



Affymetrix[®] Genotyping Console Browser 1.2 User Manual

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Chapter 1: Introduction

Genotyping Console (GTC) Browser was designed to support copy number and Loss of Heterozygosity (LOH) analysis. The visualization of chromosomal aberrations and LOH is facilitated by the analysis algorithms within GTC 4.1 which provide graphical results in both a full genome-wide karyoview and individual chromosome views. Below are specific details on the two graphical views within GTC Browser 1.2.

- *Karyoview*: In this view the entire genome is represented visually with chromosomal ideograms arranged vertically. Within this view cytobands on each ideogram are represented and annotated. Gains and losses are represented by blue and red triangles respectively.
- *Chromosome View*: In this view an individually selected chromosome is displayed along with the accompanying data graphs and annotation tracks:
 - Segment Report tracks which identify relevant copy number aberrations Log2 ratios for all markers of sample array relative to reference.
 - LOH graphical view of homozygous and heterozygous regions.
 - HMM Copy Number State (cnstate) representation.
 - Additional Copy Number tracks are available depending on platform and type of analysis performed
 - Ability to link externally to public databases (UCSD, Toronto DGV, and Ensembl).
 - FISH data
 - RefSeq genes.
 - Custom Tracks which allows users to upload regions of interest to display.

The GTC Browser is installed during the installation of GTC; see the GTC help and manual for information on installation and setup.

The following sections provide information:

- *About this Manual*
- *Technical Support*.

About this Manual

This manual presents information about GTC Browser in the following chapters:

- *Chapter 2: Browser Introduction* (page 6): Describes how to open the Browser and introduces:
 - *Karyoview* (page 16)
 - *Chromosome view* (page 23)
- *Chapter 3: Annotations* (page 31): Describes how the annotations are displayed in the Chromosome view and how to modify the display of annotations.
- *Chapter 4: Graphs* (page 45): Describes how the data is displayed in the Chromosome view and how to modify the display of data.
- *Chapter 5: Exporting Data* (page 55): Describes how to export images and data.

 **See the *GTC 4.1 User Manual* for more information about Copy Number and Loss of Heterozygosity analysis.**

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Chapter 2: Browser Introduction

The GTC Browser 1.2 displays data generated by the following analysis types:

- Copy Number
- Loss of Heterozygosity (LOH)
- Copy Number Segment

The Browser also displays annotations to help you interpret your results.

Key features include:

- Whole genome and chromosome specific views
- Multi-sample view to facilitate identification of trends in copy number or LOH data
- Dynamic links to external websites (UCSC, TorontoDGV, ENSEMBL)

The browser has two different display modes:

- *Karyoview* (page 16): see Figure 1.
- *Chromosome view* (page 23): see Figure 2.

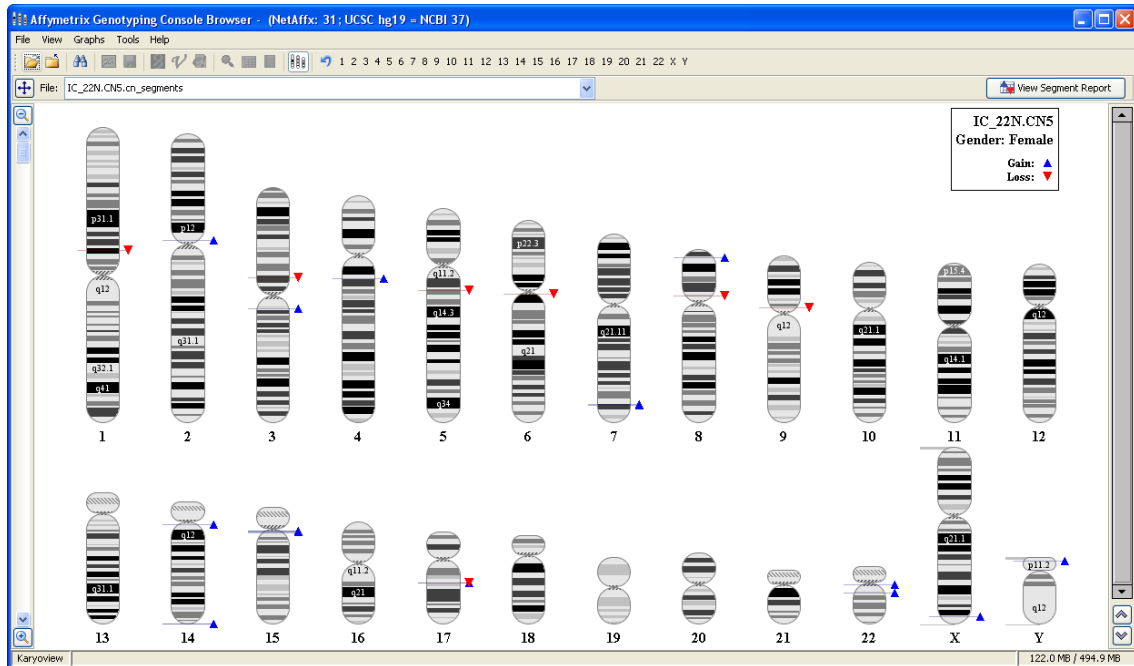


Figure 1: Browser in Karyoview



Figure 2. Browser in Chromosome view

This section covers the following material:

- *Basic workflow with Browser (below)*
- *How to load data into the GTC Browser (page 8)*
- *Browser overview (page 15)*
- *Karyoview (page 16)*
- *Chromosome View (page 23)*

Basic Workflow with Browser

The GTC Browser is used as part of the GTC Copy Number workflow. The Browser allows you to look at copy number and LOH data on a genome-wide scale as part of the following workflow (Figure 3):

1. Generate Copy Number and LOH files in GTC.
2. Run Segment Reporting Tool to generate Copy Number Segments files.
3. Review data in the GTC Browser.
4. Export data for further analysis or view the chromosomal region in a public genome browser.

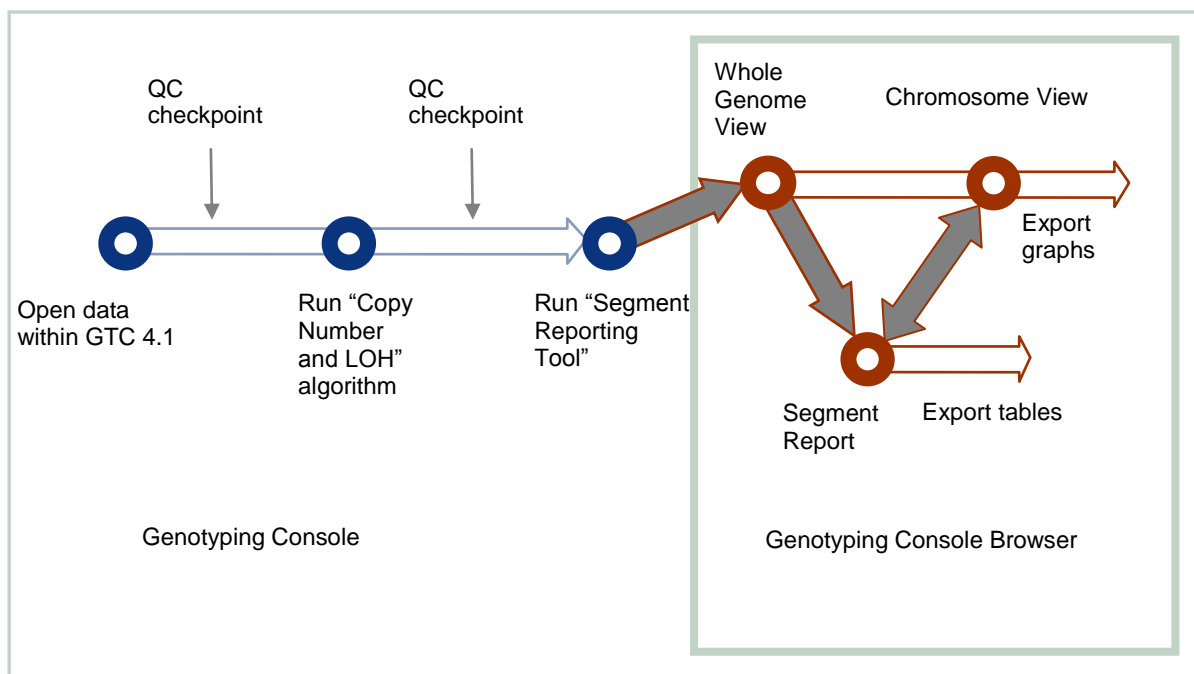


Figure 3. Basic Workflow for generating and viewing Copy Number/LOH data

Selecting the NetAffx Genome Annotation DB File

In GTC Browser, genomic assembly version information is contained inside the NetAffx Genomic Annotation Database files.

! Users must download the NetAffx Browser Genome Annotation files from Affymetrix.com product pages or support pages. Download the NetAffx Browser Genome Annotation file that corresponds to the NetAffx annotation version that was used to analyze the data. After the files are downloaded, put them in a local library folder (for example, the current GTC library folder).

Before you can load any data files, you must first load a NetAffx Browser Genome Annotation file. These database (DB) files are created by NetAffx and contain information about a particular genomic assembly version and annotations (RefSeq, Genomic Variants, FISH Clones) from public sources.

Selecting a Browser Annotation Database File

If this is the first use of the browser, you will be prompted to download a DB file (Figure 4). After you download the DB file, load it in the browser. Each time you start the browser, it will attempt to load the database (DB) file you were using in your last session. If the previously used DB file is no longer available on your disk, the browser will prompt you to select a DB file.

Changing to a different DB file will cause the browser to first unload all currently-loaded data files. For more details on selecting a different DB file, see page 10.

1. Click **OK** in the notice that appears (Figure 4).

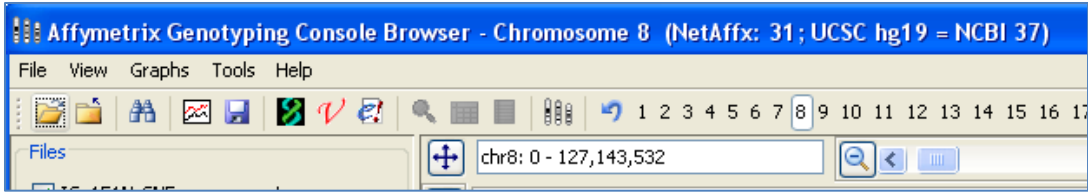


Figure 6. Title bar with annotation information

Changing the Browser Annotation Database File

Changing the annotation DB file used will automatically change the genome assembly version based on the version contained in the file.

The program will not allow you to load data files generated with a genome assembly version which does not match the genome assembly version of the NetAffx annotation database file. The currently loaded annotation version is displayed in the title bar of the browser (Figure 6).

To change the annotation DB file:

1. From the File menu, select **Load NetAffx Browser Annotation Database File** (Figure 7).

A notice appears warning that the currently loaded data files will be unloaded (Figure 8).

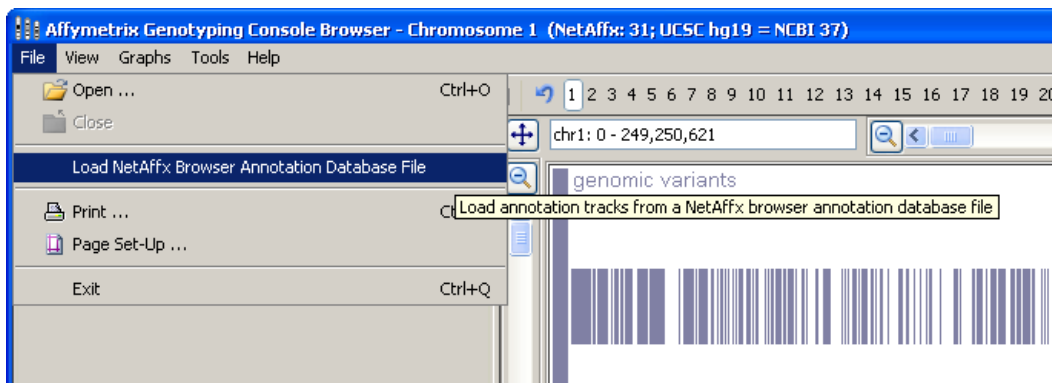


Figure 7. Menu commands to load a DB file

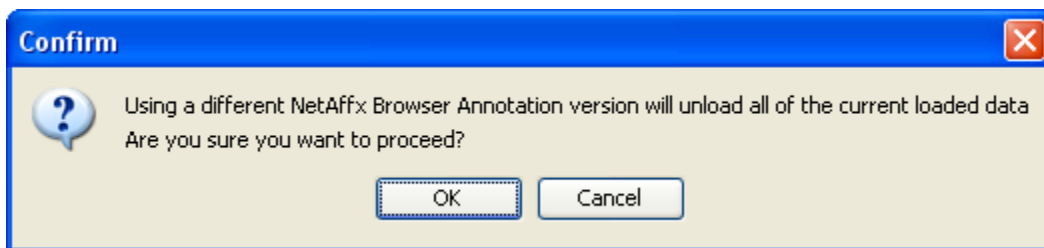


Figure 8. Notice

2. Click **OK**.

The Choose a NetAffx Annotation DB File dialog box opens (Figure 9).

