Sarek

SciLifeLab

Karolinska Institutet

GANATIONALCTAC
ATCAGENOMICSGT
INFRASTRUCTURE

NBIS

The Review

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maxulysse.github.io
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Barntumörbanken

What is Sarek?

http://sarek.scilifelab.se/

- Analysis germline and somatic workflow

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NATIONAL GENOMICS INFRASTRUCTURE

NBIS

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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

Using

https://www.nextflow.io/

https://www.sylabs.io/singularity/

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Data-driven workflow language

https://www.sylabs.io/singularity/

HPC specific container engine

Sarek exists in multiple flavors

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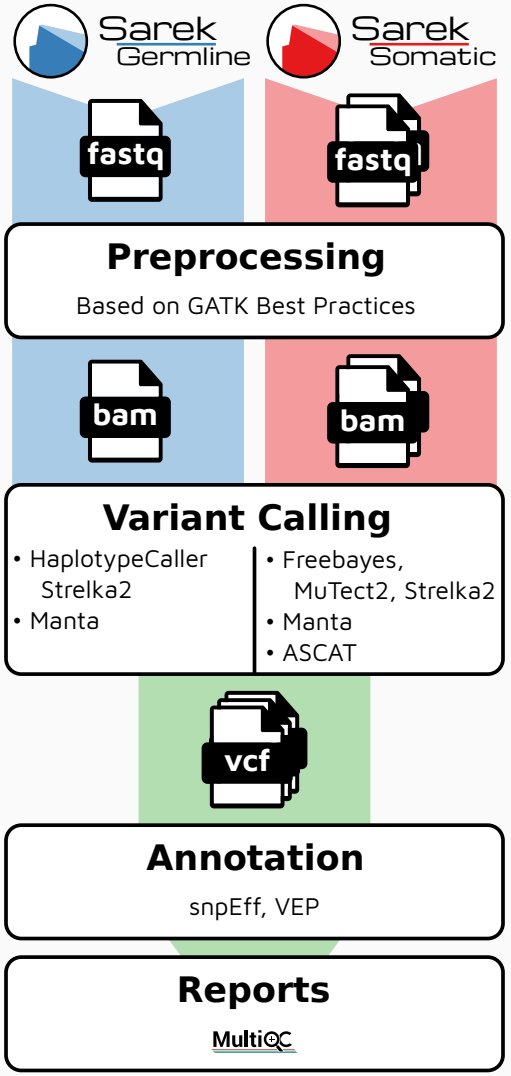
Data and files workflow



AWS iGenomes

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

- Human Grch37
- Human Grch38

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Preprocessing

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Based on GATK Best Practices (GATK 4.0)

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Based on GATK Best Practices (GATK 4.0)

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

Variant Calling

- SNVs and small indels:

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  - Freebayes 
  - HaplotypeCaller 
  - MuTect2 
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  - ASCAT 
  - Control-FREEC 

Annotation

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

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- Rank scores are computed for all variants
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- Findings are ranked in three tiers
  - 1^{st} tier: well known, high-impact variants
  - 2^{nd} tier: variants in known cancer-related genes
  - 3^{rd} tier: the remaining variants

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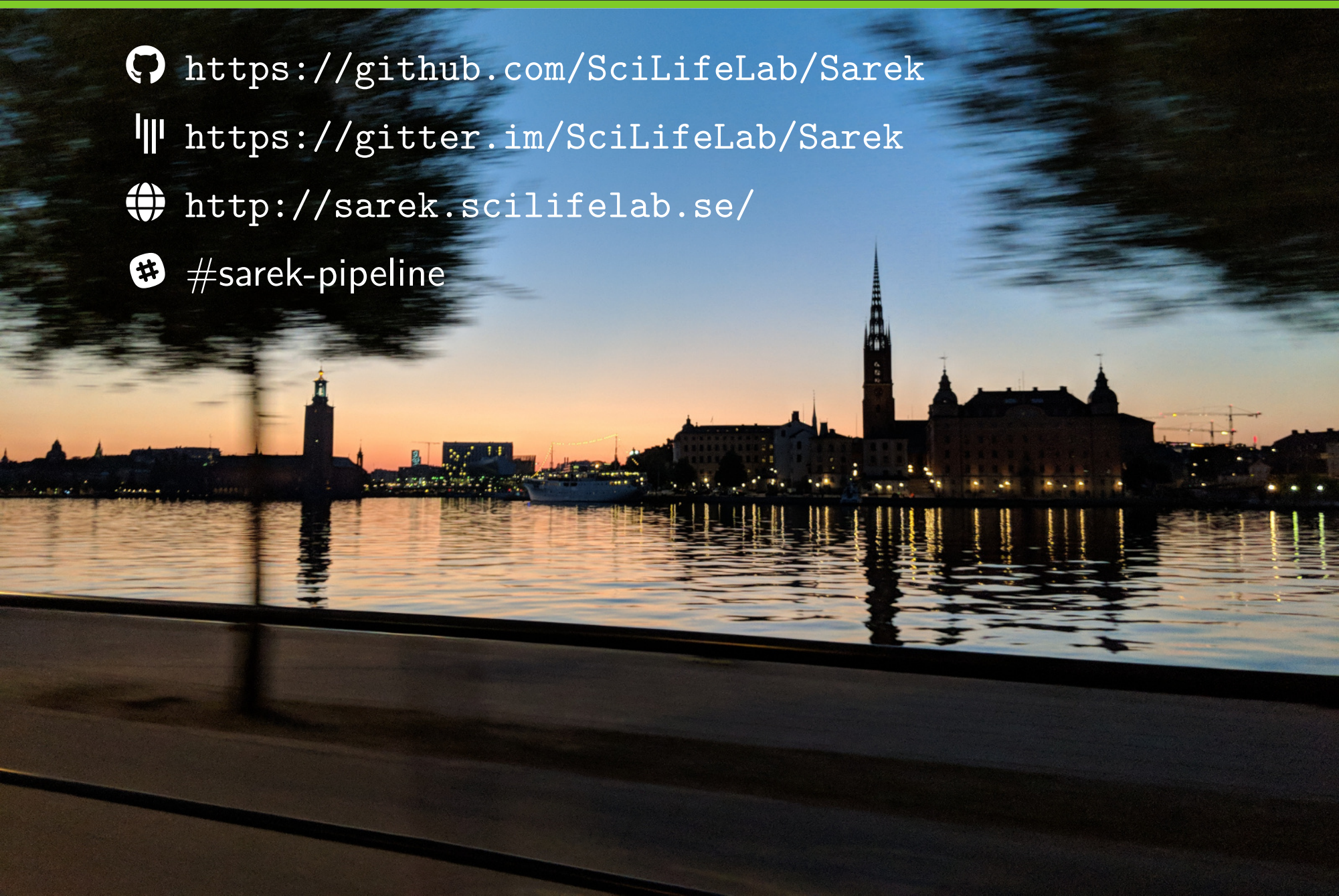
Any questions?



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https://gitter.im/SciLifeLab/Sarek

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#sarek-pipeline