MGvizPM: Precision Medicine web reports made 'siimple'

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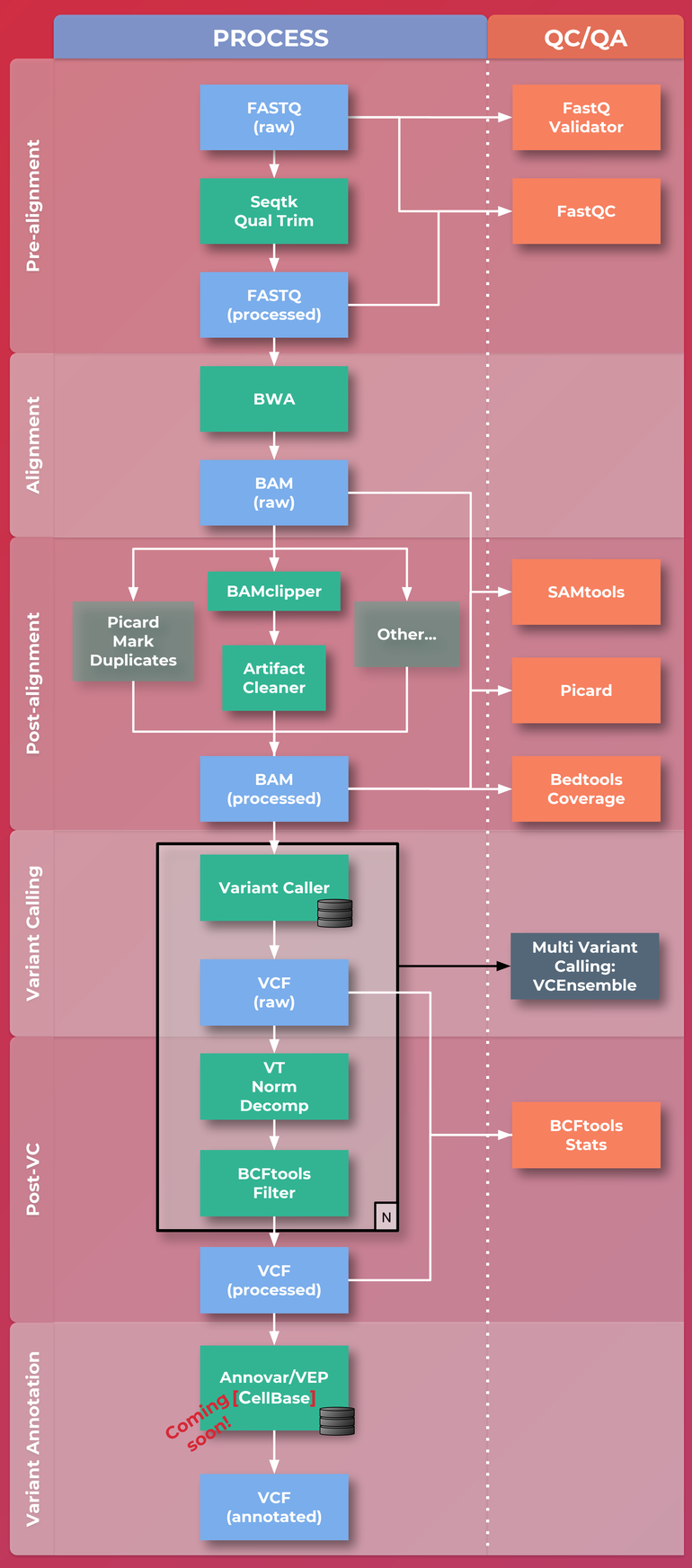
Medical Genomics Visualization Group (MGviz), Siimple OSS, Seqplexing and Kanteron Systems have jointly developed NGS data analysis workflows that create automatic technical reports for precision medicine with fully integrated QC and LIMS procedures. Our genetic and pharmacogenetic data can be easily integrated in HIS systems and use HL7 standard protocols.

We have developed a full suit of open source tools in Python, R and MERN stack for clinical bioinformatics as a service. These tools include serving variant annotation, interactive selection tools, reannotation and automatic clinical reports generation.

We are doing trials to deploy this service in a cloud platform for creating a service for analyzing customized NGS gene panels and exomes in a clinical context for diabetes, cancer and mental disabilities.

MGviz NGS pipelines



Pipeline automation following Common Workflow Language (CWL) implementations like NextFlow. Also, there are modules to implement pipelines with Snakemake or whatever workflow manager and tools the customer desires.

Summary

Clinical genomics is a reality, but better tools for creating reports without great effort and easy to interpret actionable variants are needed.

