Sarek, a workflow for WGS analysis of germline and somatic mutations

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Summary

Sarek

Portable WGS germline and normal/tumor pairs analysis **workflow** written in

nextflow

Easily deployable with **containers**

docker

Preprocessing based on GATK best practices

Sarek Germline

Variant Calling with:
- HaplotypeCaller
- Manta
- Strelka

Sarek Somatic

Variant Calling with:
- ASCAT
- Freebayes
- HaplotypeCaller
- Manta
- MuTect1
- MuTect2
- Strelka

Annotation with:
- snpEff
- VEP

Reports aggregated by MultiQC

Can be used on amazon web services

Open source, contribute on GitHub

Join the chat on Gitter

Acknowledgements

BARNCANCER FONDEN

UPPMAX

We present Sarek, a **portable** Open Source pipeline to resolve **germline** and **somatic** variants from WGS data: it is written in **Nextflow**[1], a domain-specific language for workflow building.

It **processes normal samples** or **normal/tumor pairs** (with the option to include matched relapses).

Sarek is **based on GATK best practices** to prepare short-read data, which is done in parallel for a tumor/normal pair sample.

After these preprocessing steps several variant callers scan the resulting BAM files:

- **Manta** for structural variants

- **Strelka** and **GATK HaplotypeCaller** for germline variants

- **Freebayes**, **MuTect1**, **MuTect2** and **Strelka** for somatic variants

- **ASCAT** to estimate sample heterogeneity, ploidy and CNVs

At the end of the analysis the resulting VCF files can be annotated to facilitate further downstream processing.

Fig1: CPU usage for 90x tumor/normal pair sample (hours)

