Savant Genome Browser: User Manual

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This document applies to Savant version 2.0.0

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Chapter 1

Introduction

1.1 What is Savant?

Savant stands for "Sequence Annotation Visualisation and Analysis Tool". In other words, Savant is a program for visualising and analysing genomics data. It was designed to run quickly and efficiently on conventional desktop or laptop computers.

1.2 Who Should Use Savant?

Savant makes visualisation and analysis of genomics data very efficient. If you use genome browsers like UCSC or IGV or if you are a biologist or bioinformatician who works with genomics data, you should try Savant.

1.3 Getting Started

Savant is designed to run on any operating system which supports Java Standard Edition 1.6 or 1.7. In practice, it has been tested on Windows (XP, Vista, and 7; 32-bit and 64-bit), Mac OSX (10.6 and 10.7), and Linux (32-bit and 64-bit). An internet connection is strongly recommended.

When you launch Savant, you will be presented with the start screen, which shows a list of recent projects and a feed of news stories from the Savant web-site. If this is the first time you have launched Savant, there will be no recent projects, so the first thing you should do is choose *File > Load Genome* to tell Savant what reference genome you wish to work with.

Chapter 2

Projects and Tracks

When working with Savant you will typically be working with a number of "tracks" which are organised together into a "project". In addition to the tracks, a project file also maintains associated information, such as the reference genome, your browsing location, and any saved bookmarks. Each track normally corresponds to single data file, although there are exceptions (e.g. a BAM alignment track may also be accompanied by a TDF file containing coverage information to be displayed at lower resolutions). In order to be loaded as a track by Savant, a file must be in one of the formats described in Table 3.1. Files which are not already in one of those formats can be converted as described in §3.2.

2.1 Projects

Projects have a file extension of *.svp*, and are typically stored in the *.savant/projects directory*. The project stores the following information:

- Reference genome
- Absolute paths of all loaded tracks.
- Current display mode of all tracks
- Bookmarks
- For variant data, a summary of which participants are controls or cases (see Chapter 11)

2.2 Genomes

Before loading any tracks, the project's reference genome must be established using the *File > Load Genome* command. This genome is used to specify the lengths and names of all chromosomes or contigs in the data

set; no other tracks can be loaded until the genome has been set. In many cases, the genome will itself be a sequence track, which can be loaded from a file, from a URL, or from a remote data-source.

For the convenience of users, a variety of standard published genomes are available from the Savant web-site. The commonly-used genomes include a full sequence track along with associated tracks such as UCSC and RefSeq genes. Less popular genomes may provide only a list of the chromosome names and their sizes. The exact list is subject to change, and we welcome requests to add your favourite genomes to Savant's standard set.

In some unusual cases, you may wish to run Savant without any existing genome information. In such cases, you can use the Load Genome dialog to specify a reference name and length.

2.3 Tracks

A track is an individual data set, usually corresponding to a single data file. In a few situations a track may draw data from multiple data sources; this is the case for BAM alignment tracks which can have an associated TDF file containing coverage information to be displayed at lower resolutions. In order to be loaded as a track by Savant, a file must be in one of the formats described in Table 3.1. Files which are not already in one of those formats can be converted as described in §3.2. If you attempt to load a track from one of the compatible file-types described in §3.1.2, Savant will offer to format it first.

2.3.1 Display Modes

Every track type has one or more display modes. Each of these modes is intended to emphasise a different aspect of the data. For instance, the *Mismatch* mode for read alignment tracks, uses colours to emphasise mismatches in reads. In contrast, the *Read Pair (Arc)* mode emphasises the insert lengths, showing arcs between the mapped locations of paired reads, with the height of each arc proportional to the inferred insert size. The display modes available for each type of track are described in §2.3.3.

2.3.2 Loading a Track

A track is choosing one of the *File > Load Track* menu-items. Track data can come from any of three different sources: a local file, a URL, or an external data source. *Load Track from File* will present a file-chooser to let you select a local file. *Load Track from URL* presents a dialog which lets you type in an ftp://, http://, or https:// URL to be opened.

The *Load Track from Other Datasource* menu-item presents the Savant repository browser, which lets you select one of the tracks which is available from savantbrowser.com. In addition, this menu-item is also used by plugins such as the SQL and UCSC plugins to allow you to open a track using that plugin.

2.3.3 Track Types

Savant supports five different types of tracks, which are described below. Each track appears in a separate window, which can be manipulated as described in §6.

At the right of the track is a legend which describes its contents. Above the legend is a track-specific menu-bar which allows you to control the display and behaviour of that track. This menu-bar will have at least two items: Tools and Appearance. In addition, some tracks have menu items which let you control *Display Mode* and *Interval Height*.

The *Tools* menu provides useful functionality such as the ability to Lock a track (see §5.1) and to copy the track's URL to the clipboard. Track types which have the ability to filter their contents add a *Filter...* item to the Tools menu, which invokes a dialog to specify filter parameters.

The Appearance menu controls the track's colour settings. For tracks which have a vertical scale (all but sequence tracks), the *Scale to Fit* menu-item controls whether the track scales its y-axis to fit the size of the window or uses a vertical scrollbar when necessary.

Sequence Tracks

Sequence tracks take their data from a .fa or .fa.savant[1] file. They display a sequence of colour-coded nucleotides.

Sequence tracks are often used by *File* > *Load Genome* to provide the reference genome for a project. If you have more than one sequence track open in a project, you can use a sequence track's *Tools* > *Set as Genome* menu-item to make it the reference genome.

