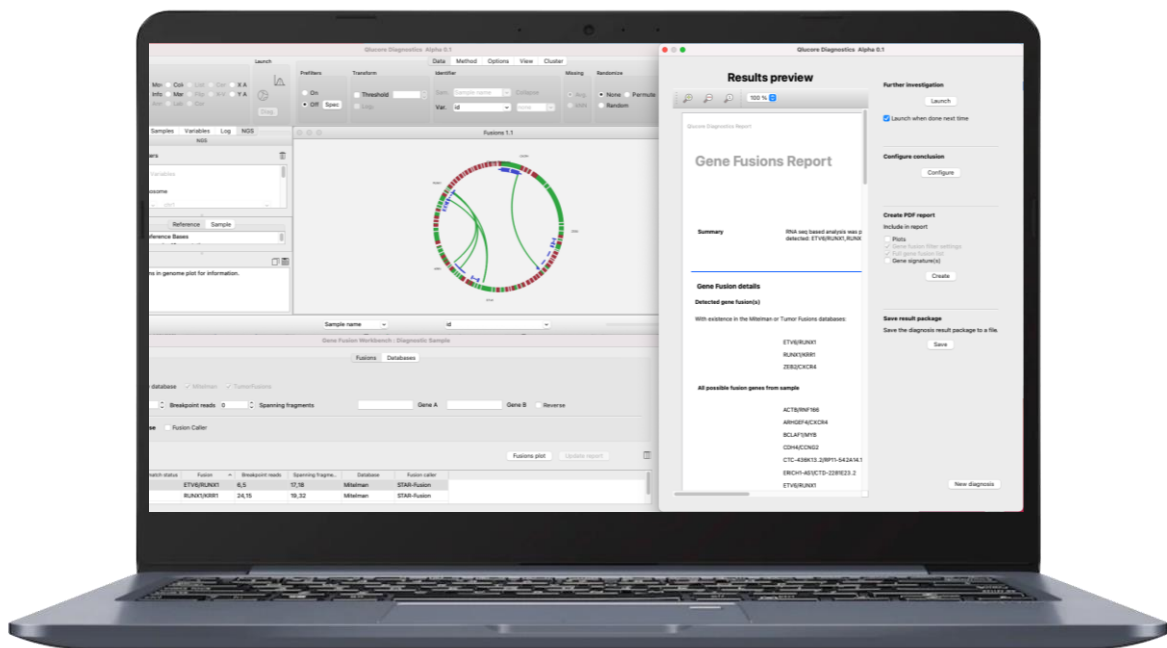


# GETTING STARTED MANUAL



## GENE FUSION MODELS

**For research use only**

## GETTING STARTED

This document offers help to get started using the Gene fusion model. It will go through the basic steps required to generate a report with detected gene fusions. It is not a comprehensive manual and some basic information about the windows and the overall layout of functionality in the Qlucore software is assumed.

### SELECT DATA

The Qlucore software processes one sample (patient) at a time and the first step is to define the data that belongs to a certain sample.