Here we present a professional platform for generating semi-automatic genetic clinical reports.

Consistency and clarity of reports are needed in order to translate new clinical genomics insight into personalized healthcare and this tool helps clinicians and genetic counselors to create these reports.

References

1. https://doi.org/10.1093/bib/bbx144
2. https://doi.org/10.1038/s41375-017-0007-7
3. http://www.nature.com/doifinder/10.1038/gim.2015.30
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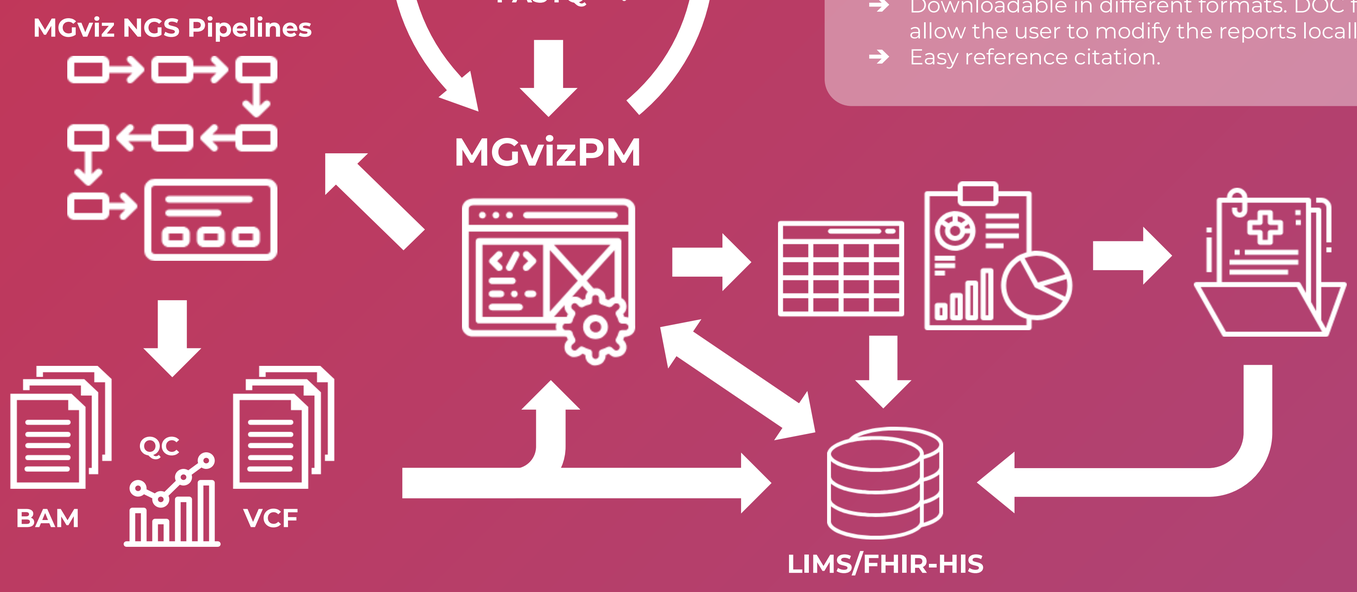
MGvizPM workflow:

1. Upload SE/PE FASTQ files.
2. Select the configuration parameters to run a NGS pipeline.
3. As the samples are analyzed, the results can be consulted.
4. QC plots can be visualized and are interactive to explore the results.
5. Filter actionable and variants of interest, annotate additional variant information (stored in the user history), consult the integrated data from different reference databases and select the variants to report.
6. Fill the report to generate the clinical-technical report, download the DOC and modify as desired.

Customized NGS pipelines:

- Optimized for each experimental design and sequencing library protocol.
- Standard and customized QC/QA steps.
- Atomized pipelines to facilitate extension.
- Scalable for production.
- Docker microservices for scalable deployment.



Variant prioritization:

- Variant annotation includes clinical, evolutionary and pharmacogenetics annotation for prioritization.
- Extended clinical information of each variant include databases such as COSMIC, ClinVar, InterVar (and other genic/project specific databases).
- Variant calling QC metrics are included to discard possible false-positive variants.
- All the information of the samples and the decisions made by the user are saved in a LIMS system for reproducibility, audits and access to the history. Data storage is compatible with FHIR-HIS system integration.

Automatic report generation:

- Fast and intuitive access to clinical database information.
- Customized templating.
- Embedded QC results.
- Downloadable in different formats. DOC files allow the user to modify the reports locally.
- Easy reference citation.

USE CASE: OGBS-TP53

http://tp53report.seqplexing.com:3001