ciriano-lab/SComatic

Keywords

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