Finalist DX

TARGETED SOLUTION FOR ANALYSIS OF DIAGNOSTIC NGS DATA

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INTRODUCTION TO FinalistDX

FinalistDX is a powerful complete computing package designed for analysis of sequencing data produced by any of the Illumina NGS sequencing platforms such as low throughput systems MiniSeq and MiSeq as well as high-throughput NextSeq and HiSeq. FinalistDX can also analyze data from any other sequencer that generate FASTQ files. FinalistDX consists of a desktop computer and analysis program running on Linux Operating System (Ubuntu 14.04 LTS). Software provides a friendly graphical user interface even for inexperienced users such as biologists or clinicians. FinalistDX is connected to the sequencer machine and serves at the same time as a backup server.

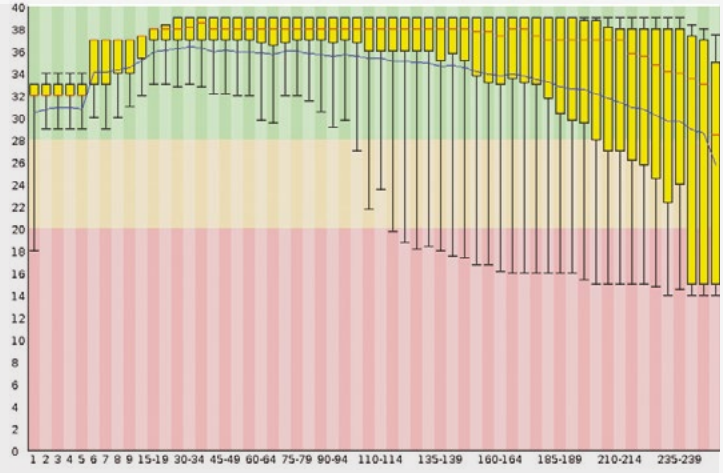
SOFTWARE MODULES

FinalistDX represents integrated bioinformatics workflow which includes quality control, alignment, visualization, coverage analysis, variant calling and variant annotation tools.

FinalistDX main window.

QUALITY CONTROL

Generate quality control plots for evaluation of sequence runs at one click using popular open source software FastQC. Users can check for sequencing quality before starting the alignment step.



Sequencing quality check before alignment step.

Sequence	Count	Percentage	Possible Source
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGACCGATCT	8122	8.122	Illumina Paired End PCR Primer 2 (100% over 40bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGATCGGAAG	5086	5.086	Illumina Paired End PCR Primer 2 (97% over 36bp)
AATGATACGGCGACCACCGAGATCTACACTCTTTCCCTAC	1085	1.085	Illumina Single End PCR Primer 1 (100% over 40bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGACCGGAAG	508	0.508	Illumina Paired End PCR Primer 2 (97% over 36bp)
AATTATACGGCGACCACCGAGATCTACACTCTTTCCCTAC	242	0.242	Illumina Single End PCR Primer 1 (97% over 40bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAAGATCGGAA	235	0.23500000000000001	Illumina Paired End Adapter 2 (96% over 31bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCGAGATCGGAAGA	228	0.22799999999999998	Illumina Paired End Adapter 2 (96% over 28bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGACCGGACG	205	0.20500000000000002	Illumina Paired End PCR Primer 2 (97% over 36bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGGATCGGAA	183	0.183	Illumina Paired End Adapter 2 (100% over 32bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGGTCGGAAG	183	0.183	Illumina Paired End Adapter 2 (100% over 32bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGACCGAACT	164	0.164	Illumina Paired End PCR Primer 2 (97% over 40bp)
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGACCGGTCT	129	0.129	Illumina Paired End PCR Primer 2 (97% over 40bp)
AATTATACTTCTACCACCTATATCTACACTCTTTCCCTAC	123	0.123	No Hit
GATCGGAAGAGCGGTTCAGCAGGAATGCCGAGACCGGACT	122	0.122	Illumina Paired End PCR Primer 2 (97% over 36bp)
CGGTTCAGCAGGAATGCCGAGATCGGAAGAGCGGTTCAGC	113	0.11299999999999999	Illumina Paired End PCR Primer 2 (96% over 25bp)

Adapter contamination check.

No Need to Read Lenghty User Guides

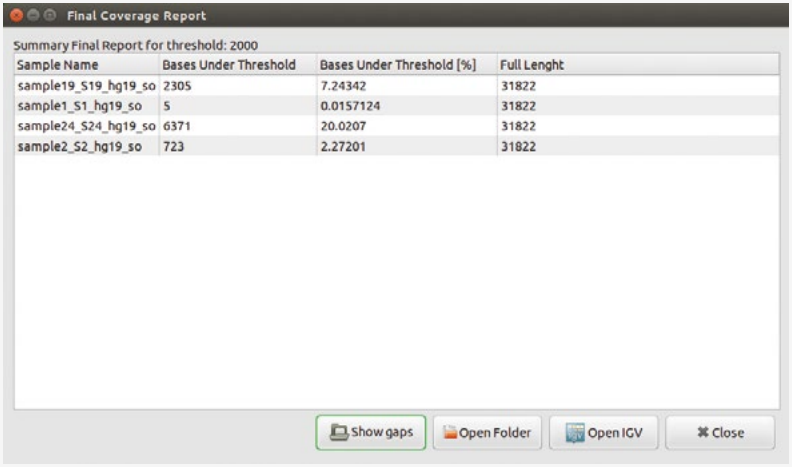
Control all Your Analysis Input Parameters