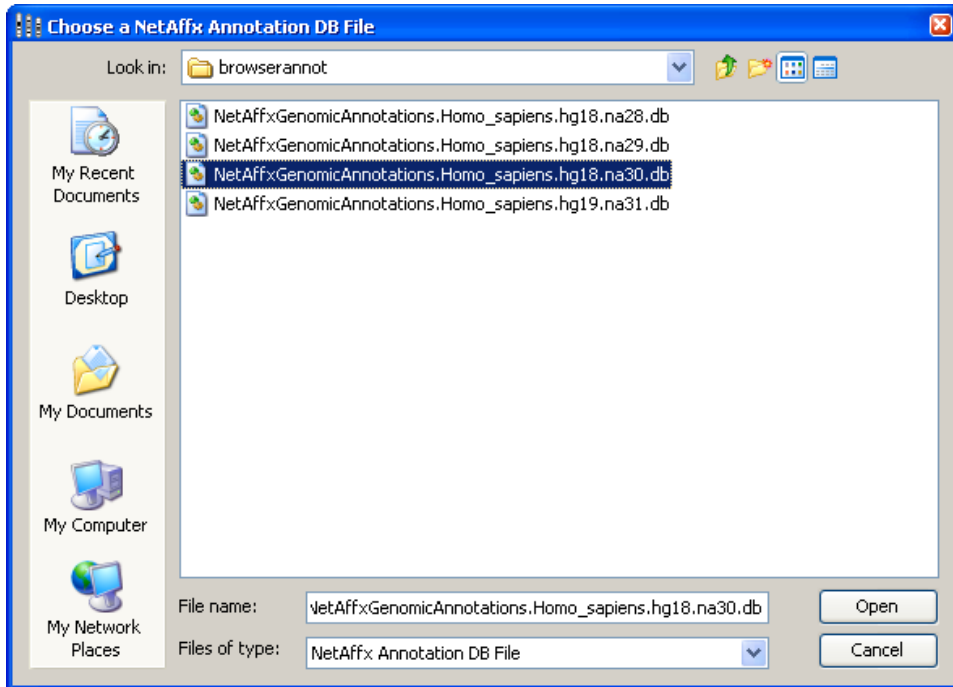


Figure 9. Choose a NetAffx Annotation DB File dialog box

3. Navigate to the directory where your database files are located and select one of the database files.

The file name uses the format:

NetAffxGenomicAnnotations.Homo_sapiens.<genome assembly number>.<NetAffx build number>.db

Examples include:

- NetAffxGenomicAnnotations.Homo_sapiens.hg18.na30.db
- NetAffxGenomicAnnotations.Homo_sapiens.hg19.na31.db

NOTE: The program will not allow you to select a file which does not use this naming convention.

NOTE: Do not confuse these genome-based annotation database files with array-specific annotation files. The array-specific files have names which begin with the name of an Affymetrix array and end with “.annot.db”. Example: “GenomeWideSNP_6.na24.annot.db”.

4. Click **Open**.

The selected database file is loaded

After a database file is loaded, the application title bar will display both the NetAffx build number and the genome assembly version (Figure 10).



Figure 10. Title bar with annotation information

NOTE: You will only be able to load data files created with the same genome assembly version as the version shown in the title bar. This is because different genome assembly versions use different coordinates to refer to the same genomic locations in the reference genome assembly.

NOTE: You will be allowed to load data files created with a different NetAffx build version from the one shown in the title bar, as long as the genome assembly version matches. A warning will be displayed, but you will be allowed to continue.

How to Load Data into the GTC Browser

Upon running Segment Reporting Tool, you are given the option to open the new files in the Browser

See the *Genotyping Console 4.1 User Manual* for information on:

- Performing Genotyping analysis
- Performing Copy Number/LOH analysis
- Selecting data in the data tree

To display copy number data in the GTC browser:

1. In the Genotyping Console data tree, select the copy number data you wish to display (Figure 11).

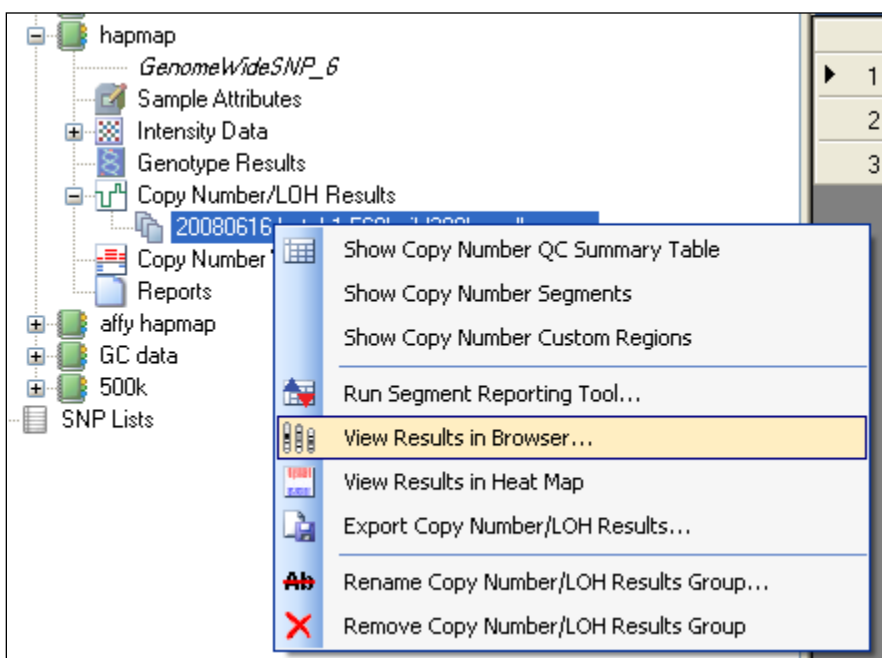


Figure 11. Selecting results in the GTC

2. Right-click on the data and select **View Results in Browser** from the context-sensitive menu; or

From the Workspace menu, select **Copy Number/LOH Results > View Results in Browser**; or

In the toolbar, click the **View Results in Browser** button.

The Select Copy Number Results dialog box opens (Figure 12).

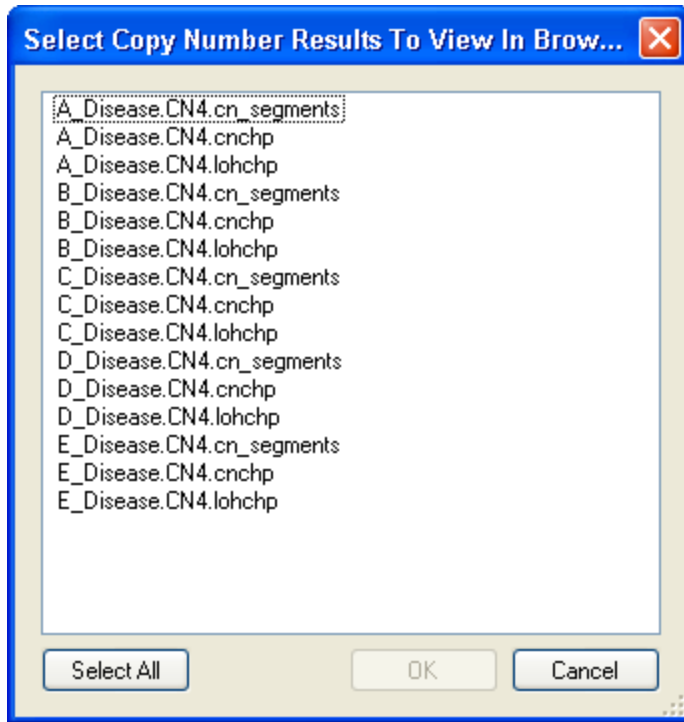


Figure 12. Select Copy Number Results dialog box

The dialog box displays a list of the Copy Number, LOH, and Copy Number Segments files available in the Results set.


 **Note: Not all the file types may be available depending upon the type of array used.**

3. Select the files you wish to view; or
Click Select All.
4. Click **OK**.

The GTC browser opens and displays the data, along with the default annotation files.

Loading Additional Data

To load additional data:

1. From the File menu, select **Open**; or
Click the **Open** button  in the Browser toolbar.

The Open dialog box appears (Figure 13).

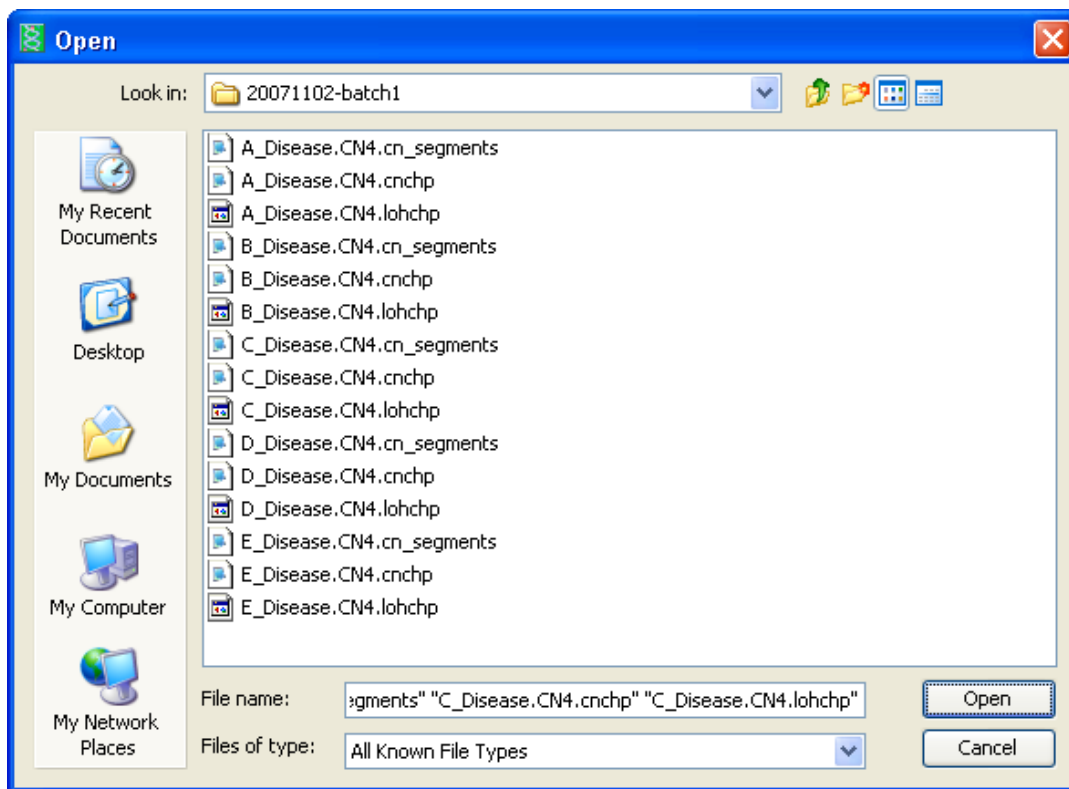


Figure 13. Open dialog box

2. Browse to the location with the copy number files you want to open.
3. Select the files.
4. Click **Open**.

The files are displayed in the Copy Number Browser.

Overview of Parts

If the CN Segments data is available, the Browser opens to the Karyoview (Figure 14).