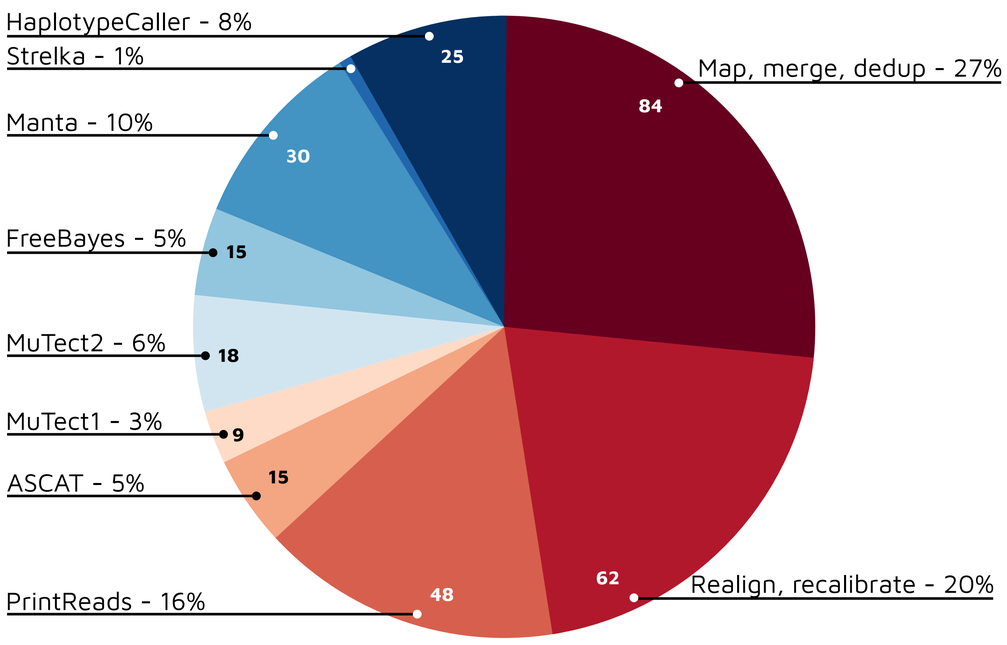
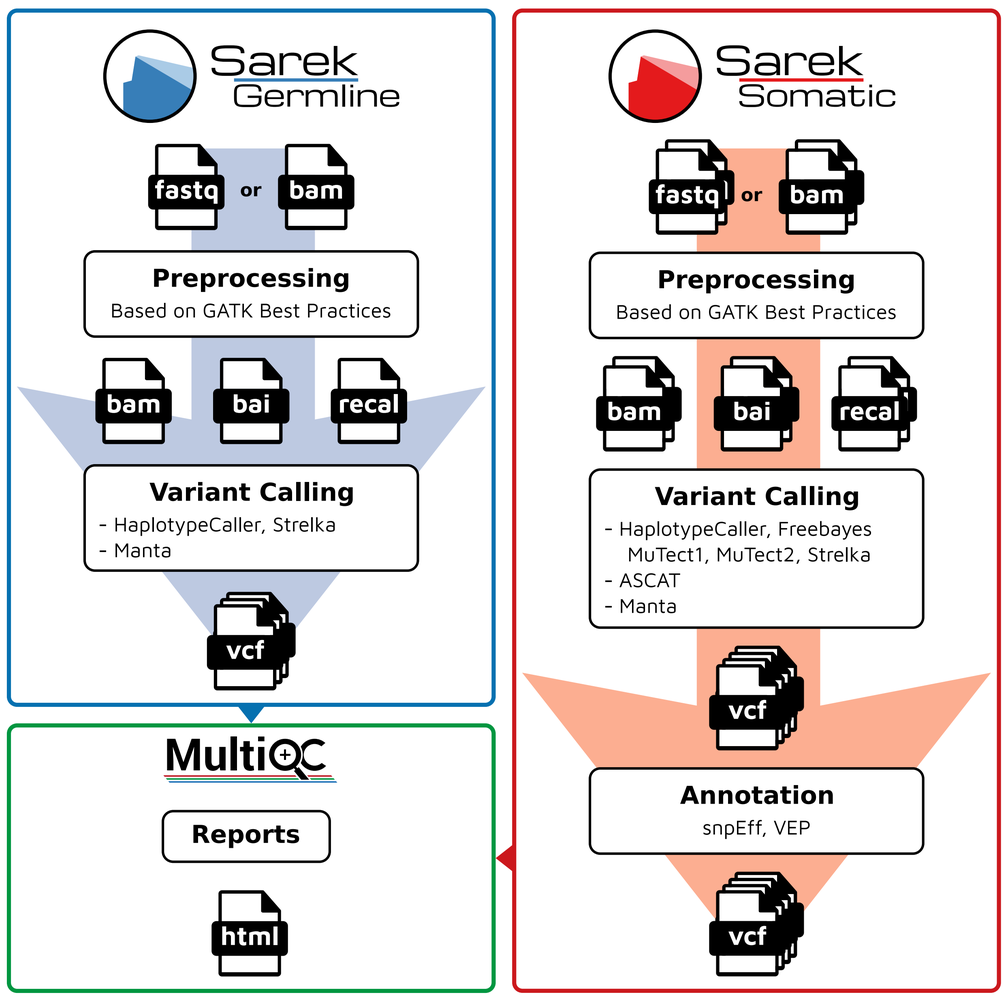


Fig2: Workflow organization



Sarek is based on **Docker** and **Singularity**[2] **containers**, enabling version tracking, reproducibility and handling sensitive data.

The workflow is capable of accommodating further variant callers.

Besides variant calls, the workflow provides quality controls presented by **MultiQC**[3].

Checkpoints allow the software to be started from FastQ, BAM or VCF.

The pipeline currently use **GRCh37** or **GRCh38** as a reference genome, it is also possible to add **custom genomes**.

The **MIT licensed** Open Source code can be downloaded from GitHub.

References

1: doi.org/10.1038/nbt.3820
2: doi.org/10.1371/journal.pone.0177459
3: doi.org/10.1093/bioinformatics/btw354

Links

opensource.scilifelab.se/projects/sarek
github.com/SciLifeLab/Sarek
gitter.im/SciLifeLab/Sarek
ngisweden.scilifelab.se

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