ALIGNMENT

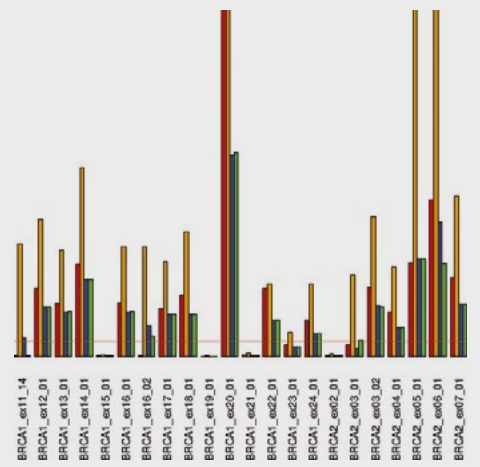
FinalistDX is designed to align reads ranging from 70 bp to 1 Mbp against a reference genome and supports split alignments. Human genomes hg19 and hg38 were selected as standard references. The alignment process is running in parallel using all available cores that make computation time very short.

COVERAGE QUALITY ANALYSIS

Users can evaluate coverage in details across all samples, looking at single-base coverage data. FinalistDX provides full information about base by base coverage for any selected region, minimum and maximum coverage and automatically notifies if coverage falls under threshold in any selected region or base. The software contains predefined selection of BED files to limit the analysis and also supports import of external BED files.



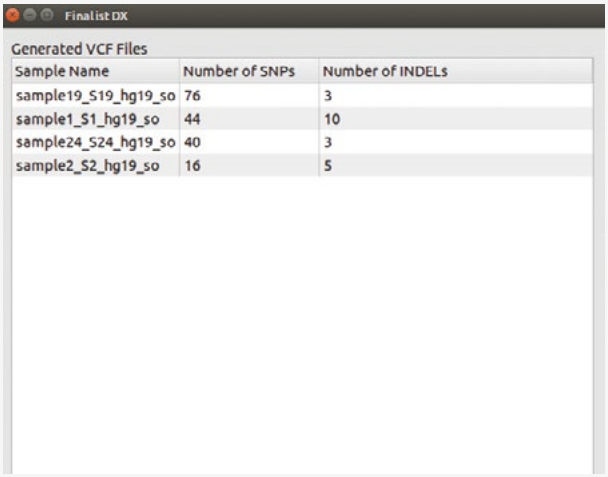
Sequencing coverage evaluation for all samples.



Coverage visualization across selected regions.

VARIANT CALLING

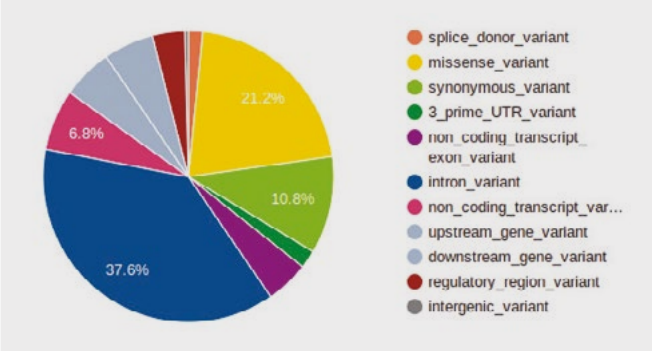
FinalistDX employs a robust statistic/heuristic algorithm for SNP/INDEL detection that meets desired thresholds for statistical significance, variant allele frequency, read depth and base quality.



Variant calling overall statistics.

VARIANT ANNOTATION

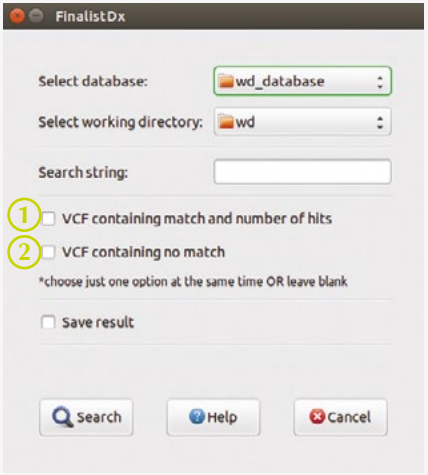
FinalistDX gives variants a meaning based on annotation and additional identifiers on the level of nucleotides, proteins, transcripts or genes. It enables users to select between Ensembl or RefSeq preinstalled off-line databases (hg19 and hg38) or to import their own databases. As an example, annotation can provide information about gene symbol, CCDS, protein identifier, Uniprot and dbSNP number, HGVS notation, exon/intron, codon, cds and strand orientation, transcript biotype, protein domains, SIFT and PolyPhen predictions, GMAF, identifies canonical transcripts, and many other.