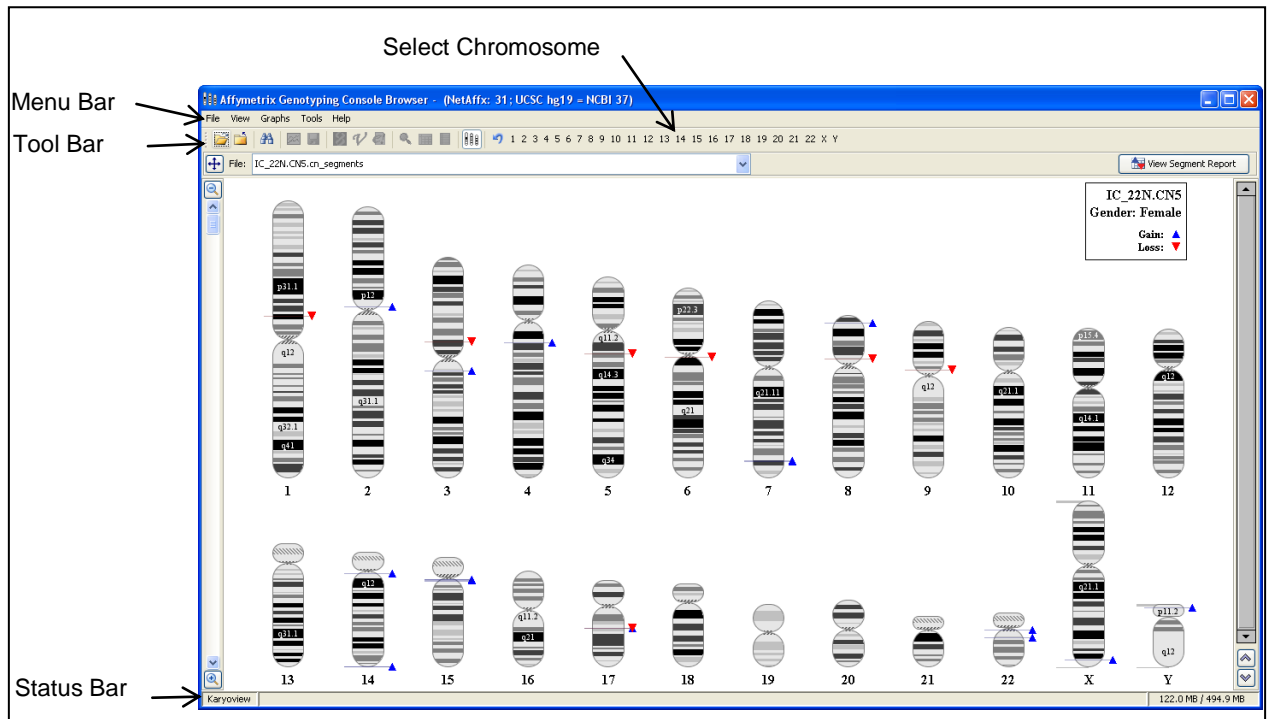


Figure 14. Karyoview (common Browser parts)

If Copy Number Segments data is not available, or if you click on a chromosome icon, the Chromosome view is displayed (Figure 15).

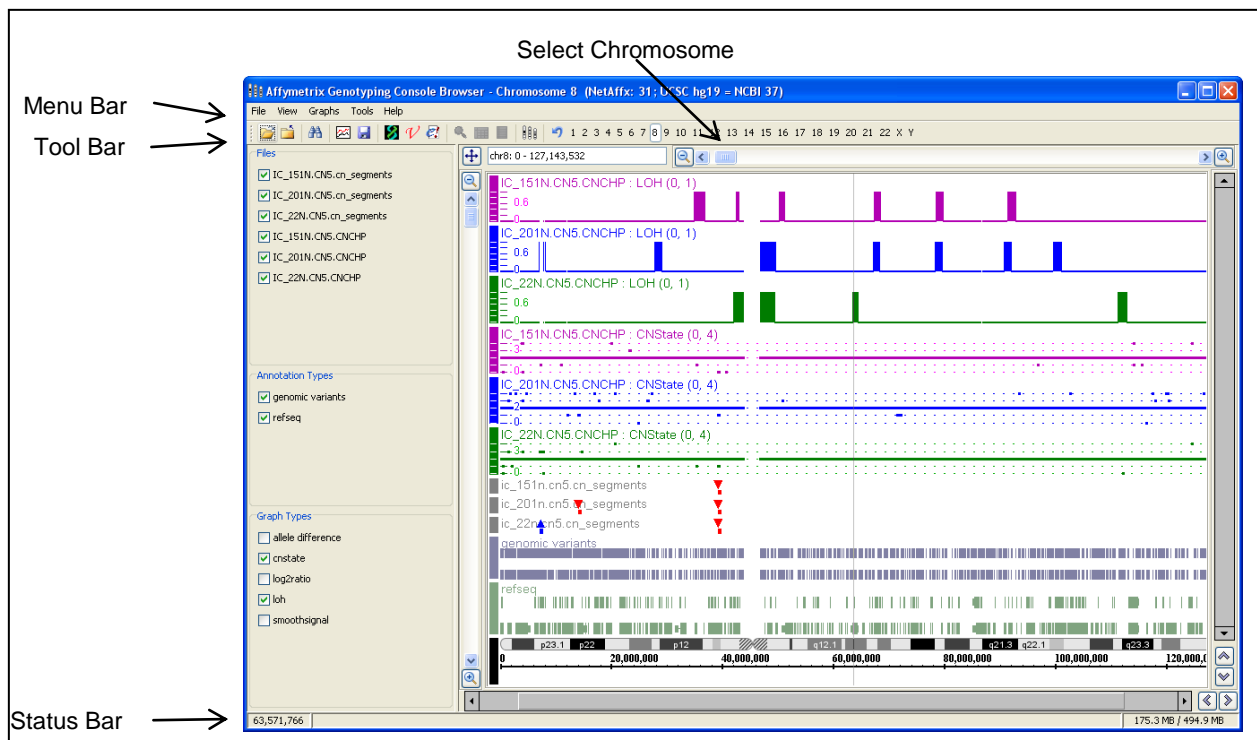


Figure 15. Chromosome view (common browser parts)

The two views share the following common components:

- Menu Bar: Access to the functions of the browser.
- Tool Bar and Chromosome Selector: Quick access to the functions of the Browser/Select chromosome for display in Chromosome display.
- Status Bar: displays:
 - Genomic position of the hairline cursor in Lower left corner
 - Memory in use/available in lower right corner. The maximum memory available is set by a start-up script (GenotypingConsoleBrowser.config) and can be modified from 512m to higher numbers based on system capacity if necessary by editing that file.

Karyoview

The Karyoview (Figure 16) provides an overview of the copy number change regions. In this view the entire genome is represented visually with Chromosome ideograms arranged vertically. Within this view cytobands on each ideogram are represented and annotated. On this view gains and losses are represented by blue and red triangles respectively.

The Segment Report provides segment-by-segment gain and loss information in a table.

Clicking on rows in the CN_Segments table or upon regions of the chromosome zooms into the Chromosome View of that region



Figure 16: Karyoview

It displays the copy number region data arranged by genomic position on a chromosome-by-chromosome basis.

- Copy Number Gains are indicated by blue triangles and by a semi-transparent blue stripe over the chromosome. The stripe is scaled to the exact size of the gain region on the chromosome.
- Copy Number Losses are indicated by red triangles and by a semi-transparent red stripe over the chromosome (Figure 17). The stripe is scaled to the exact size of the loss region on the chromosome.

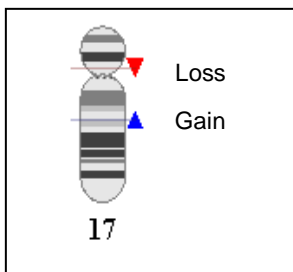


Figure 17. Loss and Gain indicators

For SNP 6.0 Arrays the Segment Report also displays a gender determination made by the Segment Reporting Tool for the sample, based on the detected copy number state for the X and Y chromosomes. See *Appendix D: Gender Call Issues in GTC 4.1* in the *Affymetrix® Genotyping Console 4.1 User Manual* for more information about the CN Segment Report gender call.

To select a file for a different CN Segment Report:

- Select from the File drop-down list (Figure 18).

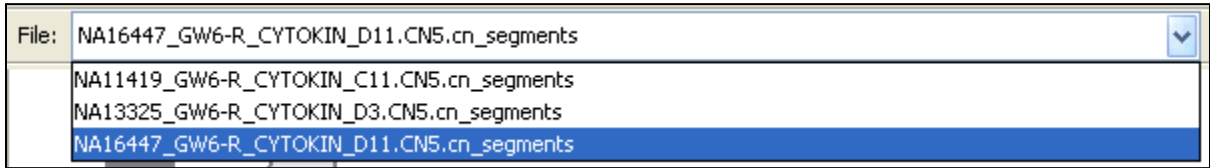


Figure 18. Selecting file

Using the Stretch Slider to Magnify the Vertical Scale

The Stretch Slider controls the vertical stretch of the Display area (Figure 19).

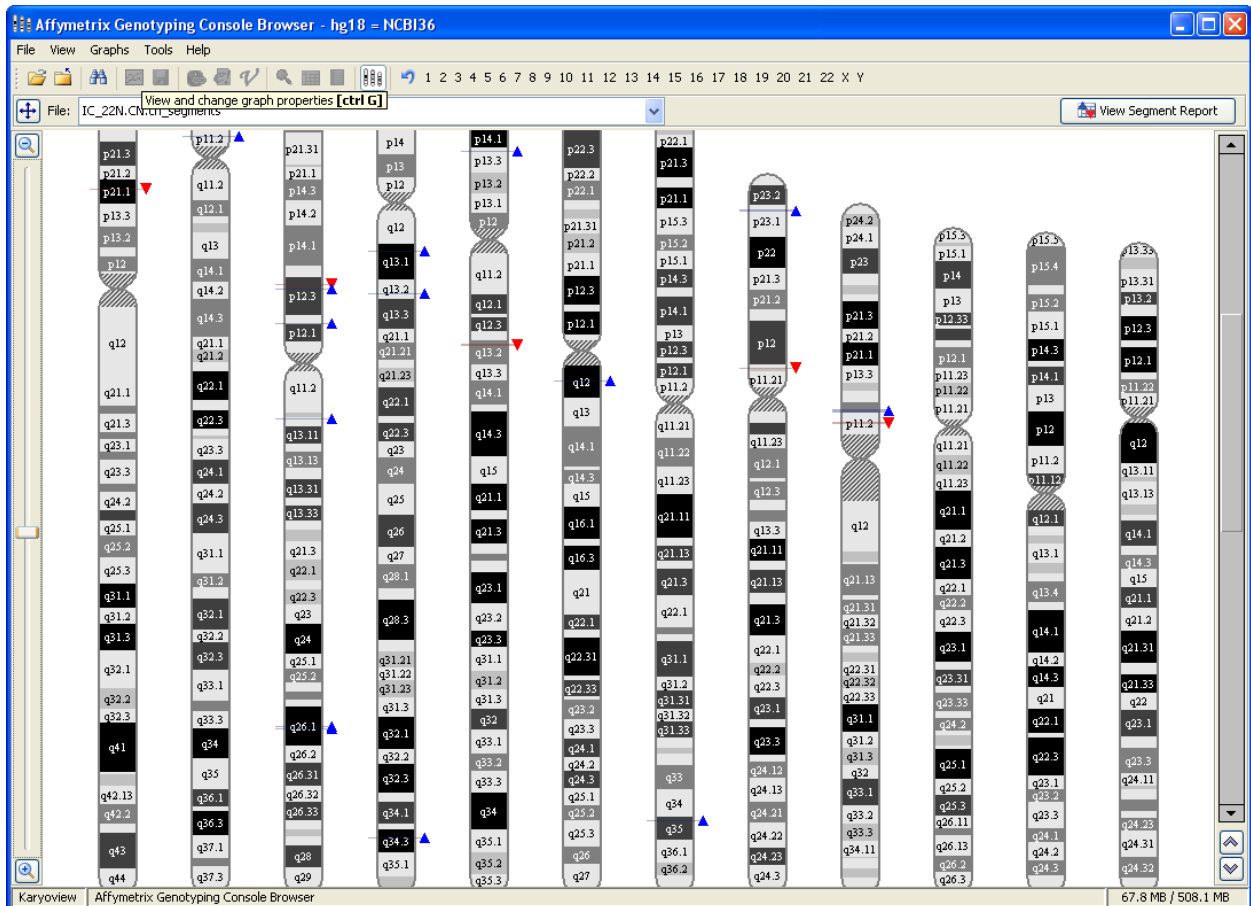


Figure 19. Vertical stretch enlarged using the slider

At higher magnifications, more details of the cytobands are displayed in the Karyoview

Selecting Chromosome or Region for Display

To select a Chromosome for display:

- Left-click on the chromosome in the Karyoview display (Figure 20).