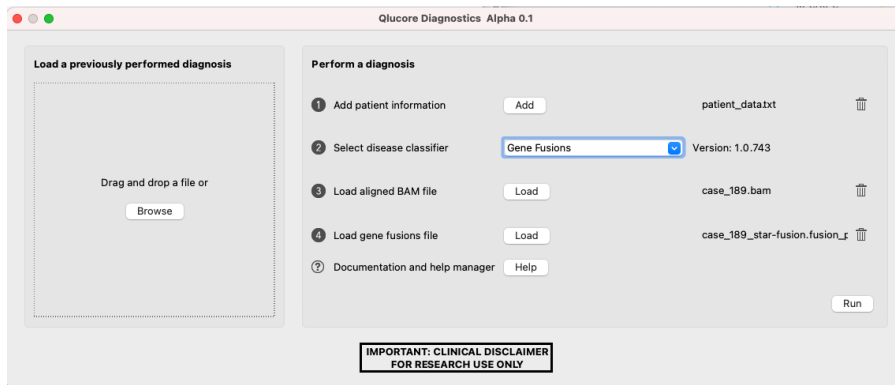


Figure 1. Data setup

1. Add the **Patient information** data (.txt file). The file content and structure are presented in an example file: patient.txt. If you do not have it, request it from your Qlucore representative.
2. Select disease analysis model **Gene fusions**.
3. Load the **RNA seq data** file (.bam file).
4. Load the **gene fusion data** file (.tsv or .txt file).
5. When all data has been specified, analysis can be started with the **'Run' button** in the lower right corner of the window.
6. Note: By pressing the **'Help' button** at the bottom you reach the Documentation and Help system and can obtain additional information.

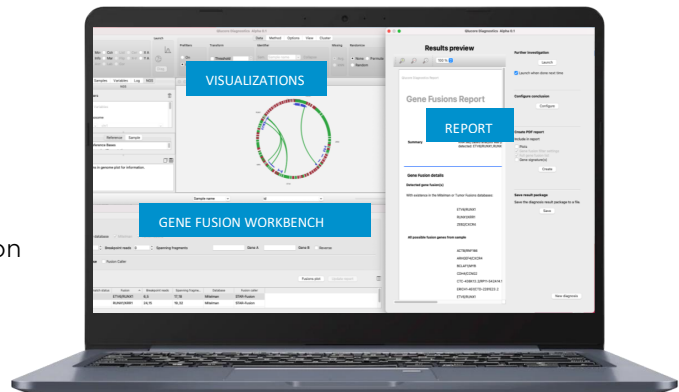
The analysis takes a few minutes.

When the analysis has been performed, the result is presented on the screen, in a set-up with multiple window components.

Figure 2. Three main components

The main components are (Figure 2):

1. Gene Fusion Workbench
2. Visualizations
3. Result preview and report generation



## GENE FUSIONS WORKBENCH

After the sample has been processed it is recommended to start investigating the detected fusions in the workbench. The Gene Fusion Workbench include several filter options and a flexible table that can include a rich set of columns based on available information and your requirement. Use the column selector to define what to show.

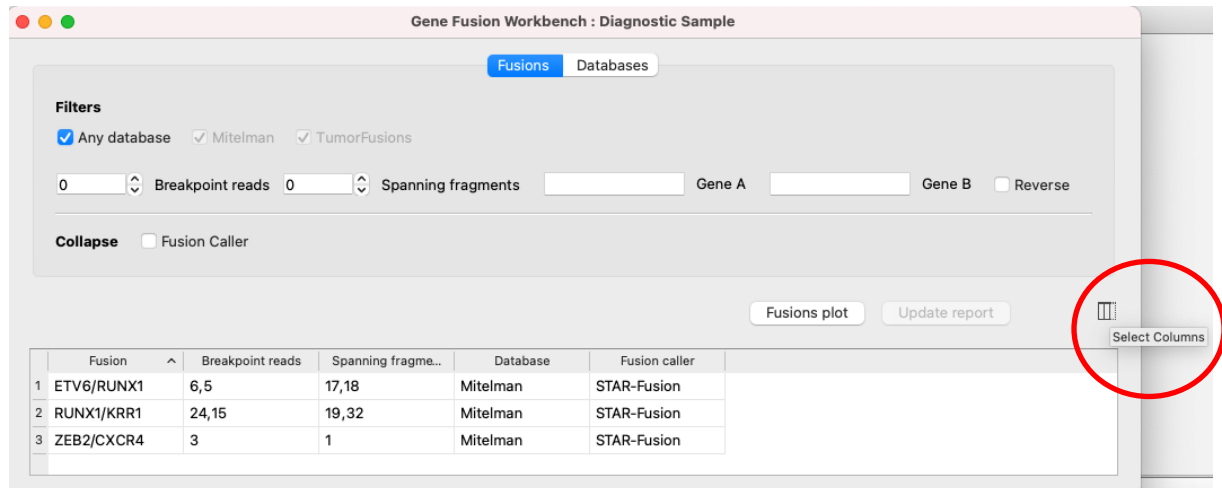


Figure 3. Gene Fusion Workbench

The Gene Fusion Workbench includes, for instance, the Mitelman database that makes it easy to compare identified gene fusions with already reported fusions. It also includes quality-oriented filters that can be applied when known and novel gene fusions are studied.