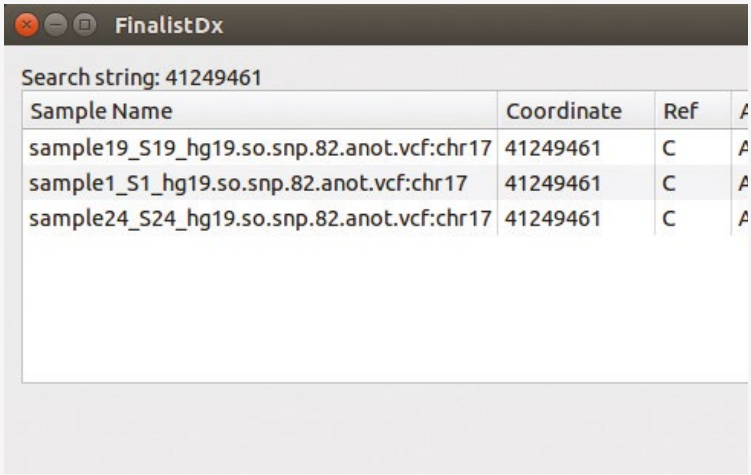
Variant consequence chart.

VARIANT DATABASE

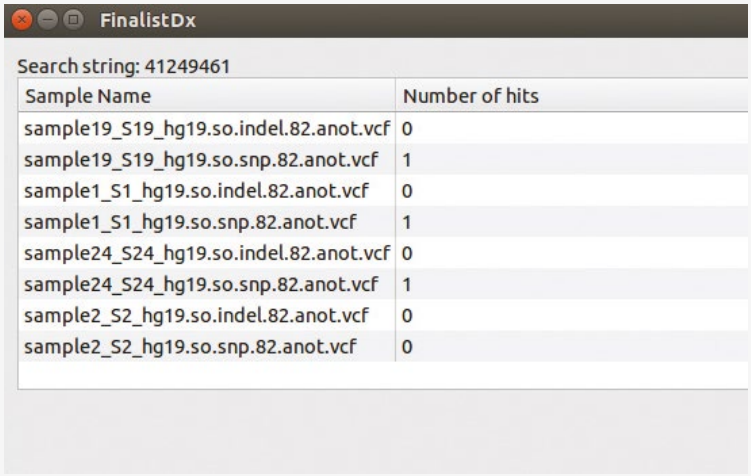
Annotated results in VCF files are stored in a searchable database in user-defined folders to easily find common and different variants according to the selected criteria (by coordinates, gene name, HGVS, rs number and others).



Variants comparison by different searching criteria.



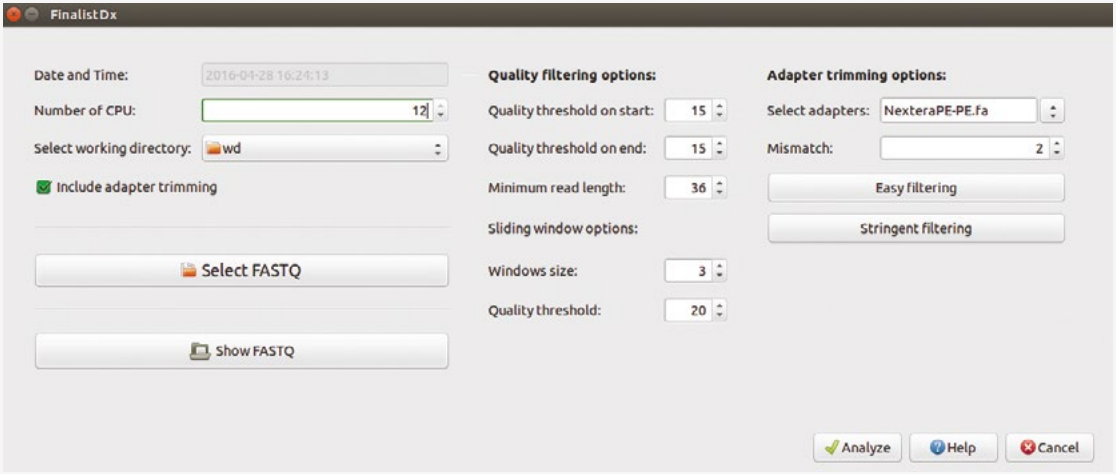
Comparison Result.



Comparison Result.

TOOLS

FinalistDX can filter FASTQ files based on Phred quality score and supports adapter trimming, preventing low quality bases and adapters to be included into analysis.



Base quality and adapter filtering options.

Free Technical and Data Analysis Support

Easy to read outputs

FinalistDX MINIMUM HARDWARE SPECIFICATION: Procesor: Intel Xeon E5-2620v2, 6 xcores (12 threads), 12 x 2.4 GHz; HDD: 2 TB, 7200 RPM; SSD: 250 GB; Monitor: AOC 27”

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