

Sarek

A portable workflow for WGS/WES analysis
of germline and somatic mutations

SciLifeLab

NATIONAL CTAC
ATCAGENOMICS
INFRASTRUCTURE

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What is Sarek?

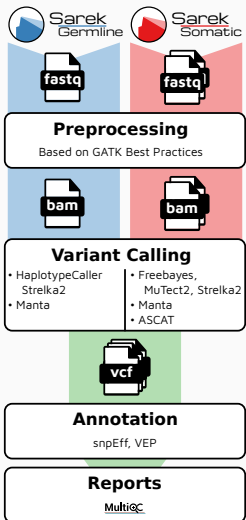


 <http://sarek.scilifelab.se/>

- Analysis germline and somatic workflow
- Whole genome or targeted sequencing
- Developed with NGI and NBIS
- Support from The Swedish Childhood Tumor Biobank



Data and files workflow






🌐 <https://software.broadinstitute.org/gatk/best-practices/>

Based on GATK Best Practices (GATK 4.0)

- Reads mapped to reference genome with `bwa`
- Duplicates marked with `picard MarkDuplicates`
- Recalibrate with `GATK BaseRecalibrator`

- SNVs and small indels:
 - HaplotypeCaller
 - Strelka2
- Structural variants:
 - Manta

- SNVs and small indels:
 - MuTect2
 - Freebayes
 - Strelka2
- Structural variants:
 - Manta
- Sample heterogeneity, ploidy and CNVs:
 - ASCAT
 - Control-FREEC ( adding)

- VEP and SnpEff
-  ClinVar, COSMIC, dbSNP, GENCODE, gnomAD, polyphen, sift, etc.

- First step towards clinical use
- Rank scores are computed for all variants
 - COSMIC, ClinVar, SweFreq and MSK-IMPACT (cancerhotspots.org)
- Findings are ranked in three tiers
 - 1st tier: well known, high-impact variants
 - 2nd tier: variants in known cancer-related genes
 - 3rd tier: the remaining variants

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Any questions?

🌐 <http://sarek.scilifelab.se/>

🐙 <https://github.com/SciLifeLab/Sarek>

