GensearchNGS-Viewer

A complete NGS data visualization experience

B. Wolf, P. Kuonen, *University of Applied Sciences Western Switzerland*
T. Dandekar, *University of Würzburg, Germany*

Introduction

We present GensearchNGSViewer, an interactive next generation sequencing alignment viewer with the primary goal of being user friendly and to help the user to quickly visualize interesting features in the data. GensearchNGSViewer is a multiplatform application running on all major operating systems, able to visualize local and remote data. The visualized data can be enhanced through annotations provided by the ensembl webservice, allowing for easier and quicker analysis of interesting features. The primary usecase for GensearchNGSViewer is the visualization and annotation of variants. Most features are based around this usecase, including interactive variant filtering and visualization of variants loaded from a standard VCF file.

Features

- DNA and RNA alignment visualization (BAM files)
- Standard annotation formats, VCF, BED, WIG, GFF

- Genome annotation import through ensembl
  - -Gene and transcript locations
  - -Variant annotations

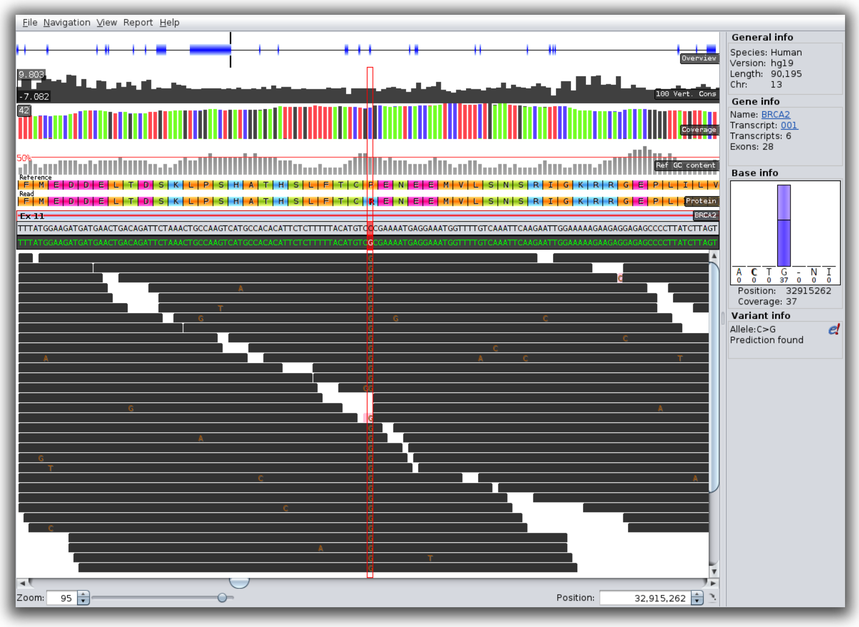
- Interactive variant file filtering
- Live variant effect prediction
- External annotation files (BED, WIG, GEF)
- Variant (vcf, csv, html) and coverage (html) reports

Availability

GensearchNGSViewer is for the moment a closed source application, but freely available for non profit use at:
http://viewer.phenosystems.com/

It can be run directly from the website be downloaded locally.

The freely available version does have some restricted functionality, it does for example not contain the report generation capabilities.