Figure 20. Left-Clicking on the Chromosome

The full chromosome is displayed in the Chromosome Display (Figure 21).

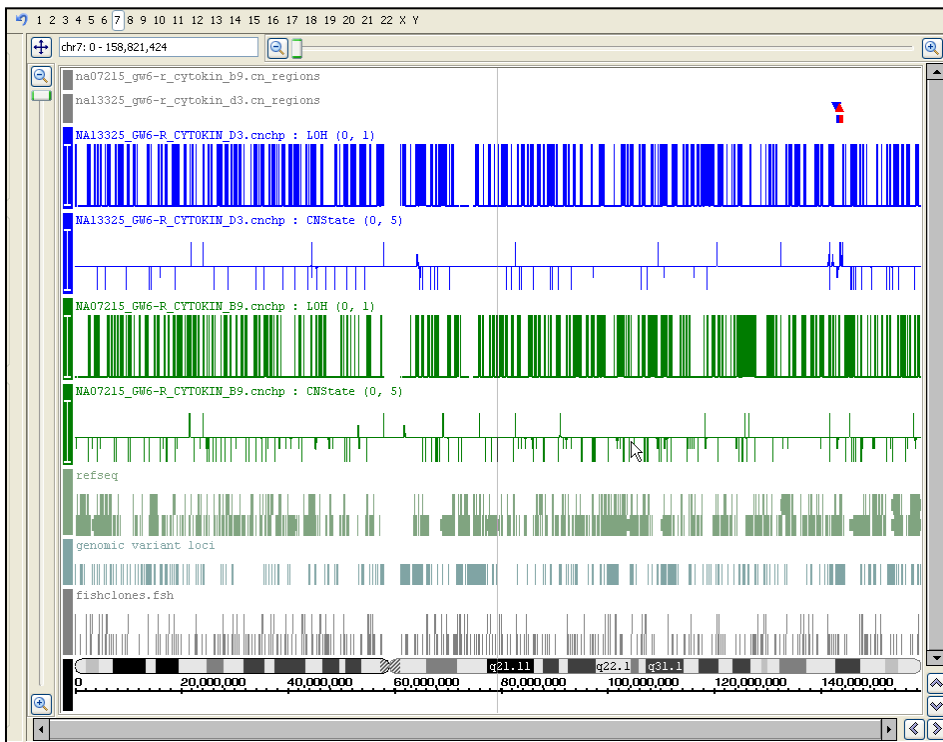


Figure 21. Full chromosome displayed

To select a copy number region for display:

- Left- click on the copy number segment indicator (Figure 22).

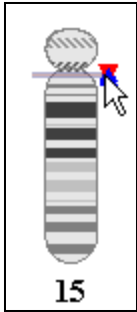


Figure 22. Selecting the copy number segment indicator

A zoomed-in view of the segment is displayed in the Chromosome Display (Figure 23).

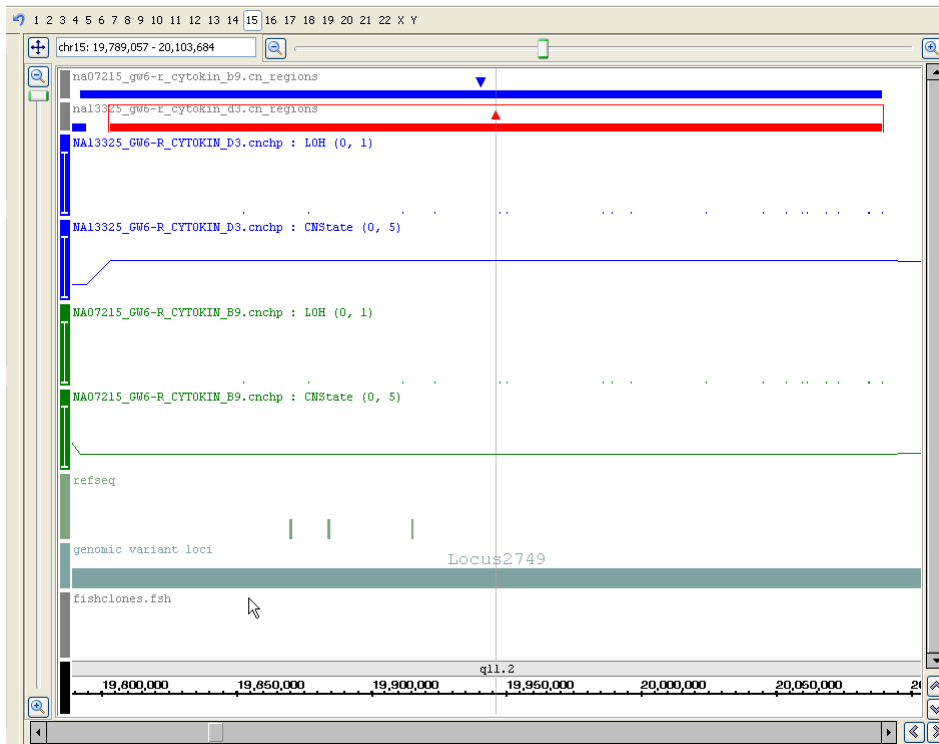


Figure 23. Zoomed-in region

Viewing the Segment Report

Segment Report displays the information in the Segment file in a table format.

To open the Segment Report:

- Click on the View Segment Report button in the Karyoview.
The Segment Report appears (Figure 24).

Sample	Copy Number State	Loss/Gain	Chr	Cytoband_Start_Pos	Cytoband_End_Pos
CCL-256D.CN4.cnchp	1	Loss	1	p36.33	p36.32
CCL-256D.CN4.cnchp	1	Loss	1	p36.32	p36.32
CCL-256D.CN4.cnchp	1	Loss	1	p36.21	p36.21
CCL-256D.CN4.cnchp	3	Gain	1	p34.3	p31.1
CCL-256D.CN4.cnchp	1	Loss	1	p31.1	p31.1
CCL-256D.CN4.cnchp	1	Loss	1	p31.1	p21.1
CCL-256D.CN4.cnchp	3	Gain	1	p21.1	p13.3
CCL-256D.CN4.cnchp	1	Loss	1	p13.3	p13.3
CCL-256D.CN4.cnchp	3	Gain	1	p13.2	p13.2
CCL-256D.CN4.cnchp	1	Loss	1	p13.2	p12
CCL-256D.CN4.cnchp	3	Gain	1	q21.1	q22
CCL-256D.CN4.cnchp	3	Gain	1	q43	q43
CCL-256D.CN4.cnchp	3	Gain	1	q44	q44
CCL-256D.CN4.cnchp	3	Gain	1	q44	q44
CCL-256D.CN4.cnchp	3	Gain	1	q44	q44
CCL-256D.CN4.cnchp	3	Gain	1	q44	q44
CCL-256D.CN4.cnchp	3	Gain	2	p25.3	p25.3
CCL-256D.CN4.cnchp	3	Gain	2	p25.3	p25.3

Figure 24. Segment Report

This Report is the table version of the output of the Segment Reporting Tool, the cn_segments file. Each row represents a particular copy number change segment which deviates from the expected normal.

- ! This table can be used to “drive” the Browser by allowing users to “hop” from Copy Number change segment to segment.
- ! Data can be sorted by clicking on column headers. You can re-sort the row order in the results by one or more criteria. Click on the column header to set the primary sort column. Control-click on additional column headers to set secondary sorting criteria.
- ! Columns can be moved by clicking, dragging and dropping them by their headers

Sample	File name.
Copy Number State	CN state of the last marker in a segment as estimated by the HMM.
Loss/Gain	Whether the Copy number change is a decrease or increase from the expected normal value.
Chr (Chromosome)	Chromosome where the segment is located.
Cytoband_Start_Pos	The Chromosome’s cytoband within which a Copy Number change segment begins.
Cytoband_End_Pos	The Chromosome’s cytoband within which a Copy Number change segment ends.
Size (kb)	Size of the segment of Copy Number change.
#Markers	Number of SNPs+CNV markers within the segment.
Avg_DistBetweenMarkers(kb)	Length of segment divided by number of markers encompassed by that segment.

%CNV_Overlap	Percentage of markers in a segment which overlap the boundaries of a known CNV.
Start_Linear_Pos	Base pair position on the Chromosome at which the first marker in the segment begins (going from top of the p-arm to the bottom of the q-arm of the chromosome).
End_Linear_Position	Base pair position on the Chromosome at which the last marker in the segment begins (going from top of the p-arm to the bottom of the q-arm of the chromosome).
Start_Marker	Name of the first SNP or CN marker of a Copy Number change segment.
End_Marker	Name of the last SNP or CN marker of a Copy Number change segment.
CNV_Annotation	Information from the Toronto Database of Genomic Variants about the CNV variants which overlap the Copy Number change segment.

You can export data for use in other applications.

To export to TSV format:

1. From the Tools menu, select **Export All to TSV...**; or

Click the **TSV button**  in the toolbar.

The Save dialog box opens (Figure 25).

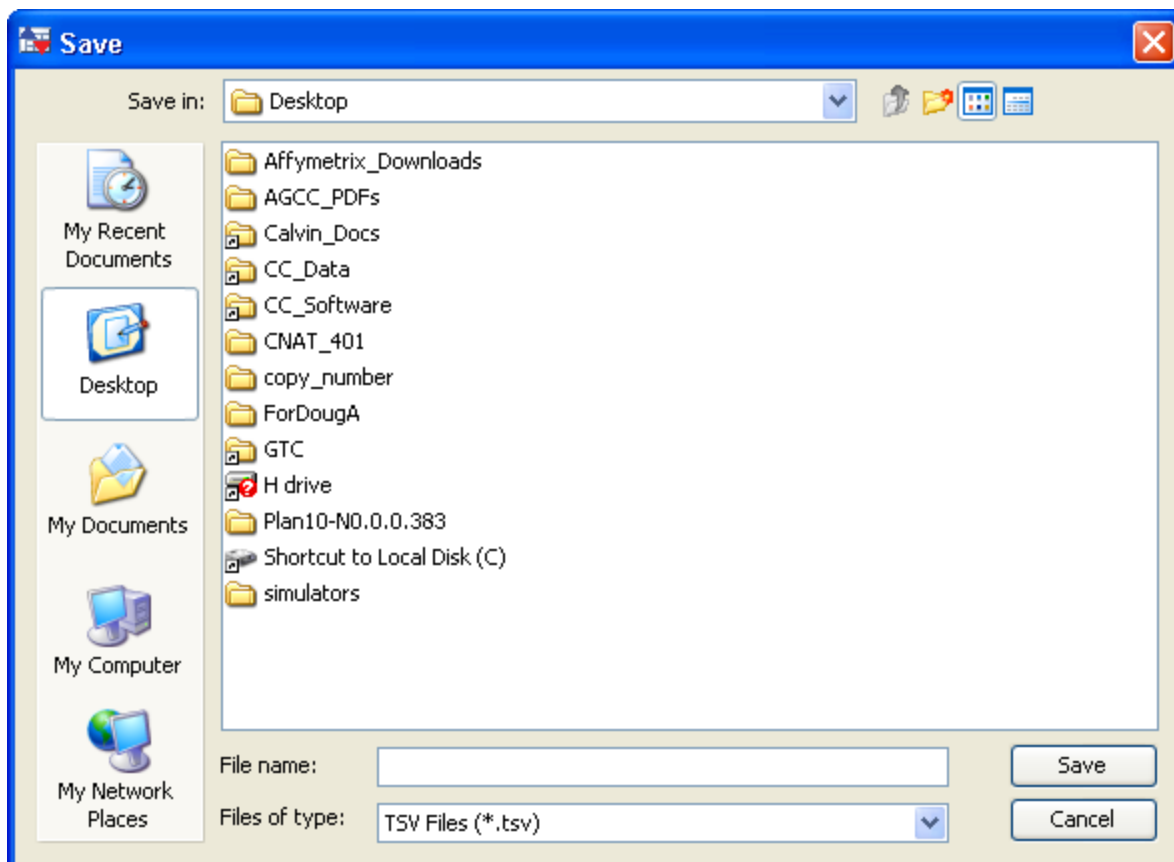


Figure 25. Save dialog box.

