PhenoSystems

GensearchNGS

MOLECULAR GENETICS MADE EASY

Hes·so

Haute Ecole Spécalisée de Suisse occidentale

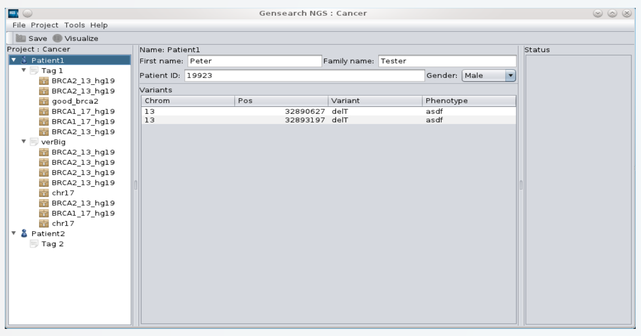
Fachhochschule Westschweiz

University of Applied Sciences Western Switzerland

Description

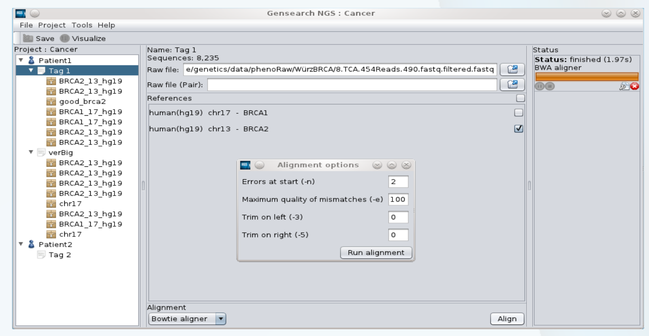
GensearchNGS integrates all steps from quality control to alignment, variant detection and visualization, variant interpretation support (UMD Predictor, Human Splice Finder, dbSNP,…) up to reporting, both internally and publicly to the Cafe Variome platform developed through Gen2Phen project.
GensearchNGS is built around the concept of plug-ins, allowing use of either proprietary tools for alignment and variant calling or any major public tool (e.g. BWA, Stampy, Bowtie, VarScan). The framework hides the complexity of those tools from the user, who doesn't need to worry about technical details such as file formats.
Also integrated is a visualization tool that visualizes the alignments done trough the framework in a fast, efficient and user friendly way.
The visualizer integrates external databases, like Ensembl, to provide the user with the needed meta information about the data he is viewing. During the visualization of a certain patient, the variants found on other patients can be displayed for comparison purposes. Those variants can also be filtered, to minimize the number of variants to check manually.
From the start, GensearchNGS was developed as a multiplatform application, running under Windows and Linux.

Overview



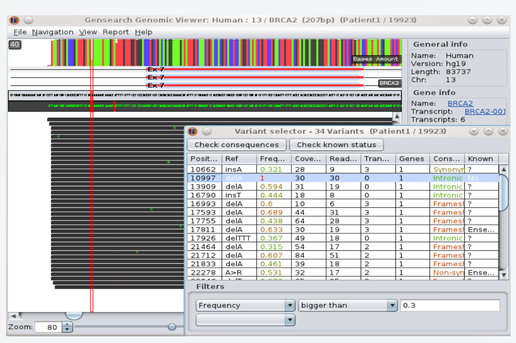
Patient management

GensearchNGS lets you manage your patients and their variants trough one interface, with direct access to the data used for the analysis. Patient reports can be created directly from within the application and the Patients separated over multiple Projects. A central variant database stores the Variants for all Patients.



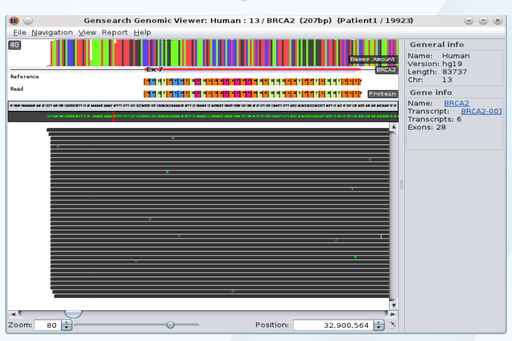
Sequence alignment

GensearchNGS allows you to align your raw sequencing data coming out of Illumina, Roche-454 or other popular sequencers in a easy way, using one of several integrated aligners. Several aligners are offered, such as BWA, Bowtie, Stampy and a custom developed aligner. During the alignment process, you can continue to work with your data and visualize it as soon as it is ready.