Interval Tracks

Interval tracks display data from a Tabix file. This is the track-type for genes and other interval-oriented data.

Interval tracks have two display modes: Standard and Squish. In Standard mode intervals are packed neatly so that none overlap; in Squish mode, the intervals are squished together on a single line. These correspond to the Pack and Squish modes of the UCSC browser.

In addition, interval tracks also have a number of Appearance options. The *Enable ItemRGB* option colours records based on the value of their ItemRGB column (for BED files which have that column). Similarly, for BED files with a Score column, the *Enable Score* option allows you to draw records with a transparency based on the Score value. The *Display Alternate Name* option allows you to control which labels are displayed for the features (e.g. for gene tracks which may identify genes with both accession numbers and protein IDs).

[1] .fa.savant files store sequence data in a format specific to Savant versions prior to 2.0.0.

Alignment Tracks

Alignment tracks display data from a BAM file. They present reads in a choice of seven different display modes, listed in Table 2.3.3.

Standard	Displays the reads stacked vertically in the most efficient manner. Colour indicates strand direction.
Mismatch	Like Standard, but also displays SNPs and indels. Requires that the reference genome be an actual sequence track.
Read Sequence	Displays the reads stacked vertically, but colour indicates nucleotides.
Read Pair (Standard)	Reads are stacked vertically, with paired reads are at the same altitude.
Read Pair (Arc)	Read pairs are shown using arcs, where the height of the arc corresponds to the inferred insert size.
SNP	For each location, all the base-reads supporting a particular call are stacked vertically. This is intended to make SNPs more prominent.
Strand SNP	Like SNP mode, but reads from the forward and reverse strands are grouped separately.

Table 2.1: Alignment Track Display Modes

In addition to the display mode, you can also adjust the appearance of the Standard, Mismatch, and Read Sequence modes by using the *Enable Base Quality* and *Enable Mapping Quality* items from the *Appearance* menu.

When the visible range is large enough that individual reads would not be visible, Savant will display a coverage graph (§GeneratingCoverageFiles) in place of the normal display.

Continuous Tracks

Continuous tracks display continuous-valued data from TDF or BigWig files.

Variant Tracks

Variant tracks display structural variation data from Tabix-formatted VCF files. The vertical axis is used to indicate the individuals whose data makes up the file. Data from all loaded variant tracks is aggregated in the Variation sheet described in §11.

Chapter 3

Formatting and Loading Data

3.1 Supported File Formats

Savant natively supports a number of standard file formats which are indexed to ensure speedy data retrieval. In addition, Savant has the ability to reformat several other common formats in order to make them usable by the program.

3.1.1 Native File Formats

Savant natively supports the following standard file formats. These are formats which Savant can read directly with no extra processing. In most cases, these files consist of a main file which contains the actual data and an index file which is must be present in the same location as the data file in order to provide fast random access to that data.

Format	Description	Data File	Index File
BAM	Nucleotide sequence alignments	.fa	.fai
BigWig	Continuous-valued data	.bw	
FASTA	Nucleotide sequences	.fa	.fai
Tabix	Genes, intervals, variants, and other localised data	.gz	.tbi
TDF	Any continuous-valued data	.tdf	

Table 3.1: Native File Formats

3.1.2 Compatible File Formats

These are formats which Savant can convert into one of its native formats. When using a file in one of these formats, it must first be converted into one of Savants native formats. You can convert these files either by using choosing Format from Savants File menu, or by using Savants *FormatTool* utility.

Format	Description	Converted To
Bed	genes, intervals, variants, and other localised data	Tabix
BedGraph	continuous-valued data	TDF
GFF/GTF	genes and other features associated with DNA, RNA and protein sequences	Tabix
Tab-delimited	any feature-oriented data in tab-delimited form	Tabix
VCF	structural variations	Tabix
Wig	continuous-valued data such as GC percent, probability scores, and transcriptome data	TDF

Table 3.2: Compatible File Formats

Genomic annotations are also available from other databases. Downloading and formatting data from these and other popular data sources is encouraged:

UCSC	http://genome.ucsc.edu
1000 Genomes Project	http://www.1000genomes.org
NCBI	http://www.ncbi.nlm.nih.gov
EBI	http://www.ebi.ac.uk

3.2 Using the Format Dialog

The Format Dialog can be used to format text files (e.g. ones downloaded from the various data sources listed above) for use with Savant. In most cases, Savant is able to infer the file's format from its extension. Given the size of data files associated with bioinformatics, formatting a file may take a considerable amount of time.

The Format Dialog can be opened by choosing *File* > *Format File*.

Components of the Format Dialog:

Input file	The text file to be formatted.
Format	The format of the input file.
Input is 1-based	Whether or not the input file's positional annotations start at 0 or 1. In most cases, this is determined by the choice of Format.
Output file	The output file to be produced which can subsequently be loaded into Savant.