2. Select a save location and enter a file name for the TSV file.
3. Click **Save**.

The TSV file is saved in the specified location.

To copy data to the clipboard:

1. Select the data you want to copy in the table.
2. From the Tools menu, select **Copy Selections**; or

Click the **Copy Selections button**  in the toolbar, or use Control+C.

The selected data is copied to the clipboard and can be pasted into other files.

Chromosome View

The Chromosome view (Figure 26) enables you to look in detail at the available data and annotations for copy number and LOH.

The GTC Browser can work with two distinct types of data:

- annotations

Annotations indicate the known or suspected locations of features, such as mRNA's, exons, FISH clones, structural variants, and so forth. Annotation data can be loaded from files.

For more information about annotations, see *Chapter 3: Annotations* (page 31).

- graphs

Graphs indicate scores or other numeric values as a function of genomic position. Graphs are generally displayed as some form of plot (x,y-plot, bar plot, etc.). The results from GeneChip[®] tiling arrays and from chromosome copy number analysis are generally represented as graphs. Simple graphs represent values for individual genomic positions. Graph data is generally loaded from files.

For more information about graphs, see

Chapter 4: Graphs (page 45).

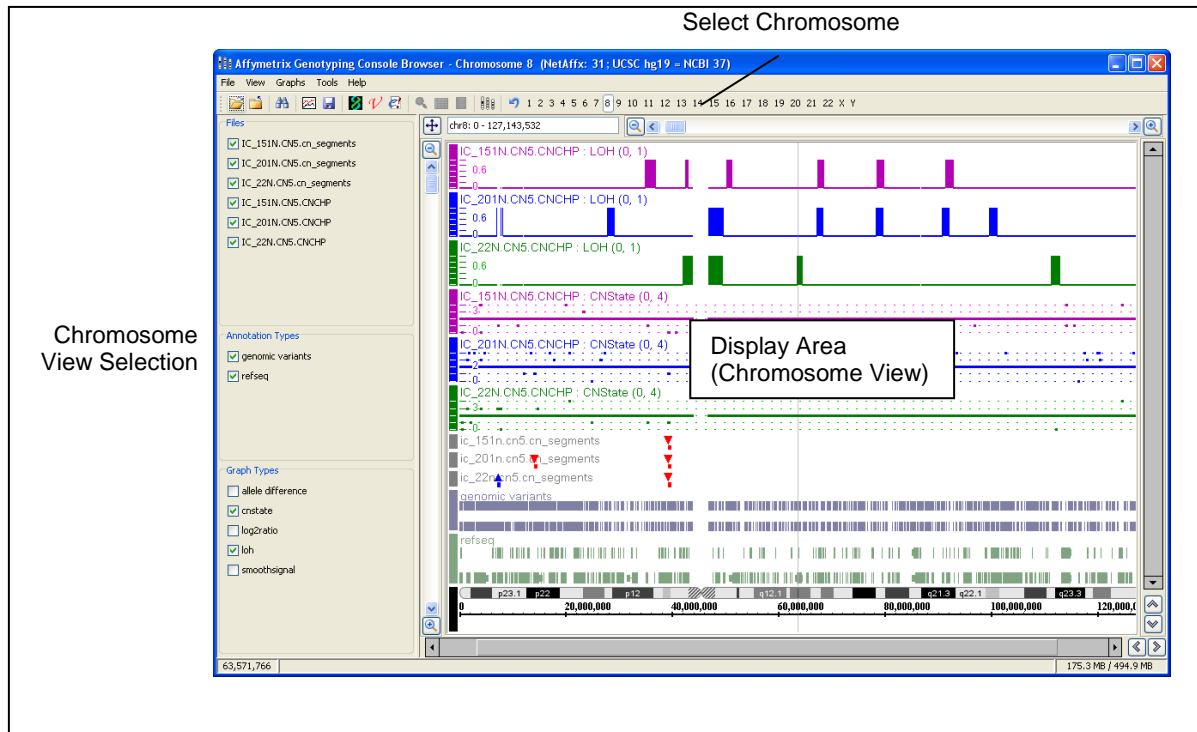


Figure 26. Parts of the GTC Browser (Chromosome view)

The Chromosome view has the following components:

- *Chromosome selection bar*: selects chromosome for display.
- *Chromosome view selection*: selects files, annotation types, and graph types for display.
- *Display Area*: location on screen where the data representations and annotations are displayed.

Selecting Files and Chromosomes for Display

To select a different Chromosome for display:

- Select from the Chromosome Selection bar (Figure 27), or
From the View menu, select **Display Chromosome**.
- You can also type the chromosome and coordinates into the Range box below the Chromosome Selection bar in the format “chr12: 10000-20000”.

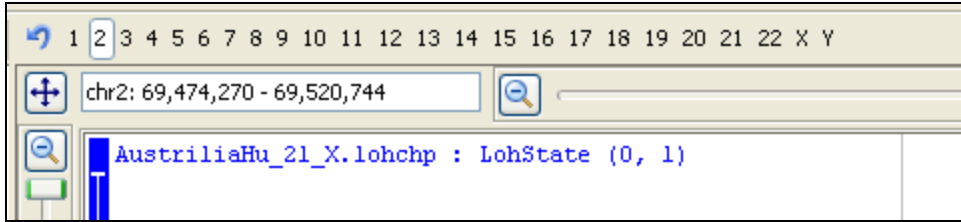


Figure 27. Chromosome selection bar

To select files for display:

- Select or deselect from the Files list on the left side of the screen (Figure 28).



Figure 28. Files list

Selecting annotations is described in [Selecting Annotations for Display](#).

Selecting graphs is described in [Selecting graph types for Display](#).

Display Area

The annotations and graphs are displayed in the Display area (Figure 29).

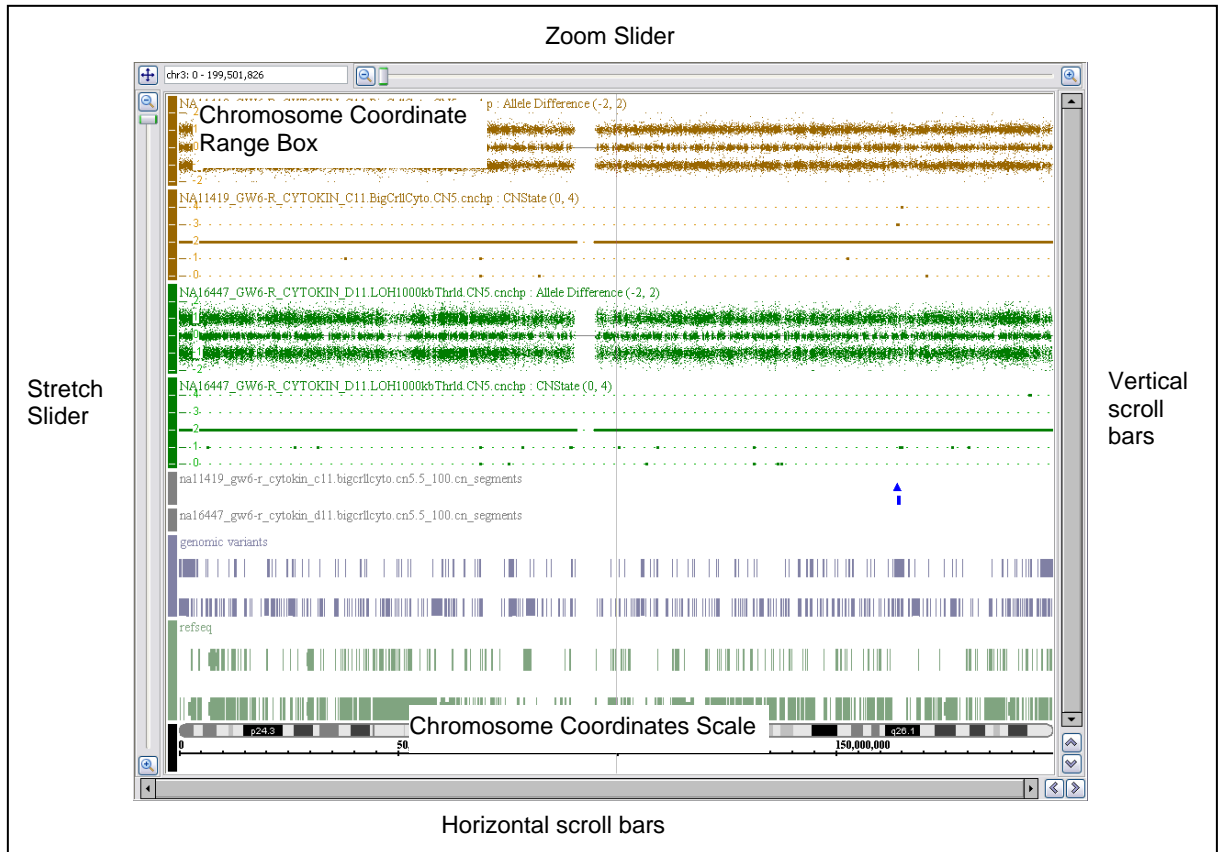


Figure 29. Chromosome View, display area controls

The display area has the following features and controls:

Chromosome Coordinates range box	Displays and selects the range of genome position coordinates displayed in the display box.
Chromosome Coordinates scale	Shows the position along the genome.
Zoom Slider	Controls the horizontal zoom and the area of the chromosome displayed.
Stretch Slider	Controls the vertical stretch of the Display area.
Scroll bars	Used to select the area displayed after zooming or stretching the vertical or horizontal scale.

Navigation in the Chromosome view

This section describes how to manually navigate around in the Chromosome display to see annotations on the genomes and chromosomes you have loaded and how to zoom-in on areas of interest. Many of these navigation functions have “shortcut” keys. Hold your mouse over the button to see the tool-tip telling you what the shortcut key is for each button. For example, zoom in vertically with “control + shift + right arrow”.

Zooming and scrolling horizontally

The horizontal zoom slider (Figure 30) is located along the top edge of the viewer.

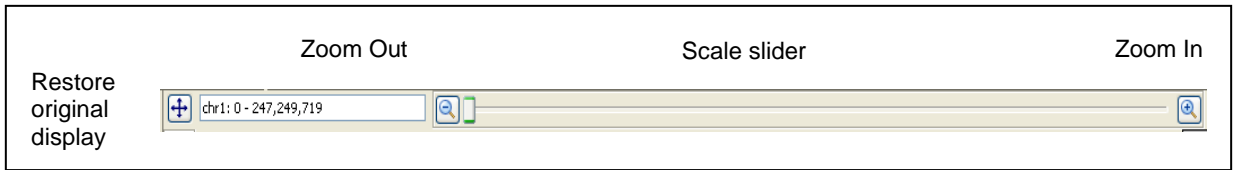


Figure 30. Horizontal Zoom Slider (with Chromosome Coordinate range box and Restore Original button)

To zoom in, move the zoom slider to the right. To zoom out, move the zoom slider to the left.

The hairline marks the focus of horizontal zooming. Click in any empty position in the view to reset the hairline position and thus reset the center of zooming.

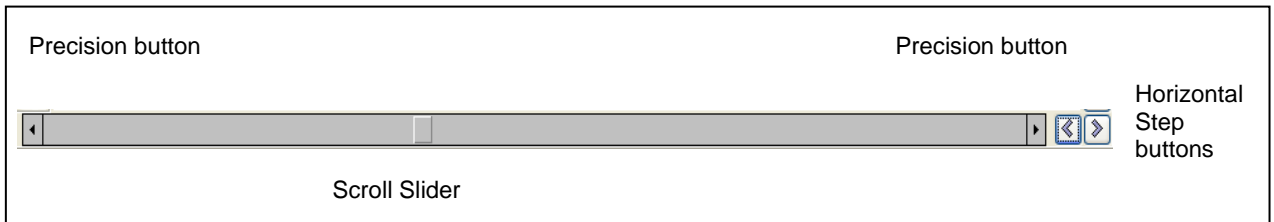


Figure 31. Horizontal Scroll controls

When you are zoomed in, you can scroll through the view in several ways:

- Use the horizontal scroll box slider (Figure 31) at the bottom of the viewer panel to scroll along the chromosome. Slider size and speed of movement will vary according to the current zoom level.
- Click the Horizontal step buttons to move in larger increments.
- Click in the scroll bar trough on either side of the slider to move it in the direction of your click.
- For finest precision when zoomed in, click the arrows at the ends of the scroll bar.