If the Gene fusion is available in the Mitelman database you can for example add the PubMed reference column and get PubMed ID(s) to study the published article(s) mentioning this fusion, see figure 4.

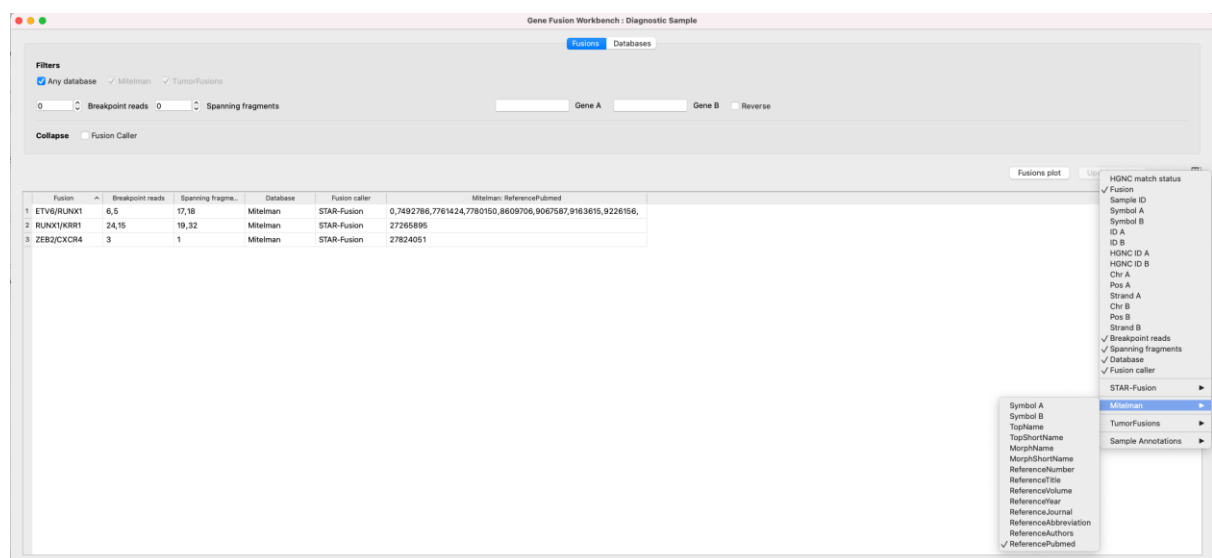


Figure 4. Include PubMed reference

## VISUALIZATIONS

The Gene Fusion Workbench is connected to two visualizations, the Genome Browser, and the Circle plot.

### Genome browser

If you double-click on a gene fusion row in the gene fusion workbench the **Genome browser** is positioned to show the gene fusion in context (Figure 5). Detailed information such as direction, exact sequence, position, and corresponding amino acids are presented here.

In the **NGS tab** you can examine the full sequence including the breakpoint. For more details see the reference manual. In the method tab you can generate a circle plot with the gene fusions.

