Sarek and nf-core for Reproducibility SPHN Workflow Interoperability Workshop



SciLifeLab

Science for Life Laboratory



SciLifeLab is a national centre for molecular biosciences with focus on health and environmental research









UPPSALA UNIVERSITET

Science for Life Laboratory



Infrastructure Services

Genomics	Proteomics	Metabolomics
Single Cell Biology	Bioimaging and Molecular Structure	Chemical Biology and Genome Engineering
Drug Discovery	Diagnostics	Bioinformatics

National Genomics Infrastructure



https://ngisweden.scilifelab.se/

Our mission is to offer a **state-of-the-art infrastructure** for massively parallel DNA sequencing and SNP genotyping, available to researchers all over Sweden.

We provide **guidelines and support** for sample collection, study design, protocol selection and bioinformatics analysis.

National Genomics Infrastructure



https://ngisweden.scilifelab.se/





https://ngisweden.scilifelab.se/

- Stockholm
- Uppsala



https://ngisweden.scilifelab.se/

- Genomics Production Genomics Applications Development
- SNP&Seq Uppsala Genome Center

NGI Portal



https://ngisweden.scilifelab.se/



4/23

National Bioinformatics Infrastructure Sweden



https://www.nbis.se/



- Distributed infrastructure
- Swedish node in ELIXIR

Sarek



James Frain as Sarek, "Star Trek: Discovery"





Nextflow pipeline



- Nextflow pipeline
- Developed at NGI





- Nextflow pipeline
- Developed at NGI
- In collaboration with NBIS







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- Developed at NGI
- In collaboration with NBIS
- Support of The Swedish Pediatric Tumor Biobank















What does Sarek do?





- WGS analysis (Tumor/Normal pair or Germline)
 - FASTQ(s) to annotated VCF(s)
 - Multiple entry points



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- WGS analysis (Tumor/Normal pair or Germline)
 - FASTQ(s) to annotated VCF(s)
 - Multiple entry points
- Handles both GRCh37, GRCh38 and custom genomes
- Uses containers (Reproducibility, Portability)
 - Docker or Singularity

My own Computer

- My own Computer
- UPPMAX Irma NGI Production Cluster
- UPPMAX Rackham Cluster
- UPPMAX Bianca Secure Cluster

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- AWS

Any POSIX compatible system

Reproducibility

- Reproducibility
- Portability

- Easy Reproducibility
- Easy Portability























NouGAT (de-novo)

Participating Institutes

NGI-RNAseq is now used by a number of core sequencing and bioinformatics facilities. Some of these are listed below. If you use this pipeline too, please let us know in an issue and we will add you to the list.

GANATIONAL CTAC ATCAGENOMICS INFRASTRUCTURE	National Genomics Infrastructure (NGI), Sweden	https://ngisweden.scilifelab.se/
Q BiC	Quantitative Biology Center (QBiC), Germany	https://portal.qbic.uni-tuebingen.de /portal/

NGI-RNASeq

https://github.com/SciLifeLab/NGI-RNAseq





Just had a error-free run of @tallphil's RNA-Seq pipeline using @PaoloDiTommaso's @nextflowio on our LSF cluster! Started yesterday, came back today, all done! Thanks a lot guys for your great work!

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https://nf-core.github.io/

A community effort to collect a curated set of Nextflow analysis pipelines

- GitHub organisation to collect pipelines in one place
- No institute-specific branding
- Strict set of guideline requirements
- Automated testing for code style and function



https://nf-core.github.io/

All Nextflow pipelines must have:

- MIT licence
- Software bundled using Containers
- Continuous Integration testing
- Stable release tags
- Common pipeline structure and usage
- Excellent documentation























NouGAT (de-novo)

A first pipeline transfered



Phil Ewels @ewels We have a pipeline!

nf-core/ 🛨 methylseq

https://github.com/nf-core/methylseq



Sebastian DiLorenzo Jesper Eisfeldt Phil Ewels Maxime Garcia Szilveszter Juhos Max Käller Malin Larsson Marcel Martin Markus Mayrhofer Monica Nistèr Björn Nystedt Pall Olason Markus Ringnér Pelin Sahlén Johanna Sandgren Teresita Díaz De Ståhl



- The National Genomics Infrastructure (NGI), SciLifeLab, Stockholm, Sweden
- The Quantitative Biology Center (QBiC), Universität Tübingen, Germany
- The Genomics Institute of Singapore (GIS), A*STAR, Singapore

The List of People Involved



Chih ChuanDenis MorenoPaolo Di TommasoRemi-Andre OlsenPhil EwelsSenthilkumar PanneerselvamSven FilingerAlexander PeltzerMaxime GarciaChuan WangRickard HammarénAndreas Wilm

We have gitter channels
 I||I https://gitter.im/SciLifeLab/Sarek
 I||I https://gitter.im/nf-core/Lobby

- We have gitter channels
 III https://gitter.im/SciLifeLab/Sarek
 III https://gitter.im/nf-core/Lobby
- Our code is hosted on Github
 https://github.com/SciLifeLab/Sarek
 https://github.com/nf-core