When you are zoomed all the way out (the horizontal zoom control slider is at the far left), you are already viewing the entire range of the loaded data (entire chromosome) and the scroll box slider is not available.

Zooming and scrolling vertically

Zooming and scrolling vertically are similar to zooming and scrolling horizontally. Zooming vertically is especially useful for viewing annotation types with many rows of annotations. To learn about collapsing and expanding annotations in the display, see **Expanding and Compressing**.

The vertical zoom slider is located immediately to the left of the main view. To zoom the view vertically, drag the vertical slider down.

After expanding vertically, use the vertical scrollbar on the right of the display to view different annotation rows.

Going to a Specific Coordinate

To go to a specific coordinate or coordinate range, use the coordinate range box (Figure 32) located at the left-hand side of the horizontal zoom slider, which is directly above the main view.

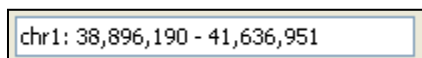


Figure 32. Coordinate Range Box

The range box displays the current coordinate range. But you can also type a new location into the box to go there quickly. Enter the desired location in any one of these formats then press the <Enter> key:

- “**chromosome : start - end**”: sets the view to the given start and end coordinates on the given chromosome. This is the same format used by the UCSC genome browser, so you can easily copy coordinates from that application. Example: “chr3:100000-200000”.
- “**start : end**” or “**start - end**”: sets the view to the given start and end coordinates of the current chromosome.
- “**chromosome : start + width**” or “**start + width**”: sets the view to the given start coordinate and sets the zoom level such that the given width of coordinates is visible.
- “**position**”: moves the hairline to the given position. Example: “100,000”. If possible at the current zoom level, the view will be scrolled such that the given position is in the center of the view.

If the specified chromosome or position does not exist in the currently loaded genome, an error message will be shown.

Viewing the Selected Region in another Browser

You can view the region selected in the Display area at one of the following public sites:

- UCSC
- Ensembl
- Toronto DGV

To view the region at a public site:

1. Adjust settings as described above so that the entire region of interest is visible in the Display Area (Figure 33).

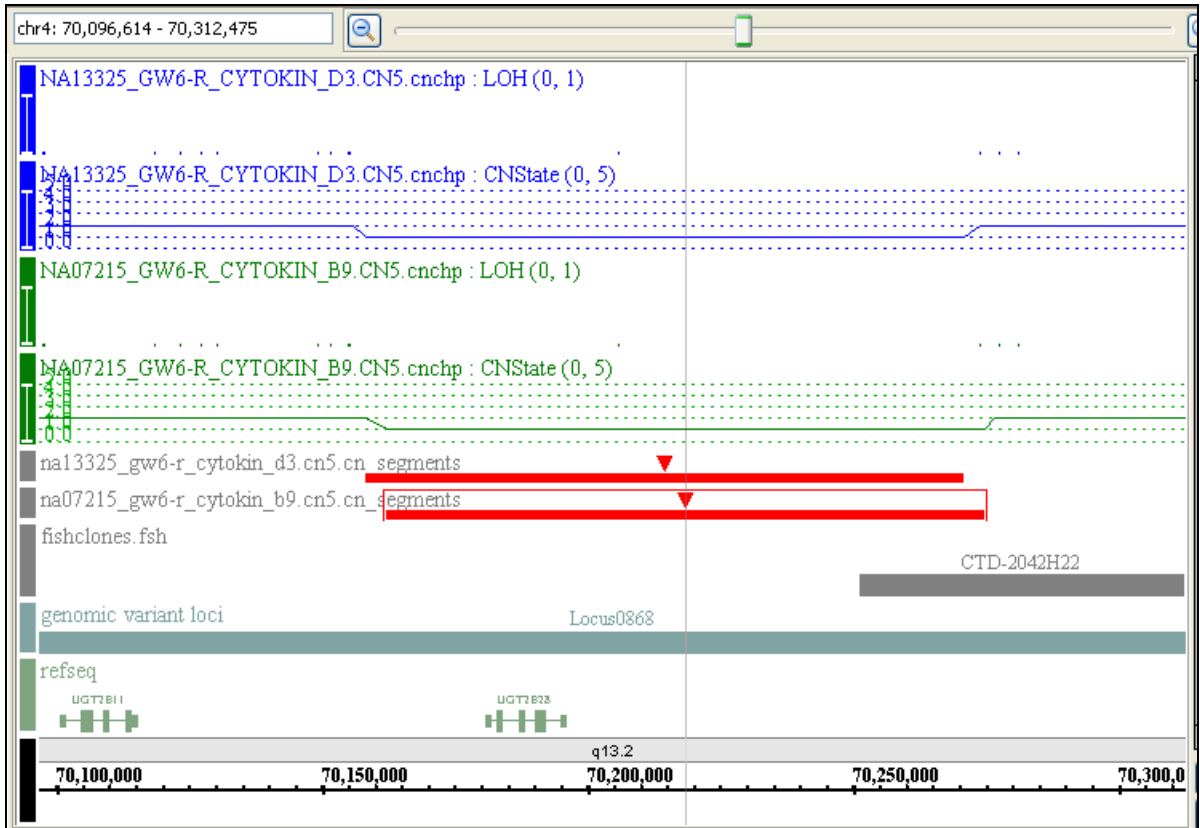


Figure 33. Selected region in Display area

- From the View menu, select **View Region at [site name]**.

A web browser opens with the public site displaying the chromosome region selected in the GTC Browser (Figure 34).

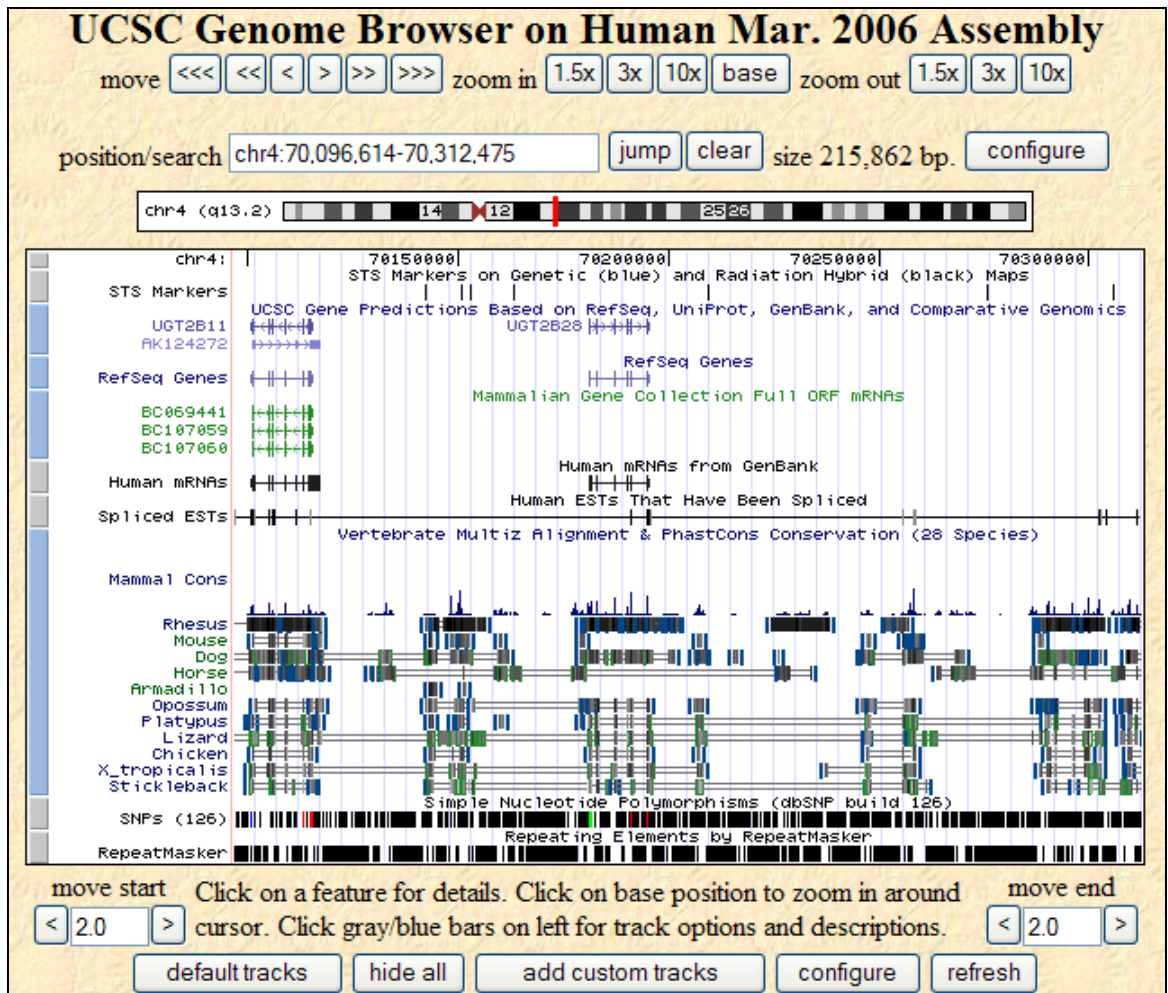


Figure 34. Region displayed in UCSC Genome Browser

Chapter 3: Annotations

Annotations indicate the known or suspected locations of features, such as mRNA's, exons, promoter regions, pseudogenes, genomic variants, and FISH/BAC clones. They provide information you can use to evaluate your results.


This section discusses:

- *Annotation Types* (below)
- *Selecting Annotations for Display* (page 32)
- *Loading Other Annotations* (page 33)
- *Expanding and Compressing Annotations* (page 33)
- *Learning More about Annotations* (page 34)

Annotation Types

The GTC 4.1 browser can display the following annotation types:

- Fishclones
- Genomic Variants
- RefSeq
- Copy Number Segment data generated by GTC.

 **Some annotation types may not be available in all annotation builds.**

Fishclones

This track shows the location of fluorescent in situ hybridization (FISH)-mapped clones along the sequence. The locations of these clones were contributed as a part of the BAC Consortium paper Cheung, V.G. *et al.* (2001). More information about the BAC clones, including how they may be obtained, can be found at the *Human BAC Resource* and the *Clone Registry* web sites hosted by *NCBI* and at <http://genome.ucsc.edu/cgi-bin/hgTrackUi?g=fishClones>.

 **The Fishclones track is not yet available for the current NetAffx browser Genome Annotation file (NetAffxGenomicAnnotations.Homo_sapiens.hg19.na31.db).**

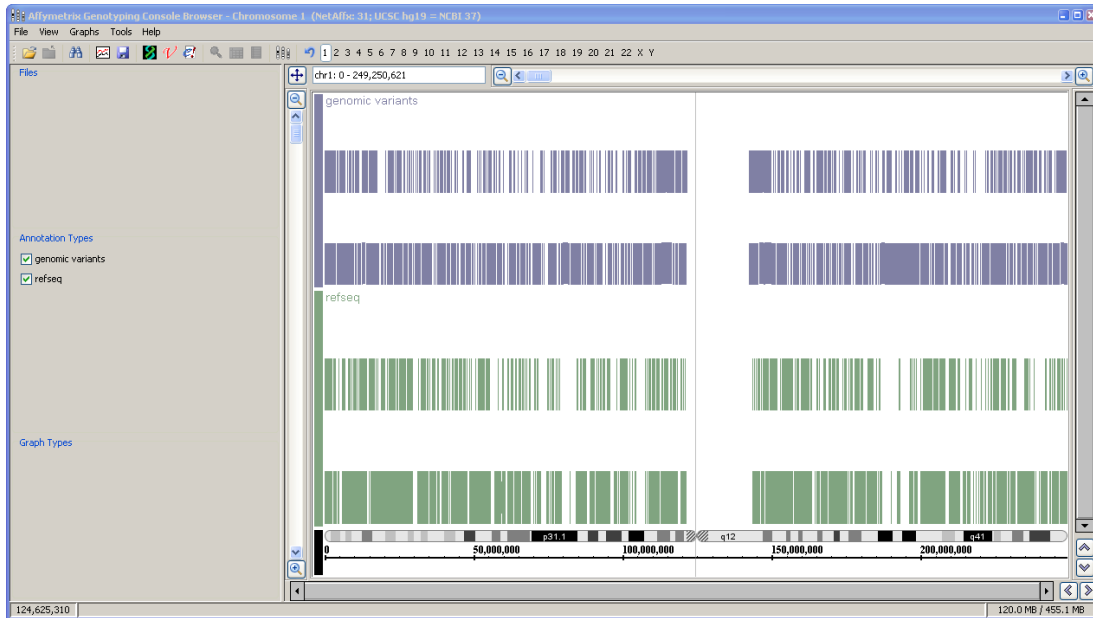


Figure 35. GTC browser without Fishclones track

Genomic Variants

This annotation shows regions detected as putative copy number polymorphisms and sites of detected intermediate-sized structural variation. They were determined by various methods; more information can be found at: <http://projects.tcag.ca/variation/>

The term "variant" is used to describe each Copy Number polymorphic or other structural variant region as reported in a published study. Each variant is assigned a unique identifier, which is static and does not change over time.