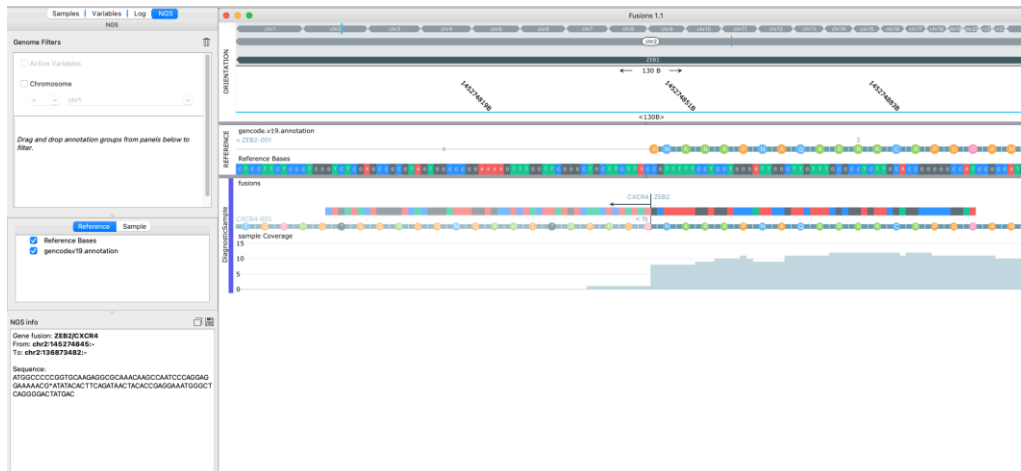


Figure 5. Genome browser

### Circular plot

To visualize gene fusions with a circle plot, select Fusion circular in Method tab (Figure 6).

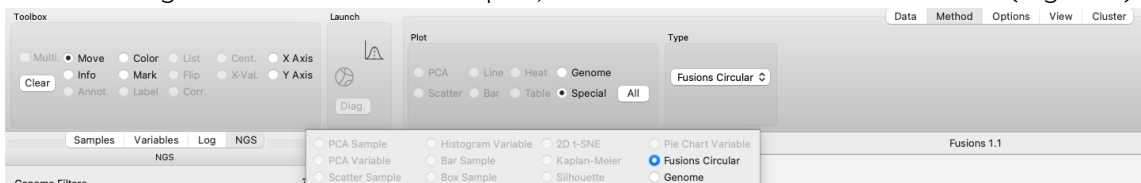


Figure 6. Launch circular plot in Method tab

The gene fusions presented in the circle plot correspond to the gene fusions that are filtered out in the Gene Fusion Workbench.

To adjust the presented fusions in the circle plot, adjust the filter in the Gene Fusion Workbench.

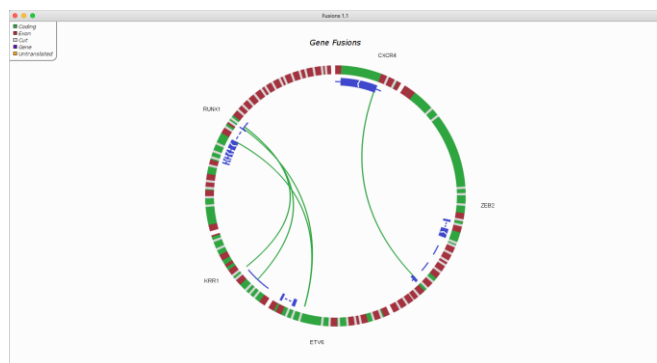
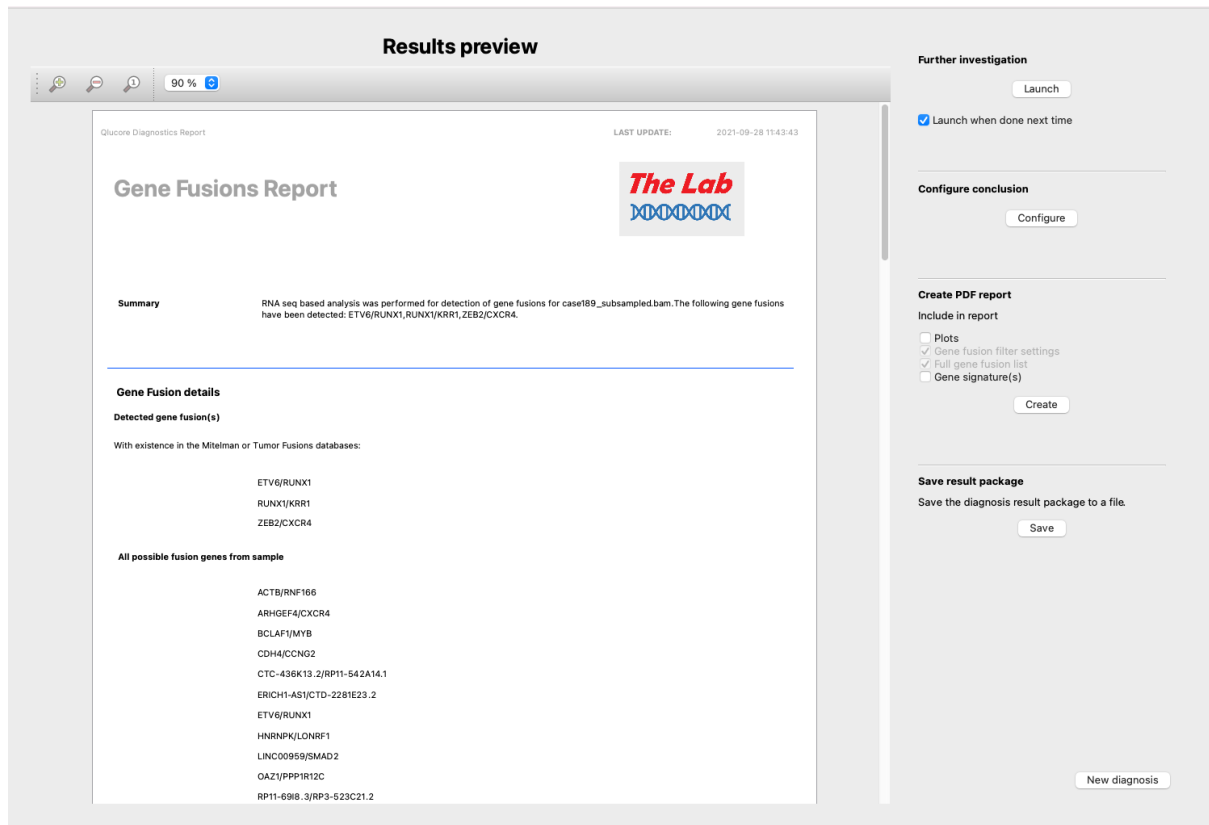


