Tomte: a pipeline for RNA-seq analysis in rare disease diagnostics

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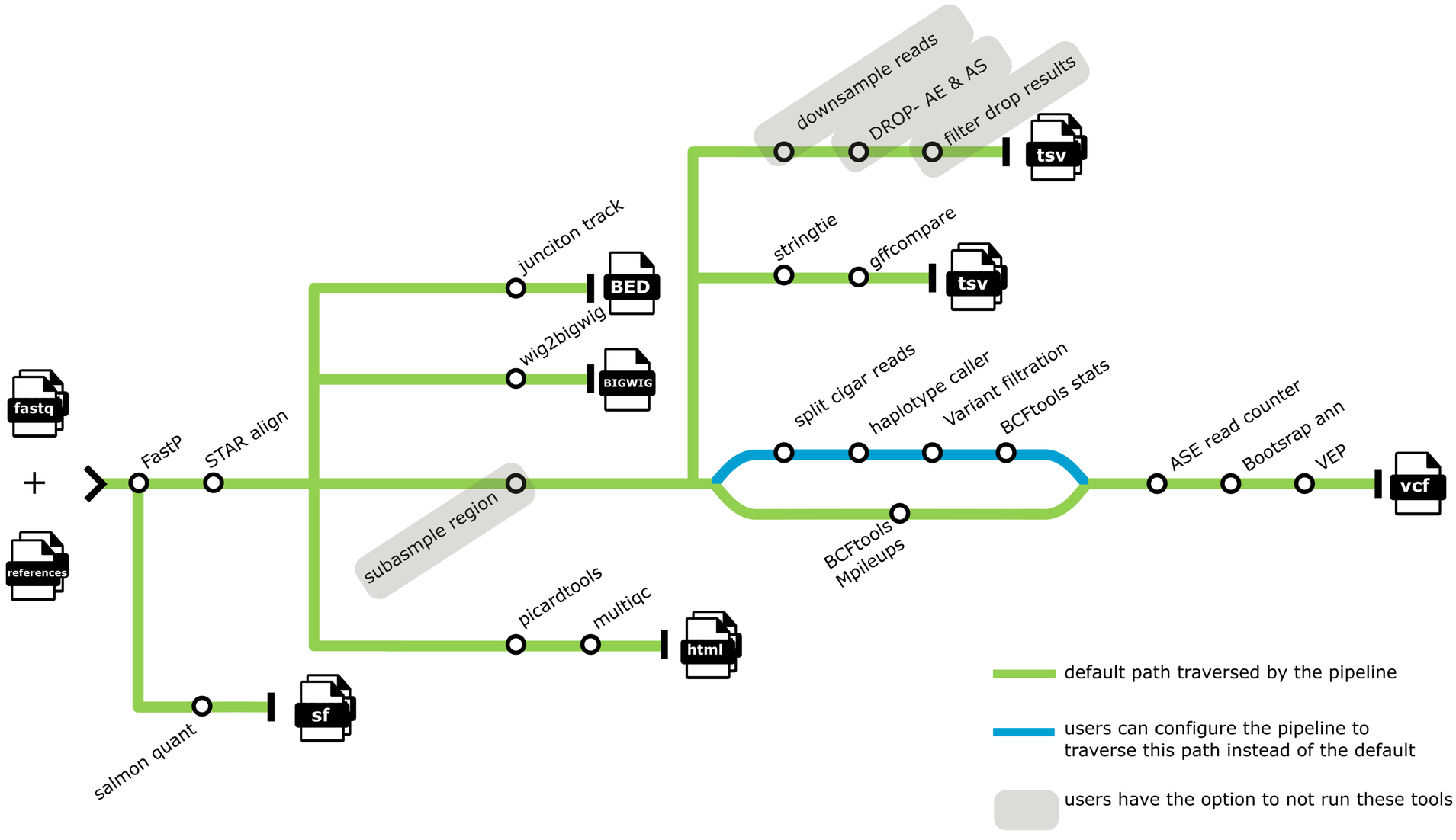
Background

- A rare disease is a condition affecting less than 1 in 2000 people
- They are rare individually but common as a whole, resulting in about 30 million people affected in the EU
- Most of these diseases have a clear genetic component but the cause of many of them remains unknown
- The diagnostic rate after performing WES or WGS is estimated at 30-50%

Motivation

- WGS analysis often yields many non-coding and deep intronic variants, the effect of which can be difficult to predicted reliably by in silico algorithms
- Whole transcriptome sequencing has shown to be a useful tool, particularly in seeing the effect of such variants in transcripts
- It allows for comprehensive detection of aberrant expression, aberrant splicing, and mono allelic expression in expressed genes

Workflow



Results & Conclusions

- Tomte is written in Nextflow DSL2 following nf-core recommendations allowing for reproducibility, portability, and continuous integration
- The pipeline will be used for investigating RNA-seq data from rare disease patient in the Stockholm healthcare region both in a research and clinical setting
- Providing a complete analysis including QCs, tracks, quantification, transcript assembly, and detection of aberrant expression, aberrant splicing, and mono-allelic expression

tomte

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