3.2.1 Generating Coverage Files

In addition to formatting the file types described in Table 3.1.2, the Format dialog is also used for generating coverage files. These are used to display alignment data when the viewable range is too large to make indi-

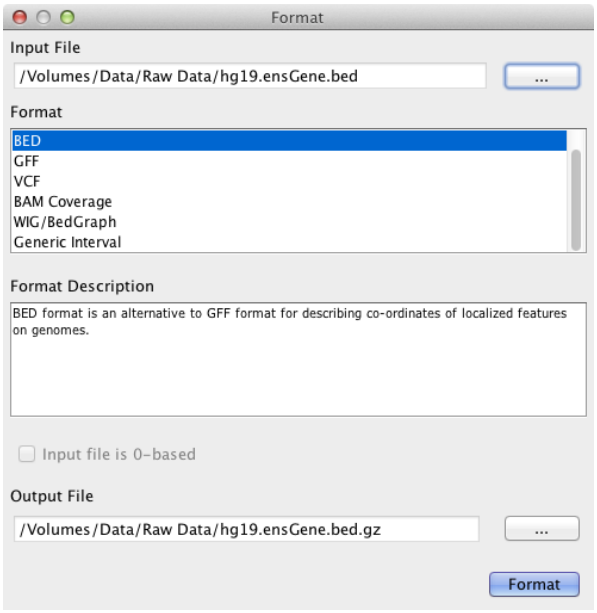

Figure 3.1: The Format Dialog

vidual reads discernible. In such cases, Savant uses the coverage file to draw a continuous track indicating the level of read coverage across the viewable range. Simply choose a .bam file as the input, and Savant will generate the corresponding .bam.cov.tdf file. In order to serve as a coverage file, the .bam.cov.tdf file must be stored in the same location as the .bam file which it summarises.

Chapter 4

Navigation

Navigation refers to changing the region of the genome which is being viewed by the browser. You can navigate either by interacting with the navigation toolbar (§4.1) located at the top of the Savant main window, or by using the appropriate keyboard and mouse shortcuts (§4.2).

In Savant a location consists of two parts: the current reference and the current visible range. The current reference is typically a chromosome, so the term "chromosome" is used below, even though the reference in question could actually be any contiguous range of bases, and not necessarily a chromosome *per se.* The current range specifies a range of bases in the coordinate space of the current reference.

4.1 Navigation Toolbar

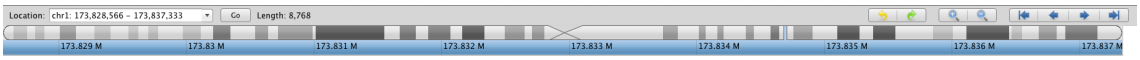

Figure 4.1: Navigation Toolbar

The top row of the navigation toolbar contains the location field (§4.1.1) and the navigation buttons (§4.1.2). Below that are the genome bar and the ruler (§4.1.3).

4.1.1 Location Field

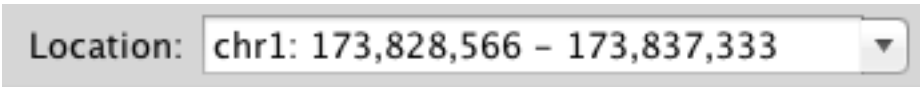

Figure 4.2: Location Field

The location field displays the current visible range within the genome. It can also be used to specify a new range in a number of ways, described in the table below. Changes take effect by hitting RETURN or clicking the *Go* button.

1)	Clicking the downward-pointing triangle to reveal a menu of chromosomes.	
2)	Type a gene name into the text field. Type a partial gene name and hit TAB to pop up a menu of matching genes.	
3)	Type a range specification into the text field:	
	chr2:1000-2000	chr2, range 1000-2000
	chr2	chr2, range 1-1000
	1000-2000	in current chromosome, range 1000-2000
	1000-900	in current chromosome, range 900-1000 (equivalent to 900-1000)
	1000+2000	in current chromosome, range 1000-3000
	1000	in current chromosome, start position at 1000, keeping current range-length
	+1000	1000 bases to the right of the current start, keeping same range-length
	-1000	1000 bases to the left of the current range, keeping same range-length

Table 4.1: Specifying Ranges Using the Location Field

4.1.2 Navigation Buttons

Figure 4.3: Navigation Buttons

At the top right of the Navigation Toolbar are eight buttons which perform a variety of navigation-related tasks. From left to right these are: *Undo, Redo, Zoom In, Zoom Out, Beginning, Pan Left, Pan Right,* and *End.*

Undo lets you undo your previous navigation action. It can be used repeatedly to undo multiple navigation actions. *Redo* cancels the effect of the most recent *Undo.*

Zoom In and *Zoom Out* change the visible range by a factor of two, keeping it centred on the same location.

Pan Left and *Pan Right* shift the visible location left or right by half the width of the screen.

Beginning and *End* allow you to navigate quickly to the beginning or end of the current chromosome.

Figure 4.4: Genome Bar and Ruler

4.1.3 Genome Bar and Ruler

Below the location field and the navigation buttons lies the genome bar. This bar highlights in blue the current visible region in the context of the genome. If the current genome has associated cytoband information, it will be displayed in this bar. You can select a new range by clicking and dragging within the bar.

Underneath the genome bar is the ruler, which indicates the current viewable range in the coordinate space of the current chromosome.

4.2 Mouse and Keyboard Shortcuts

Navigation can be done very quickly using a number of mouse and keyboard shortcuts described in the tables below.

Keys	Action
SHIFT+LEFT	pan left
SHIFT+RIGHT	pan right
SHIFT+HOME	move to start of chromosome
SHIFT+END	move to end of chromosome
SHIFT+UP	zoom in
SHIFT+DOWN	zoom out

Table 4.2: Navigation Using the Keyboard

Keys	Action
Click and drag left/right	pan left/right
CTRL/CMD + scroll-wheel up/down	pan left/right
CTRL/CMD + Click and drag	zoom in on selected region
SHIFT + Click and drag	select all records in region

Table 4.3: Navigation Using the Mouse

4.3 Other Useful Shortcuts

Range changes can be undone and redone using the commands in the following table.