RefSeq

Annotations taken originally from NCBI RefSeq database, as housed on the UCSC Genome Bioinformatics site, with information on mRNA's, exons, promoter regions, pseudogenes. See:

- <http://www.ncbi.nlm.nih.gov/RefSeq/>
- <http://genome.ucsc.edu/index.html>

The annotations were updated to match the NA26 annotations used by NetAffx for release. For additional information, check the Affymetrix.com support page: http://www.affymetrix.com/support/help/releasedocs/netaffx_release_26.affx

Copy Number Segment Data

This annotation track shows segments on the chromosome of copy number variation as detected by the Copy Number Segment Reporting Tool in Genotyping Console 4.1.

Selecting Annotations for Display

To select annotations for display:

- Select or deselect from the Annotations Types list on the left side of the screen (Figure 36).

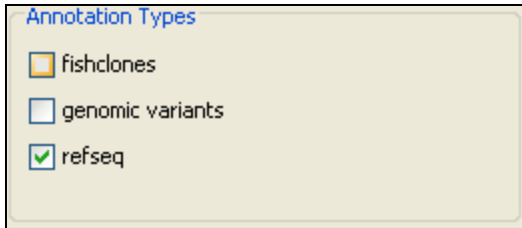


Figure 36. Annotation Types list

Loading other annotations

You can load annotation data of your own choice in BED format. The BED format is a simple tab-separated values file with three required columns containing coordinates and nine optional ones.

BED files are required to be named with the extension “.bed”.

Refer to <http://genome.ucsc.edu/FAQ/FAQformat> for details of the format. GTC Browser 1.2 supports all the variations of the BED format listed there.

Expanding and Compressing Annotations

For tracks containing multiple rows of annotations, collapsing tracks consolidates all rows within a track into 2 rows (Figure 37). Any annotations after the first one will be drawn together in the second row.

When there are multiple annotations of one type at the same coordinate, the separate annotations will be shown on separate rows.

Collapsing tracks is useful if you don't need to see all the details. But be aware that in collapsed tracks larger annotations may obscure smaller ones; annotations with introns may be obscured by annotations that don't show the intron.

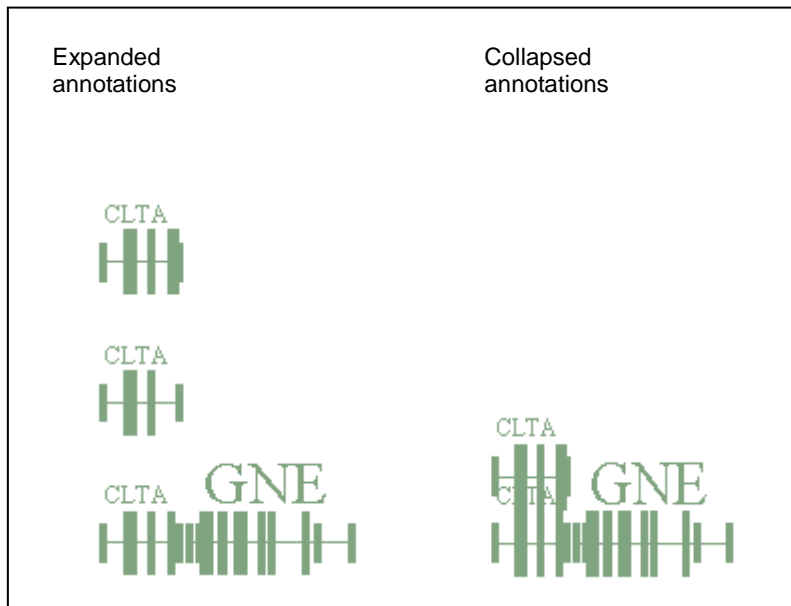


Figure 37. Collapsing and expanding annotations

To collapse or expand annotations:

- From the View menu, select:
 - Collapse All Annotations
 - Expand All Annotations

All strands of the annotations will be affected.

Learning More about Annotations

You can learn more about annotations using the:

- *Selection Details table* (below)
- *List View table* (page 41)

Selection Details

The Selection Details table displays information about a selected annotation:

To learn more about an annotation:

1. Right-click on the item in the Display area.

The context-sensitive menu appears (Figure 38).

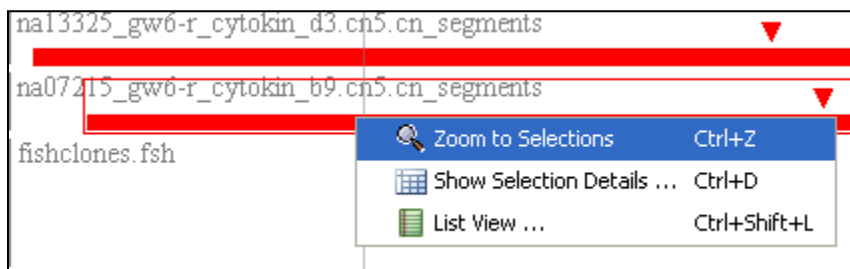


Figure 38. Context menu for a CN-Segments region

2. Select Show Selection Details.

The Selection Details dialog box appears (Figure 39).

property	Loss chr4:70162221-70274566
id	Loss chr4:70162221-70274566
start	70162221
end	70274566
length	112345
Chr	4
Start_Linear_Pos	70162221
End_Linear_Position	70274566
Cytoband_Start_Pos	q13.2
Cytoband_End_Pos	q13.2
Size(kb)	112
CNV_Annotation	Variation_0701 // chr4:70151264-70310376 //...
method	NA07215_GW6-R_CYTOKIN_B9.CN5.cn_segm...
#Markers	60
Start_Marker	CN_1113956
Copy Number State	1
%CNV_Overlap	3%
Sample	NA07215_GW6-R_CYTOKIN_B9.CN5.cnchp
End_Marker	CN_1116067
Avg_DistBetweenMarkers(kb)	2
Loss/Gain	Loss

Figure 39. Selection Details for CN_Segment

The information displayed differs depending upon what type of annotation is being selected:

- Copy Number Segment
- FISHclones
- Genomic Variant
- RefSeq

To transpose the data in the display:

- From the Tools menu, select **Transpose Table**.
The data is displayed in a single row (Figure 40).

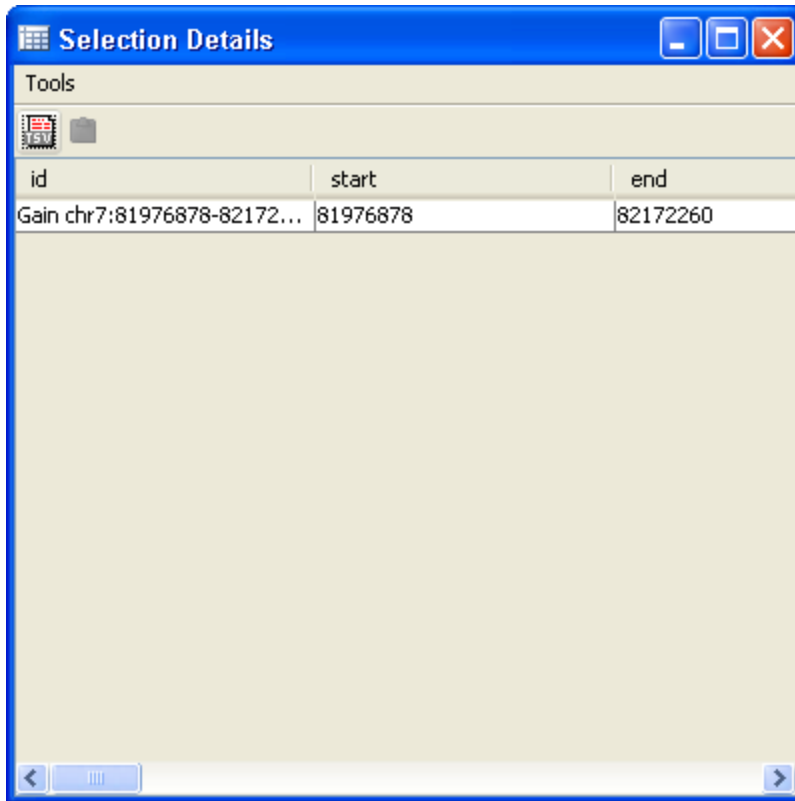


Figure 40. Transposed table

To export to TSV format:

1. From the Tools menu, select **Export All to TSV...**; or

Click the **TSV button**  in the toolbar.

The Save dialog box opens (Figure 41).

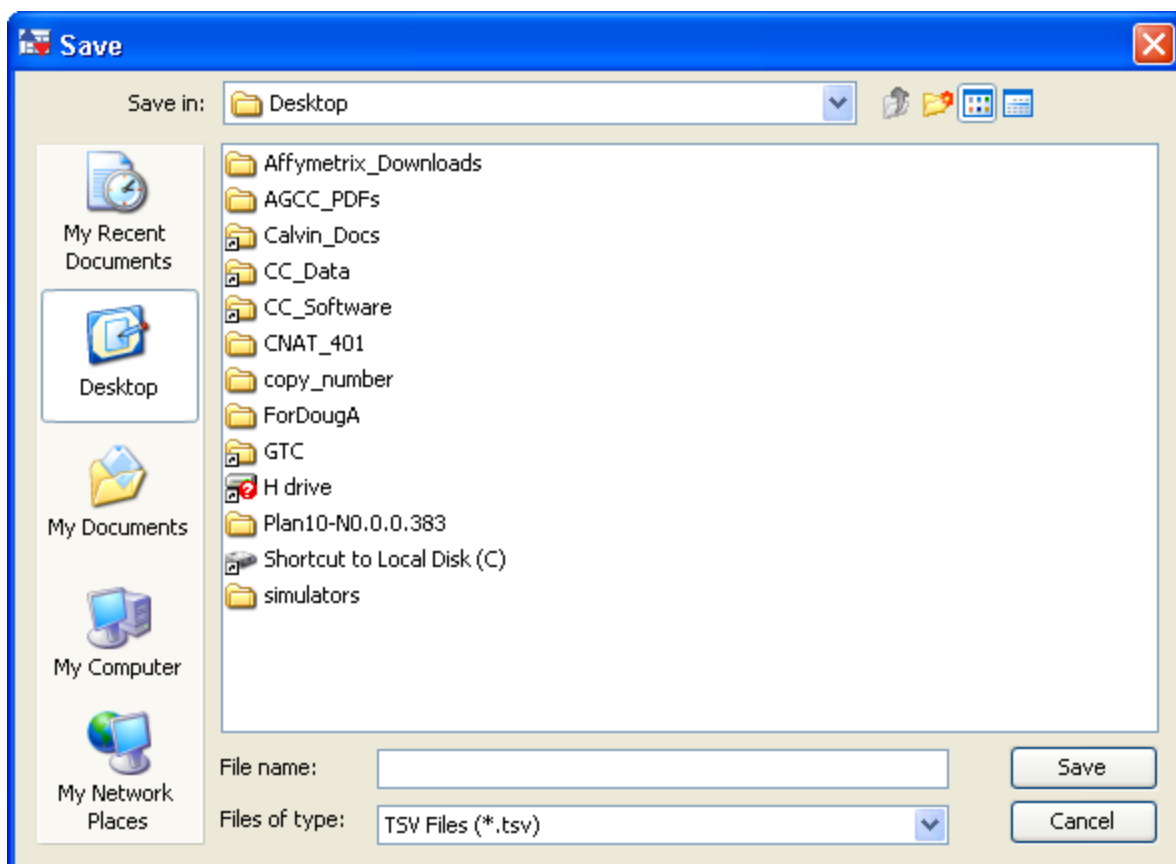


Figure 41. Save dialog box.

