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Qlucore launches Next Generation Sequencing (NGS) Module

<u>Qlucore</u>, a leader in the development of bioinformatics visualization software, today announces its beta version of Qlucore Omics Explorer 3.3. This includes a completely new Next Generation Sequencing (NGS) Module, and range of new functionalities for the base version.

The NGS Module will enable additional functionality related to data generated with NGS technologies which will make it possible to interactively and dynamically visualize, analyze and explore NGS data.

Qlucore Omics Explorer is a next-generation bioinformatics software program for broad usage known for its friendly user interface and advanced visualization capabilities. With the Qlucore Omics Explorer NGS module it is now possible for biologists and researchers who are not data analysis experts to explore NGS and data, find new answers and generate new hypotheses.

RNA-seq studies is one of the most used techniques in modern genetics and cancer research, and the NGS Module enables a comprehensive and synchronized view of both expression levels and genomic locations. A unique flexibility is handed to the user who can perform a statistical test in the expression space by simply moving a cutoff slider, observing the result in the Genome browser.

One of the main components in the NGS Module is the innovative Genome browser which, in combination with the dynamic filters, ensures that the user is always visualizing the most interesting regions of the genome which minimizes the need for zoom and scroll.

Many users in the NGS space today are prevented from undertaking analysis due to the requirements of servers and high performance computers. Qlucore introduces an innovation with assumption free pre-processing. Work is divided in two phases. The pre-process takes time but is only done once and can be run on any local computer or server running Windows or Mac. Visualization and Analysis are performed as a second phase on a normal computer (PC or Mac) which utilizes the pre-processed information.

The main components of the NGS module are:

- A project manager for project set-up and definition of which files and samples to include.
- A built in variant caller for short indels and variants.
- An interactive and fast Genome browser for many samples and many tracks per sample
- A genome filter control component for dynamic filtering

Workflow and data storage

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The project files and associated data for indexing samples are stored locally on the user's computer to secure fast access and interactivity, whilst large files such as BAM files can be stored anywhere on a network.

RNA-seq analysis

The options available for RNA-seq analysis stand out. Utilization of the existing functionality in QOE for expression data and combining it with the new NGS functionality enable significantly increased options for analysis.

When a RNA-seq project is initiated, the project manager will (during the pre-process stage) calculate both quantitative normalized expression levels as well as prepare the data for the genome browser. The program is built to seamlessly handle information both from a quantitative and genomic view. As an example the user can define discriminating genes with a t-test, visualize them in a heat map and at then study the genes in the genome browser. If a sample group is selected or deselected all samples in both plots will be updated and analysis can continue without any requirements or restarts.

Data and file types

A reference genome is mandatory. All organisms are supported provided a reference genome expressed with the .fasta file format is available.

Variant calling

Variant calling can be done with the inbuilt variant caller which uses Samtools (<u>http://www.htslib.org/</u>) or the user can load a VCF file from another variant caller.

Filters

The filter component is flexible and lets the user combine multiple filters to select the relevant regions to analyze. The filter can be applied to calculated values such as read coverage as well as presence and matches with certain names. An example is: To create a combination of filters on parameters such as variants being present in the VCF file and named "A" and not present in the VCF file named "B" and combine this with a requirement that all displayed bases shall have a read coverage of at least 50.

Genome browser

The visualization options and possibilities of the "to do" selections are extensive in the fast and interactive genome browser. Example of options are:

- Display one or several samples
- Add and display any number of annotation tracks

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- Navigation tracks for improved orientation in three levels (whole genome, chromosome and gene)
- Tracks for all accepted data types showing for instance deletions and insertions
- Tracks with coding regions, genes, introns, exons etc. extracted from GTF file
- Search
- Zoom
- Padding to control how many filtered bases are to be shown to make sure that edge effects are not affecting the analysis
- Detailed information on selected features in the annotations

Exports

- High quality 2-D graphics exports of the genome browser content in png, jpg, bmp, TIFF or pdf (vector graphics).
- Report list of variants in the browser and the corresponding information such as BED and Cytoband. HGVS (http://varnomen.hgvs.org) strings are provided as well as reference sequence specifiers missing and protein levels.

Sample management

- Samples can be organized into groups using sample annotations.
- Active/visible samples can be managed using sample annotations.
- Import of sample annotation files (*.txt, *.csv)