Sarek

SciLifeLab

KAROLINSKA INSTITUTET ANNO 1810
Karolinska
Institutet

DNA club

GANATIONALCTAC
ATCAGENOMICSGT
INFRASTRUCTURE

NBIS

Maxime U. Garcia
maxulysse.github.io
@MaxUlysse
@gau

Barntumörbanken

Science for Life Laboratory
Karolinska Institutet Science Par

What is Sarek?

Sarek

🌐 http://sarek.scilifelab.se/

- Analysis germline and somatic workflow

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

NATIONAL GENOMICS INFRASTRUCTURE   Barntumörbanken   NBIS

Using

nextflow

🌐 https://www.nextflow.io/

🌐 https://www.sylabs.io/singularity/

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Data-driven workflow language

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HPC specific container engine

Sarek exists in multiple flavors

Sarek

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Sarek
Germline

Sarek
Somatic

Sarek exists in multiple flavors

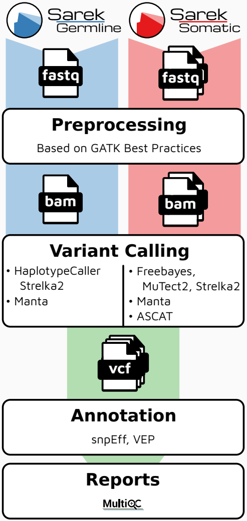
Sarek

Sarek Germline

Sarek Somatic

Sarek Exome

Data and files workflow



Preprocessing

Based on GATK Best Practices

Variant Calling

- HaplotypeCaller Strelka2
- Manta

- Freebayes, MuTect2, Strelka2
- Manta
- ASCAT

Annotation

snpEff, VEP

Reports

References Genomes

AWS iGenomes

🌐 https://ewels.github.io/AWS-iGenomes/

- Human `GRCh37`
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- Human `GRCh37`
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- Dog `CanFam3.1` 🔧
- Mouse `GRCm38` 🔧

Preprocessing

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

Based on GATK Best Practices (GATK 4.0)

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- Recalibrate with `GATK BaseRecalibrator`

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  - Control-FREEC 🔧 🔴

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- ClinVar, COSMIC, dbSNP, GENCODE, gnomAD, polyphen, sift, etc.
- Possibility to use cache directories 🔧
- Prioritization 🔧
  - Rank scores are computed for all variants, and can be explored

Singularity containers

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- Next step `Sarek` module

Conda 🔧

BIOCONDA®

🌐 https://bioconda.github.io/

Conda

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https://bioconda.github.io/

- Execute Sarek within a conda environment

AWS

🌐 https://aws.amazon.com/

AWS

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- Improving AWS usage

Acknowledgments

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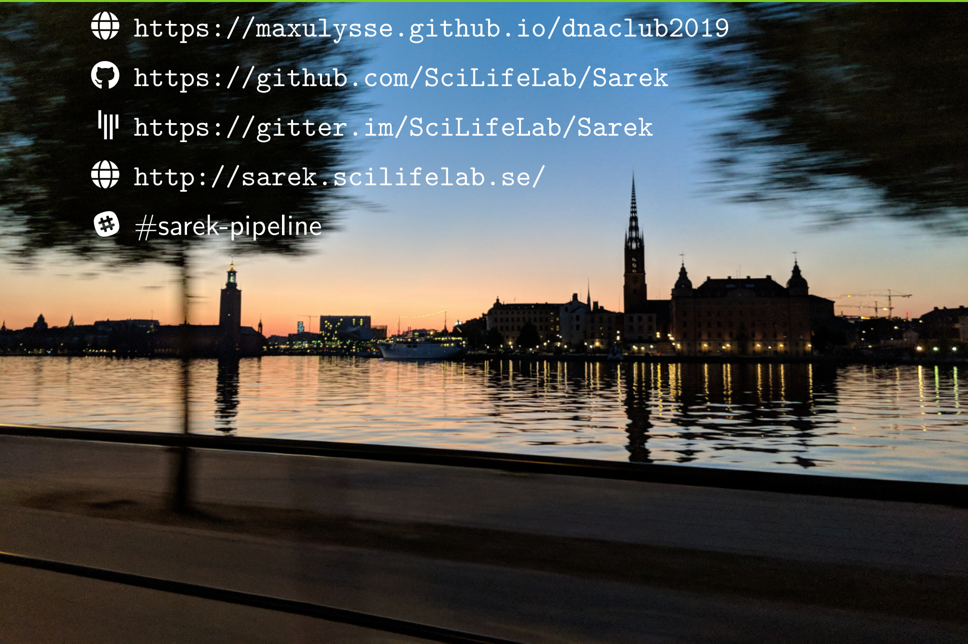
Any questions?



- https://maxulysse.github.io/dnaclub2019
- https://github.com/SciLifeLab/Sarek
- https://gitter.im/SciLifeLab/Sarek
- http://sarek.scilifelab.se/
- #sarek-pipeline