Keys	Action
CTRL/CMD + Z	undo range change
CTRL/CMD + Y	redo range change
CTRL/CMD + I	export track images (see §10.1)
CTRL/CMD + B	bookmark current location (see §7)
CTRL/CMD + J	crosshair (see §5.4)
CTRL/CMD + K	plumbline (see §5.4)
CTRL/CMD + L	spotlight (see §5.4)

Table 4.4: Other Shortcuts

Chapter 5

Visualisation Features

In addition to making it easy to navigate quickly and efficiently through the genome, Savant provides a variety of mechanisms for making it easier to visualise the data more effectively.

5.1 Track Locking

Individual tracks can also be locked to a particular range so that they are not updated until they are unlocked. Locked tracks can be used as overview profiles from which subregions can be selected to specify range changes for other tracks. To lock a track, select the *Lock Track* option from the track's *Tools* menu. While a track is locked, you may select a subrange from the track (by using the mouse zoom options, described previously) which will become the new range for other, unlocked tracks. To unlock a track, go to the track's menu and uncheck the *Lock Track* option.

5.2 Selecting Records

In most cases, a track consists of a collection of underlying records, which can be selected individually. By default, selected records are highlighted in green. Selected records remain selected when the display mode changes, so for instance you could select an arc in Read Pair (Arc) mode and then switch to Mismatch mode to see the details of the selected reads. Records selected in a track are also selected in the Data Table (Chapter 8).

5.3 Popup Menus

Hovering the mouse over a track will pop up a menu which provides information about the record under the mouse, as shown in Figure 5.1. The details of what fields are displayed in the popup is dependent on the

type of data contained in the track. To make it clearer exactly which record corresponds to the popup, the track will give the record a reddish tint while the popup is open.

At the bottom of the popup menu are a number of blue menu-items which allow you to perform specific actions on the current record. The *Select/Deselect* and *Add to Bookmarks* items are common to popups for all track types; certain track types may add additional items as appropriate.

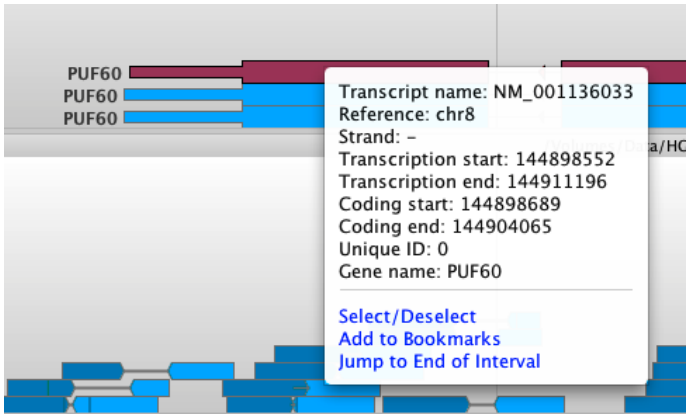

Figure 5.1: Track Popup

5.4 Highlighting Areas of Interest

Often, it is helpful to focus one's attention on a particular portion of the display. Savant provides three tools for making this easier, found under the *View* menu.

The *Crosshair* tool displays a crosshair cursor and shows the current track-space coordinates of the mouse. This is intended to make it easier to identify particular features by location. Note that the current location (without the crosshair) is also shown in the lower left of the Savant window.

The *Plumbline* tool displays vertical bars which run across all the tracks, allowing you to clearly see how features on different tracks are aligned.

The *Spotlight* tool is similar to the Plumbline tool, except that portions of tracks outside the spotlight area are dimmed to deemphasise them.

Chapter 6

Docking Framework

Savant features a docking framework which allows you to rearrange modules to their liking. Such modules include tracks and built-in items (e.g. Bookmarks, Variants, etc.) and plugins. Non-track modules are constrained to be docked to the sides of the UI and not among tracks. Similarly, track modules are constrained so that they cannot be docked among other modules.

While a number of important functions are presented here, the best way to learn all the features of the docking framework is to try using it.

6.1 Showing and Hiding Modules

By default, built-in modules are hidden. Hidden modules appear as tabs located on the region of the UI to which they are docked. A module is shown once the tab is clicked. Click the tab again to hide the module.

6.2 Resizing Modules

Modules can be resized. To resize a module, click an edge and drag it until it occupies the desired size.

6.3 Rearranging Modules

Modules can be arranged in virtually any configuration within the UI. To move a module, click its title bar and drag it to the desired new location. While dragging, a grey outline will appear showing the location the module will occupy if the mouse is released. Track modules can be docked to the top or bottom of the track space, while other modules can be docked to any edge of the UI. In addition, modules can be docked on top of each other (in which case tabs will appear allowing one to switch between modules) or beside each other.

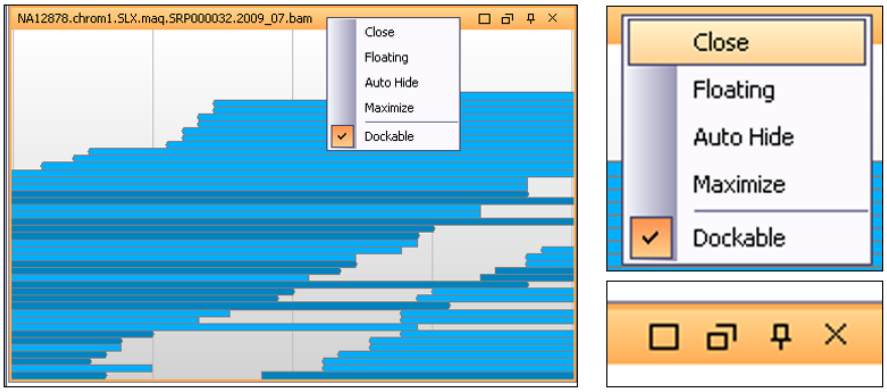

Figure 6.1: Left: A track module. Upper-right: Docking menu presented when the title bar of a module is right-clicked. Bottom-right: Docking controls embedded in the title bar of the module. The latter controls are *not* available in the Mac version.