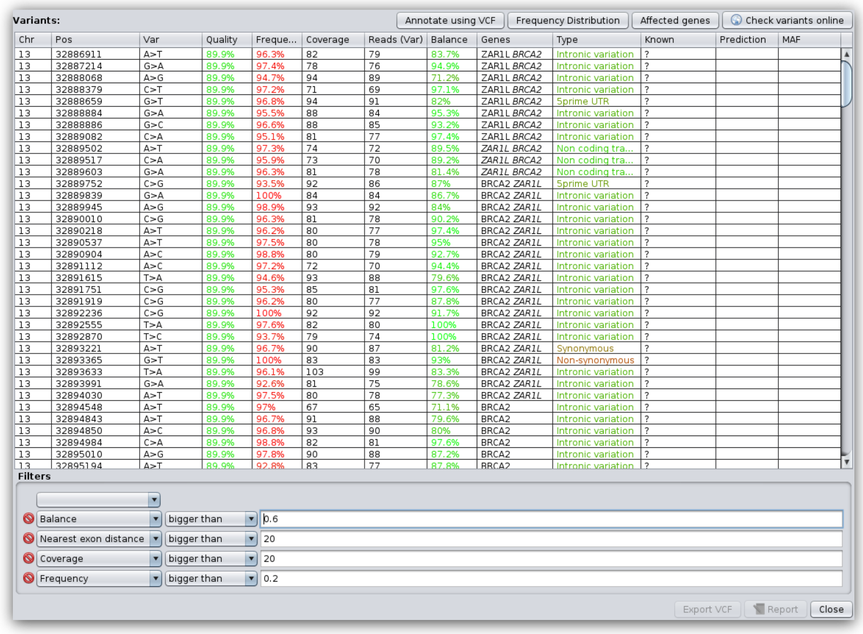


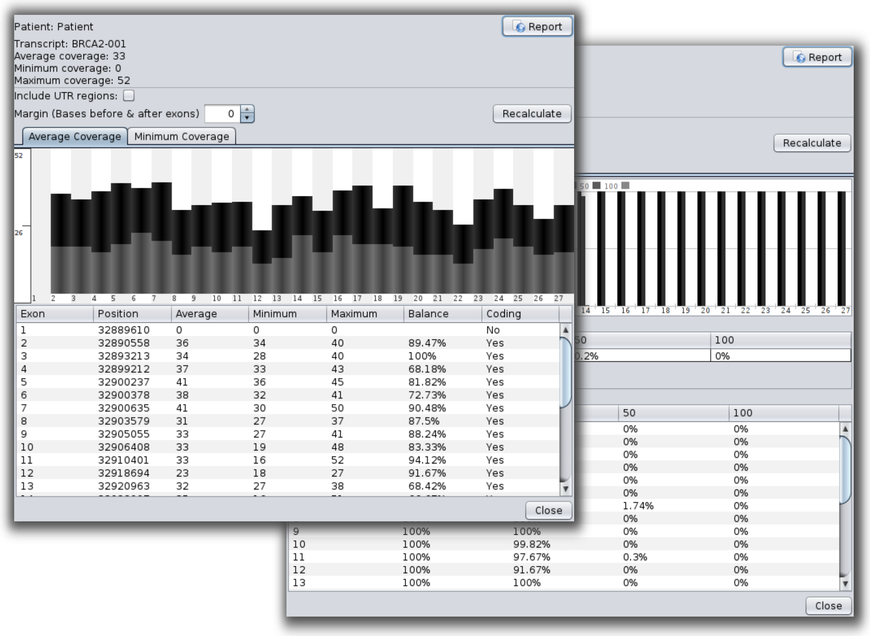
Data visualization

Easy and fast visualization of complex DNA or RNA sequence alignments loaded from BAM files with a transparent integration of genome annotations through the ensembl webservice and external files.

Variant display

Interactive variant filtering and annotation, including effect prediction and annotations through ensembl. The user can interactively filter variants to find those relevant to him.



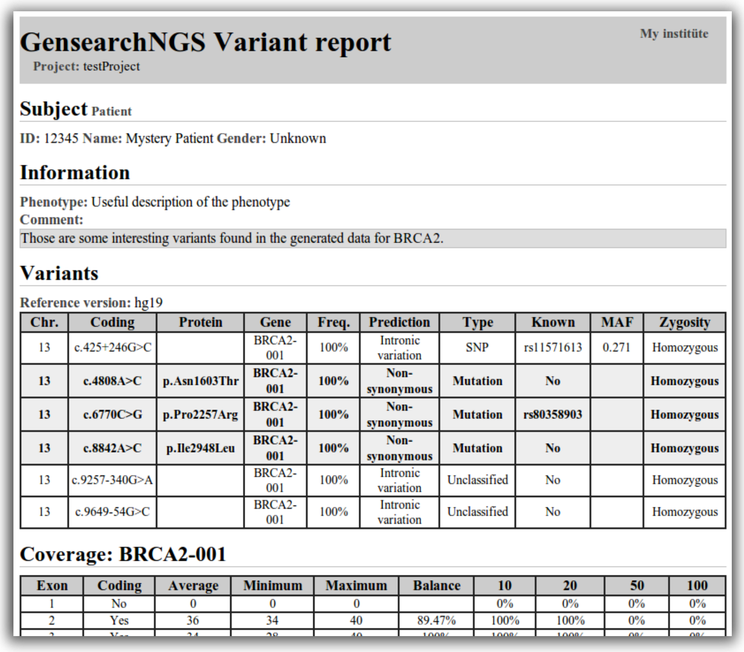


Coverage details

Detailed coverage details for user selected transcripts, gives a quick and detailed overview of the forward and backward coverage of coding or non coding areas of any transcript.

Report generation

Fast detailed report generation for user selected variants and coverage of selected transcripts. The reports can be exported in different formats, such as HTML, VCF and CSV.



GensearchNGS Variant report
Project: testProject
My institüte

Subject Patient
ID: 12345 Name: Mystery Patient Gender: Unknown

Information
Phenotype: Useful description of the phenotype
Comment:
Those are some interesting variants found in the generated data for BRCA2.

Variants
Reference version: hg19

Chr.	Coding	Protein	Gene	Freq.	Prediction	Type	Known	MAF	Zygosity
13	c.425+246G>C		BRCA2-001	100%	Intronic variation	SNP	rs11571613	0.271	Homozygous
13	**c.4808A>C**	**p.Asn1603Thr**	**BRCA2-001**	**100%**	**Non-synonymous**	**Mutation**	**No**		**Homozygous**
13	**c.6770C>G**	**p.Pro2257Arg**	**BRCA2-001**	**100%**	**Non-synonymous**	**Mutation**	**rs80358903**		**Homozygous**
13	**c.8842A>C**	**p.Ile2948Leu**	**BRCA2-001**	**100%**	**Non-synonymous**	**Mutation**	**No**		**Homozygous**
13	c.9257-340G>A		BRCA2-001	100%	Intronic variation	Unclassified	No		Homozygous
13	c.9649-54G>C		BRCA2-001	100%	Intronic variation	Unclassified	No		Homozygous

Coverage: BRCA2-001

Exon	Coding	Average	Minimum	Maximum	Balance	10	20	50	100
1	No	0	0	0		0%	0%	0%	0%
2	Yes	36	34	40	89.47%	100%	100%	0%	0%

Contact: Beat wolf, beat.wolf@hefr.ch
Website GensearchNGS-Viewer: http://viewer.phenosystems.com/
Website GensearchNGS: http://www.phenosystems.com/

PhenoSystems
GensearchNGS
MOLECULAR GENETICS MADE EASY

Hes·so FRIBOURG FREIBURG
Haute Ecole Spécialisée de Suisse occidentale
Fachhochschule Westschweiz
University of Applied Sciences Western Switzerland

BIOZENTRUM UNIVERSITAT WÜRZBURG