Figure 7. Circular plot

## CREATE REPORT

When the analysis is ready for report generation, go back to the **preview window** (Figure 8) to configure your report. You can choose to add the circular plot to the report and configure the conclusion.



The screenshot shows a 'Results preview' window with a 'Gene Fusions Report' on the left and configuration options on the right. The report includes a summary, detected gene fusions, and a list of all possible fusion genes from the sample.

**Gene Fusions Report**

Summary: RNA seq based analysis was performed for detection of gene fusions for case189\_subsampled.bam. The following gene fusions have been detected: ETV6/RUNX1, RUNX1/KRR1, ZEB2/CXCR4.

**Gene Fusion details**

Detected gene fusion(s)

With existence in the Mitelman or Tumor Fusions databases:

- ETV6/RUNX1
- RUNX1/KRR1
- ZEB2/CXCR4

**All possible fusion genes from sample**

- ACTB/RNF166
- ARHGGEF4/CXCR4
- BCLAF1/MYB
- CDH4/CCNG2
- CTC-436K13.2/RP11-542A14.1
- ERICH1-AS1/CTD-2281E23.2
- ETV6/RUNX1
- HNRNPJ/LONRF1
- LINC00959/SMAD2
- OAZ1/PPP1R12C
- RP11-698.3/RP3-523C21.2

**Further investigation**

Launch

Launch when done next time

**Configure conclusion**

Configure

**Create PDF report**

Include in report

- Plots
- Gene fusion filter settings
- Full gene fusion list
- Gene signature(s)

Create

**Save result package**

Save the diagnosis result package to a file.

Save

New diagnosis

Figure 8. Report generation

Save the report by clicking **Create** or save the diagnosis result as a package file to revisit without having to redo the analysis.

To run a new sample, press **New diagnosis**.

## PACKAGE

Package is a way to gather all information that has been generated together in a data package. This means that the sample processing and results are stored in the package.

The package can be opened later. Opening a package does not mean that the analysis is recalculated.

The intended use of the package is for teamwork efficiency. One analyst can do a first analysis of a sample and then forward information in the form of a package to another team member.