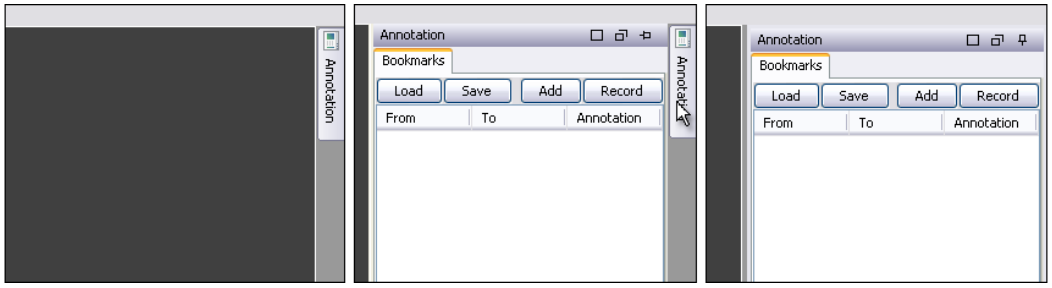

Figure 6.2: Hiding and Showing Modules. Left: The Annotation module, hidden on the right of the Savant UI. Middle: The Annotation module, shown by mousing over the Annotation tab. Right: The Annotation module, pinned so that it will remain shown even when you are interacting with other components of the UI.

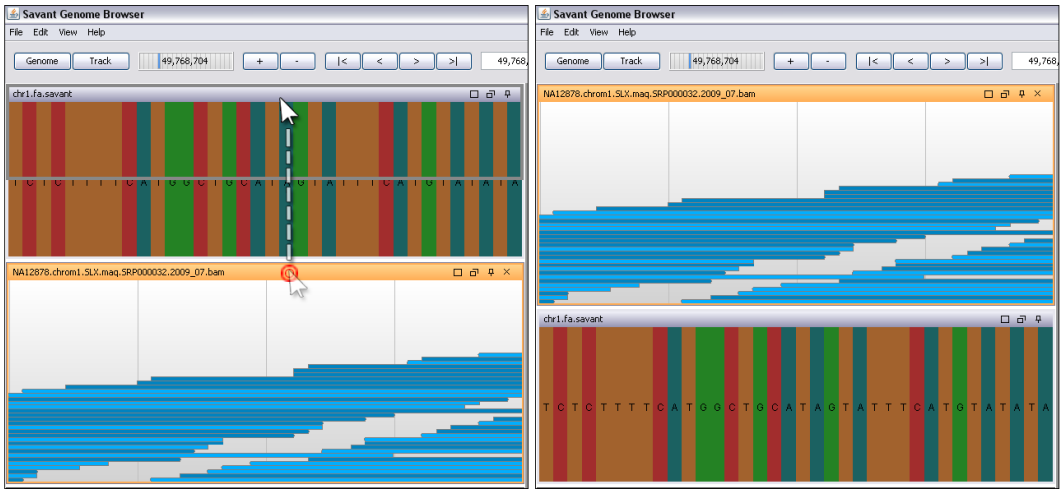

Figure 6.3: Rearranging Modules. Left: Demonstration of the rearrangement process. Right: The result of the rearrangement.

6.4 Maximising and Restoring Modules

A module can be maximised to occupy the entire screen or UI, to make interaction or visualisation with it easier, and then restored back to its original state among other modules to resume a concerted view. To maximise, press the embedded icon which resembles a square. To restore, press the embedded icon which resembles two overlapping squares (in the same location as the icon pressed to maximise). On a Mac, you can maximise and restore by double-clicking the title bar. The same functionality is possible through the title bar on Windows and Linux.

6.5 Detaching and Attaching from and to the UI

A module can be detached from the UI and moved to a separate location on the screen. This is particularly useful for multi-display setups where, for example, analytics modules can be moved to one display and tracks kept on another. To detach a module, press the embedded icon which resembles to squares. The detached module can then be moved to another location by clicking and dragging its title bar. To reattach it to the UI, press the embedded icon which resembles a square with an L-shape in it (in the same location as the icon pressed to detach it). On a Mac, you can detach and reattach by right-clicking the title bar and checking or unchecking Floating, respectively. The same functionality is possible through the title bar on Windows and Linux.

Chapter 7

Bookmarks

The Bookmarks module helps to keep track of interesting regions or to make annotations. At any time, you may add, remove, or seek to a bookmarked region by using buttons within the module or by using keyboard shortcuts.

7.1 Adding and Removing Bookmarks

A bookmark can be added by pressing the Add button ( ) in the Bookmarks module's toolbar. The current range will be used for the bookmark, although the from and to coordinates of the bookmark can be adjusted by double clicking and changing them. The keyboard shortcut CTRL+B (or CMD+B on a Mac) can be used to quickly add a bookmark.

For tracks which contain multiple features (e.g. gene tracks), the track's *Tools* menu provides a way to automatically create a bookmark for each of a track's features.