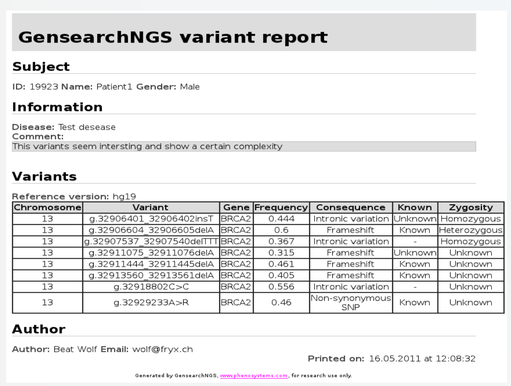
Variant analysis

GensearchNGS allows you to scan your next generation sequencing data for Variants and easily access them.
The worst consequence on all genes affected by a variant is calculated, highlighting the critical variants.
In a transparent way, public variant databases like ensembl, are queried to see if a detected variant is already known.
To find the most interesting variants, filtering allows to restrict the amount of variants shown, to find the most important ones.



Data visualization

The aligned data can be visualized in a fast and easy manner. External meta information, like genes, is integrated and directly displayed together with the data to analyze, allowing for an easy understanding functional consequences without having to gather manually information from different sources. The visualizer uses the meta data to predict the consequences of the different variants.



GensearchNGS variant report

Subject

ID: 19923 **Name:** Patient1 **Gender:** Male

Information

Disease: Test desease
Comment:
This variants seem intersting and show a certain complexity

Variants

Reference version: hg19

Chromosome	Variant	Gene	Frequency	Consequence	Known	Zygosity
13	g.32906401_32906402insT	BRCA2	0.444	Intronic variation	Unknown	Homozygous
13	g.32906604_32906605delA	BRCA2	0.6	Frameshift	Known	Heterozygous
13	g.32907537_32907540delTTTT	BRCA2	0.367	Intronic variation	-	Homozygous
13	g.32911075_32911076delA	BRCA2	0.315	Frameshift	Unknown	Unknown
13	g.32911444_32911445delA	BRCA2	0.461	Frameshift	Known	Unknown
13	g.32913560_32913561delA	BRCA2	0.405	Frameshift	Known	Unknown
13	g.32918802C>C	BRCA2	0.556	Intronic variation	-	Unknown
13	g.32929233A>R	BRCA2	0.46	Non-synonymous SNP	Known	Unknown

Author

Author: Beat Wolf **Email:** wolf@fryx.ch

Printed on: 16.05.2011 at 12:08:32

Generated by GensearchNGS, www.phenosystems.com, for research use only.

Report generation

After identifying the interesting variants, a report can be generated. The report can either be saved locally, or be printed. The information contained in the report can be sent to an online variant database like Cafe Variome.

Practical example

GensearchNGS is already used in a production environment. The intuitive user interface allowed the laboratory of the University of Würzburg, which never used next generation sequencing before to analyze several Gigabytes of raw data of several patients and identify several potential variants related to a certain disease.
The ability to quickly visualize the aligned sequencing data and filter the Variants present in each patient, helped to cut down the potential disease causing variants. The ability to filter out variants also present in other patients with the same disease, helped to track down the few interesting variants.

Partners

Hes·so

NMDCHIP

Cafe Variome

GEN2PHEN

BIOZENTRUM UNIVERSITÄT WÜRZBURG

Team

Beat Wolf
David Atlan
Pierre Kuonen
Simone Rost

The research leading to these results has received funding from the European Community's Seventh Framework Programme (FP7/2007-2013) under grant agreements: n°223026 NMDchip and n°200754 Gen2Phen