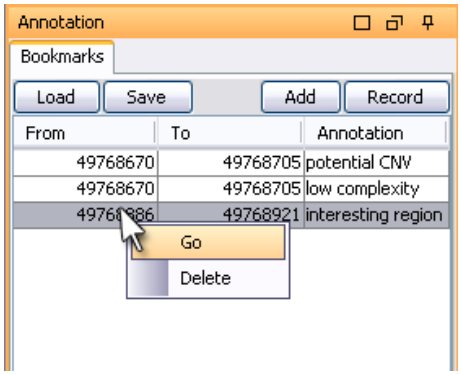

Figure 7.1: Bookmarks Module

Bookmarks can be removed by selecting them and choosing the Delete button ().

7.2 Seeking to a Bookmark

A bookmark can be sought to by selecting it and choosing the Go option.

7.3 Adding Annotations to Bookmarks

An annotation can be added to a bookmark by double-clicking the Annotation field and typing in some text.

7.4 Saving and Loading Bookmarks

As bookmarks are created, they are automatically stored as part of the current project file, and will be saved whenever the project is saved.

Bookmarks can be exported, to be reused in other sessions or to be shared with colleagues. To save the existing bookmarks, click the Save button () in the Bookmarks module's toolbar. These bookmarks can subsequently be loaded by clicking the Load button ().

Exported bookmarks are stored as a simple tab-delimited text file with four columns: chromosome, start, end, and annotation. Consequently, if you create your own tab-delimited file with this layout, it can be loaded into a Savant project as a set of bookmarks.

You can opt to append the loaded bookmarks to the existing bookmarks, or to replace the existing bookmarks entirely with the loaded ones.

Savant also gives you the option of adding padding to bookmarks to make them more prominent. For instance, if you have loaded bookmarks from a text file containing SNP coordinates, you might want to add padding so that navigating to a SNP will display it centred in the display area; without padding, selecting one of these SNP bookmarks would set the range so that the selected SNP filled the entire display.

Chapter 8

Data Table

The Data Table is a plugin (see Chapter 9) which is installed by default as part of Savant. The Data Table module displays the data in the current range in a tabular format, with the records being rows and the fields being columns.

The data is automatically updated when the range in the main track display changes, unless the *Auto Update* checkbox is unchecked. The *Show Only Selected* checkbox allows you to filter the table, showing only records which are selected in the current track.

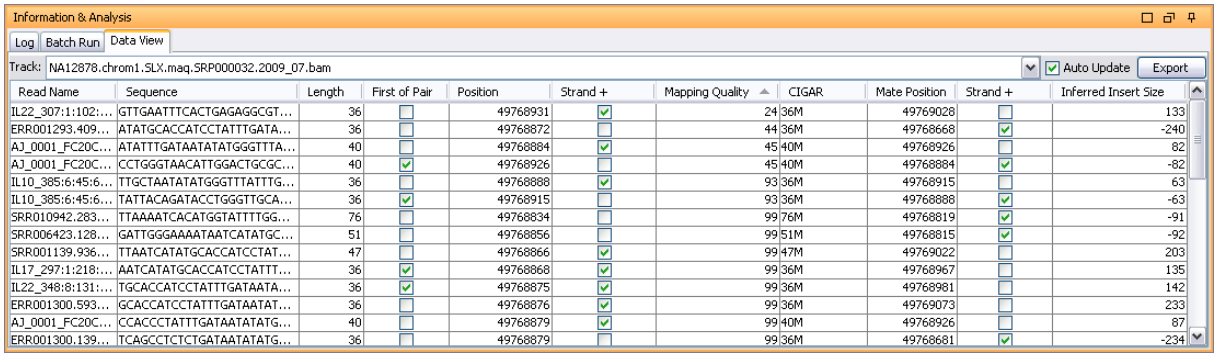

Figure 8.1: Data Table Module, showing data from a read alignment track.

8.1 Changing Tracks

The Data Table only displays data from a single track at a time. To change the track whose data is being displayed, a drop-down list of tracks is provided from which to choose.

8.2 Sorting Rows

The entries in the Data Table can be sorted by clicking the field header by which the rows are to be sorted. Subsequent clicks on the field header toggle between sorting in ascending order and descending order.

8.3 Exporting Data

The data being viewed in the Data Table can be exported to a text file by clicking the Export button.

The format of the resulting text file depends on the type of the track being exported. Sequence tracks are exported as Fasta files. BAM alignment tracks are exported as SAM files. All other track types are simply exported as tab-delimited text.

Chapter 9

Plugins

Savant is able to integrate plugins, allowing for powerful extensions of the browser. To learn how to develop a plugin, see the **Developer's Guide** downloadable from the Savant web-site.

9.1 Installing and Uninstalling Plugins

To install a plugin, choose *Plugin Manager...* from the *Plugins* menu. Savant will present the dialog shown in Figure 9.1. This dialog lists all the plugins which are currently installed. Selecting one of plugins from the list will display information about the plugin's version and status, and provide a button to allow uninstalling the plugin.

Clicking *Install from Repository* will bring up a second dialog (Figure 9.2) which lets you browse the plugins available on the Savant web-site. Plugins in the repository are segregated by Savant version, since Savant 2.0.0 is not compatible with plugins developed for older versions of Savant.

Third-party plugins which are not available through the Savant Plugin Repository can be installed using the *Install from File* command, which lets you select a JAR file to be installed. *Install from File* will copy the selected file into Savant's plugin directory and make it available to Savant.

Some third-party plugins may require ancillary files (e.g. supporting libraries or data files). Installing this sort of plugin cannot be done through the Plugin Manager dialog; in such cases the necessary files must be manually copied into the *.savant/plugins* directory (*savant\plugins* on Windows).

9.2 Using Plugins

Every plugin works differently and may or may not have a user interface. For instructions on how to use a third-party plugin, see the developer's documentation. Savant supports three general types of plugins, which are described below.

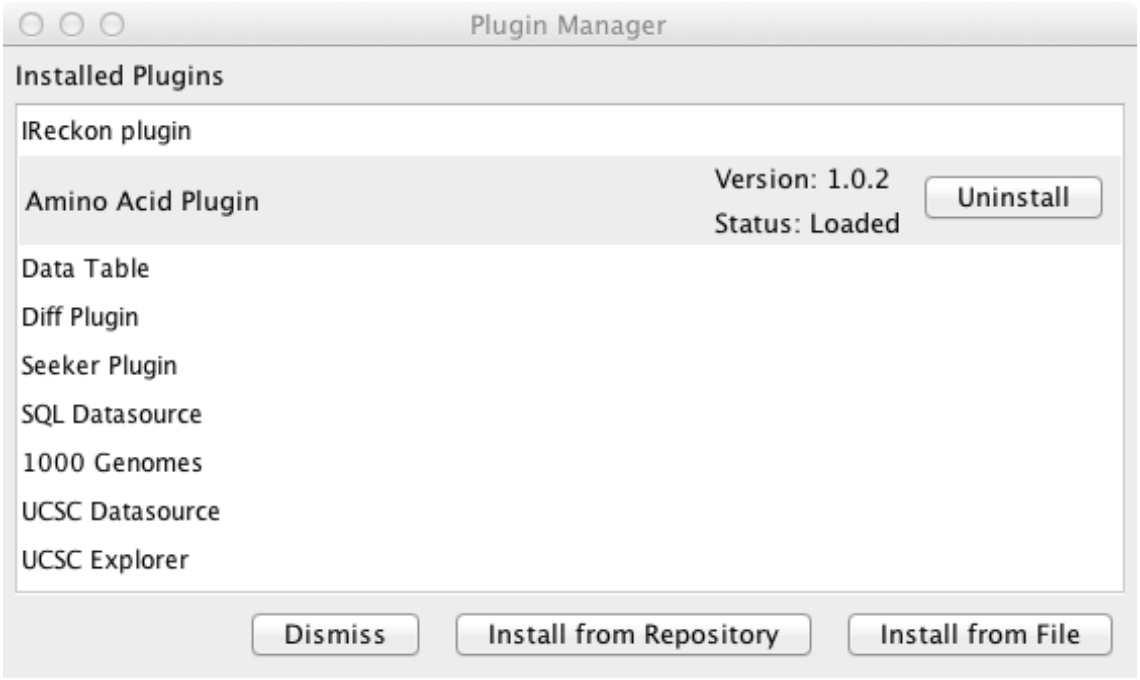

Figure 9.1: Plugin Manager Dialog

9.2.1 Panel Plugins

Typical plugins (such as the built-in Data Table and UCSC Explorer plugins) are presented in tabs at the bottom left of the Savant user interface. Click the tab to reveal the plugin's user interface. The windows for these plugins can be detached, docked and rearranged, as described in Chapter 6.

Plugins can be temporarily disabled by unchecking the corresponding menu-item on the *Plugins* menu. This is a less extreme measure than uninstalling the plugin; a disabled plugin is still installed and can easily be reenabled by reselecting its menu-item from the *Plugins* menu.

9.2.2 Tool Plugins

New in Savant 2.0 is a new variety of plugin, referred to as "tool plugins". To the end-user, these are functionally similar to panel plugins, but where panel plugins consist of Java code packaged inside a JAR file, tool plugins consist of an XML file which provides instructions on how to invoke an external application. The format of this XML file is described in the Developer Manual.

Like panel plugins, tool plugins present their user interface in a dockable tab at the bottom left of the Savant user interface, and can be enabled/disabled from the *Plugins* menu.

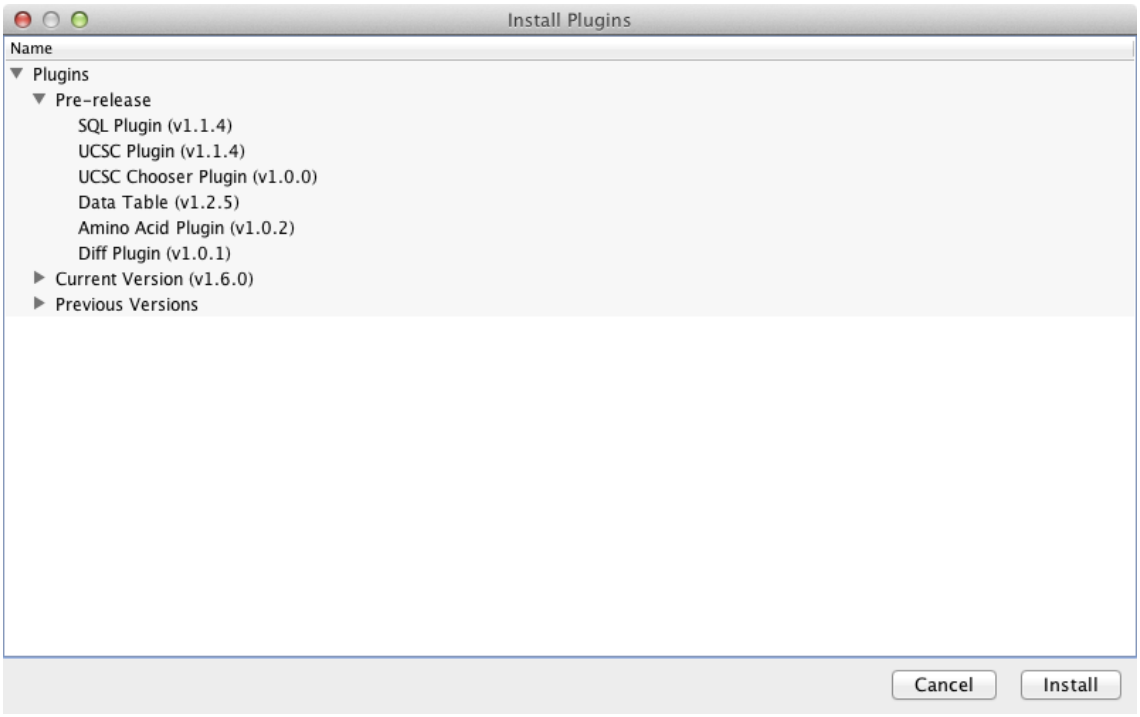

Figure 9.2: Plugin Repository Dialog

9.2.3 Data Source Plugins

Data source plugins are a special class of plugins which extend Savant, allowing it to access data in non-standard ways. Data source plugins do not have a tab at the bottom left of the Savant window, but instead are accessed through the *Load Track from Other Datasource* menu-item.[1]

The UCSC and SQL plugins are both examples of data source plugins.

[1] Or in the case of genomes, from the *Other Datasource* button on the genome dialog.

Chapter 10

Other Features

10.1 Exporting Images

Chapter 11

Variants

Savant 2 provides improved support for viewing variant data in VCF files. In addition to being able to open Tabix-formatted VCF files as variant tracks, Savant 2 also provides a Variation tab at the right of the user interface (next to the Bookmarks tab). This tab provides a variety of ways of exploring variation data, as described in §11.2.

11.1 Variant Tracks

Variant data can be loaded into Savant using the *Load Track from XXX...* commands. Loading a VCF file in this fashion will create a new track in the main display area. As with other data types, the VCF file must first be formatted and indexed as a Tabix file in order to be usable (see §3.2).

SNPs are displayed with a vertical line in the colour of the alternate (non-reference) base. Participants drawn from the VCF file are arranged vertically. A gap in the vertical line indicates that a particular participant lacks that variant. If a variant is heterozygous for a participant, that portion of the line will be drawn using a paler version of the associated colour.

Insertions and deletions are displayed in the same manner as SNPs, with a single vertical line (black for deletions, magenta for insertions). The variant track makes no attempt to visually indicate the extent of the inserted or deleted material. In order to see what was actually substituted by the indel, you will need to mouse over the variant track, and the popup will provide the details of the substitution that took place.

11.2 Variation Module

The Variation module groups together four different ways of viewing aggregated variant data. While variant tracks contain data only from a single VCF file, the visualisations on the Variation module comprise data from all currently-open variant tracks.

The variation module maintains its own visible range, which is distinct from the visible range for the main track display area. Changing the range in the variation module has no effect on the main range. The reverse is not true, since if you move the main display range outside the variation module's range (e.g. by shifting to another chromosome), the variation module will update its range to "follow" your navigation.

At the top of the variation module is a field for entering the module's desired visible range. The variation module also has its own Zoom In and Zoom Out buttons to alter its visible range.

At the top right of the variation module is the *Controls* button. This is used to select which participants in a dataset are considered to be controls (as opposed to cases). When graphing allele frequency, the control and case participants are graphed separately.

11.2.1 Variation Table

The variation table provides a spreadsheet-like summary of all variants in the variation module's current display range. Double clicking on row will centre the main track display on the position of that variant.

11.2.2 Variation Map

The variation map provides a graphical overview of all variants in the variation module's display range. Variant records are arranged vertically by position, but the scale is non-linear since the gaps between variants are squeezed down. Participants are laid out horizontally, using the same colour scheme as described for variant tracks (§11.1)

11.2.3 Allele Frequency Plot

The allele frequency plot graphs frequencies of each allele for each variant location. Like the variation map, the allele frequency display is arranged vertically by position, with a non-linear scale.

If controls have been specified, allele frequencies for controls will be displayed on the left of the central axis, while case allele frequencies will be displayed to the right of the central axis.

11.2.4 Linkage Disequilibrium Plot

The linkage disequilibrium plot graphs LD values calculated using D' or r^2. Variant locations are used as the loci for the LD calculation (i.e. there is no attempt to segregate ranges into groups).

Chapter 12

Preferences

Appendix A

Savant Home Directory

By default, Savant stores settings, plugins, and temporary files in your home directory in a folder called *.savant* on Mac and Linux, or *savant* on Windows. In most cases, it should not be necessary to delve into this directory, but it can be useful to know what is stored there. Table A.1 summarises the contents of the *.savant* directory.

cache	Keeps a cache of files accessed from remote URLs. Over time, this directory may use up a considerable amount of disk space. It can be cleared using the *Clear remote file cache* option in the *Preferences* dialog (Chapter 12).
index	Stores index files for remote BAM, Tabix, and Fasta files.
plugins	The Plugin Manager dialog (§9.1) installs plugins in this directory.
projects	Default location for Savant project (*.svp*) files.
savant.settings	Stores values for Savant preferences. Edit this file at your own risk.

Table A.1: Contents of *.savant* Directory