2. Select a save location and enter a file name for the TSV file.
3. Click **Save**.

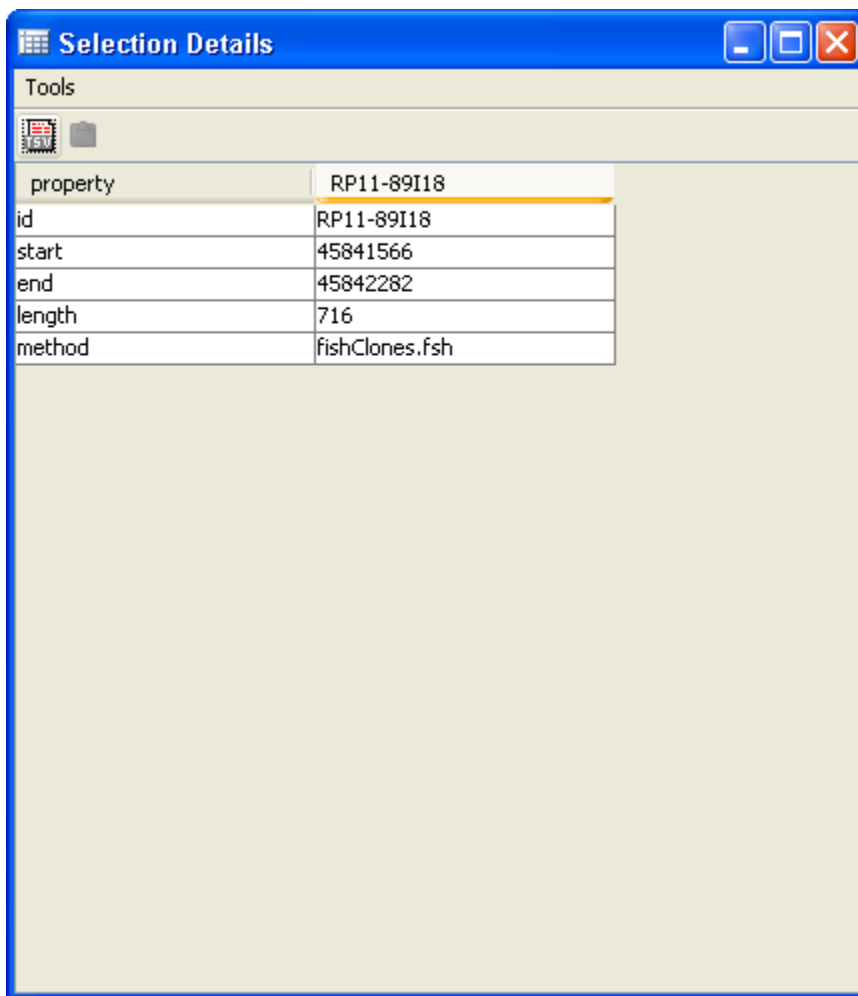
If you do not add a “.tsv” extension to the file name, this will be done the first time the **Save** button is clicked; the **Save** button must then be clicked again to save the .tsv file.

The TSV file is saved in the specified location.

Copy Number Segment Details

When you select a segment in the Copy Number Segment track, the Selection Details table (Figure 42) displays information on a particular copy number change segment. It displays the same information as in the *Segment Report*.

Fishclones Details



The screenshot shows a window titled "Selection Details" with a blue title bar and standard Windows window controls. Below the title bar is a "Tools" section with a small icon. The main content is a table with the following data:

property	RP11-89I18
id	RP11-89I18
start	45841566
end	45842282
length	716
method	fishClones.fsh

Figure 42. Selection Details for fishClones annotation

The following information is displayed for fishClones annotations (Figure 42):

Property	Field descriptor for a row in the table.
ID	Name of the fluorescent in situ hybridization (FISH)-mapped clone.
Start	Physical position where FISH clone begins.
End	Physical position where FISH clone ends.
Length	Length of FISH clone in bp.
Method	Source file for table information.

Genomic Variant Details

The screenshot shows a window titled 'Selection Details' with a 'Tools' toolbar. Below the toolbar is a table with the following data:

property	Variation_3769
id	Variation_3769
chr	chr9
start	38557168
end	46588540
length	8031372
type	Genomic Variants
Landmark	chr9:38557168..46588540
VariationType	CopyNumber
Reference	Redon et al. (2006)
PubMedID	17122850
Method/platform	WGTP CGH Array
Gain	
Loss	
TotalGainLossInv	216
SampleSize	270 HapMap individuals

Figure 43. Selection Details for Genomic Variants annotation

The following information is displayed for Genomic Variants annotations (Figure 43).

Property	Field descriptor for a row in the table.
ID	Variant's unique identifier.
Start	Position of base pair assigned as start of variant.
End	Position of base pair assigned as end of variant.
Length	Length of variant in base pairs.
Landmark	Coordinates or genomic clone.
Variationtype	CNV, insertion, deletion, inversion or translocation.
Reference	Publication cited for this variant.
PubMedID	PubMedID for the cited publication.
Method/platform	Molecular techniques or other evidence used to identify variant.
Gain	Absolute number or frequency of gains of this variant

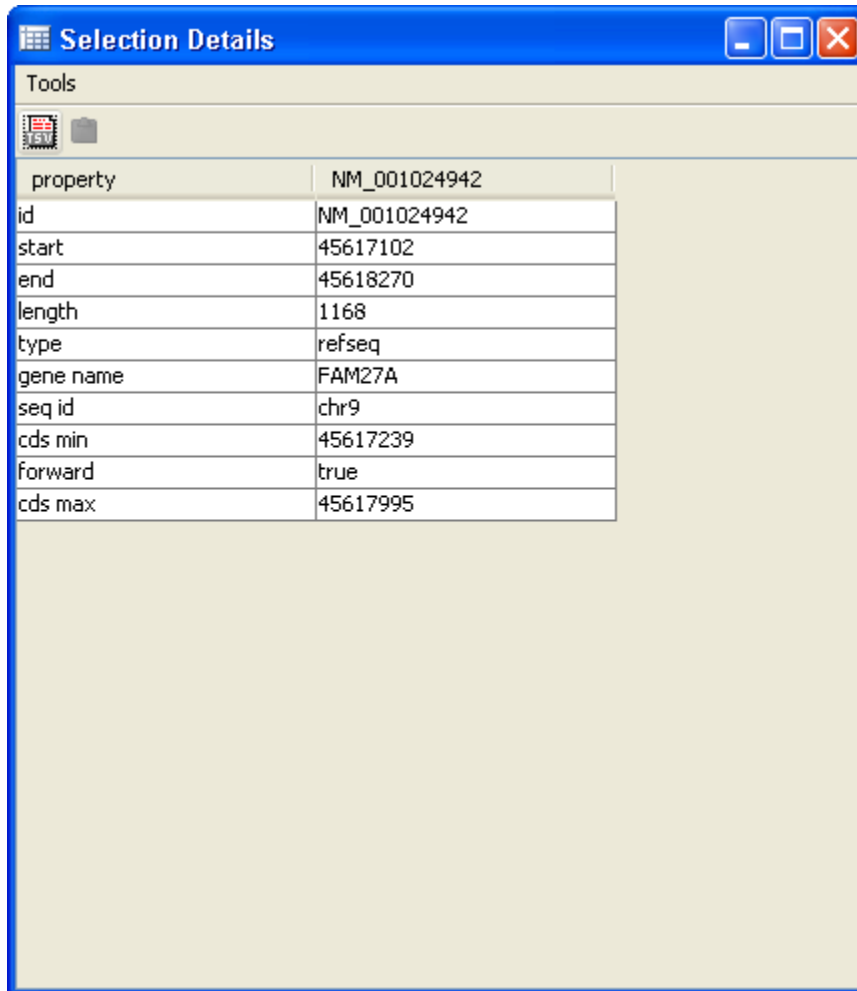
among cited study population.

Loss Absolute number or frequency of losses of this variant among cited study population.

TotalGainLossInv Absolute number or frequency of all gains+losses+inversions among cited study population.

SampleSize Number of individuals in the study population of the cited publication.

RefSeq Details



property	NM_001024942
id	NM_001024942
start	45617102
end	45618270
length	1168
type	refseq
gene name	FAM27A
seq id	chr9
cds min	45617239
forward	true
cds max	45617995

Figure 44. Selection Details for RefSeq annotation

The following information is displayed for RefSeq annotations Figure 44):

Property

id Refseq nucleotide Accession Number for gene.

Start Nucleotide position in base pairs where gene starts.

End	Nucleotide position in base pairs where gene ends.
Length	Length of selected gene or gene region (exon, promoter, etc.).
Type	Indicates which type of annotation you have selected. Always equals “refseq” for items in Refseq track.
Gene name	Gene Symbol used by Refseq.
Seq id	Chromosome name.
Cds min	Coding sequence’s smallest numerically ordered base pair.
Forward	On the + strand; value will be true or false.
Cds max	Coding sequence’s largest numerically ordered base pair.

List View

The List View table (Figure 45) can also be displayed when you right-click on an annotation in the Chromosome View:

Chromosome	Min	Max	Strand	ID	Type
chr21	34,740,884	35,047,400	+	Loss chr21:34740884-3504...	australiahu_21_x.cn_regions
chr21	35,631,536	36,049,947	+	Gain chr21:35631536-3604...	australiahu_21_x.cn_regions
chr21	37,104,302	37,208,844	+	Loss chr21:37104302-3720...	australiahu_21_x.cn_regions
chr21	37,931,173	38,152,962	+	Loss chr21:37931173-3815...	australiahu_21_x.cn_regions
chr21	39,019,706	39,301,359	+	Loss chr21:39019706-3930...	australiahu_21_x.cn_regions
chr21	39,669,876	39,953,480	+	Loss chr21:39669876-3995...	australiahu_21_x.cn_regions
chr21	41,332,614	43,883,204	+	Loss chr21:41332614-4388...	australiahu_21_x.cn_regions
chr21	44,963,638	46,924,583	+	Loss chr21:44963638-4692...	australiahu_21_x.cn_regions
chr22	16,849,599	18,014,330	+	Loss chr22:16849599-1801...	australiahu_21_x.cn_regions
chr22	20,312,249	20,751,249	+	Loss chr22:20312249-2075...	australiahu_21_x.cn_regions
chr22	27,999,648	30,006,454	+	Loss chr22:27999648-3000...	australiahu_21_x.cn_regions
chr22	31,091,715	31,604,166	+	Loss chr22:31091715-3160...	australiahu_21_x.cn_regions
chr22	32,076,526	32,396,846	+	Loss chr22:32076526-3239...	australiahu_21_x.cn_regions
chr22	34,247,492	34,915,188	+	Gain chr22:34247492-3491...	australiahu_21_x.cn_regions
chr22	35,978,628	38,639,276	+	Loss chr22:35978628-3863...	australiahu_21_x.cn_regions
chr22	39,200,740	41,677,130	+	Loss chr22:39200740-4167...	australiahu_21_x.cn_regions
chr22	43,945,366	44,149,283	+	Loss chr22:43945366-4414...	australiahu_21_x.cn_regions

1637 results

Figure 45. List View table

This table gives the positional information about a selected Browser object, allows export of this information as a BED file which can be loaded into the browser as a track

It displays the following information:

Chromosome	The chromosome the annotation is on.
Min	Minimum coordinate on the chromosome.
Max	Maximum coordinate on the chromosome.
Strand	Which strand of the DNA the annotation is on.
ID	The annotation ID, if any. For data from the CN_SEGMENTS

files, an ID is constructed from the location coordinates .

Type Identifies the file or annotation type for the annotation.

To export to TSV format:

1. From the Tools menu, select **Export All to TSV...**; or

Click the **TSV button**  in the toolbar.

The Save dialog box opens (Figure 46).

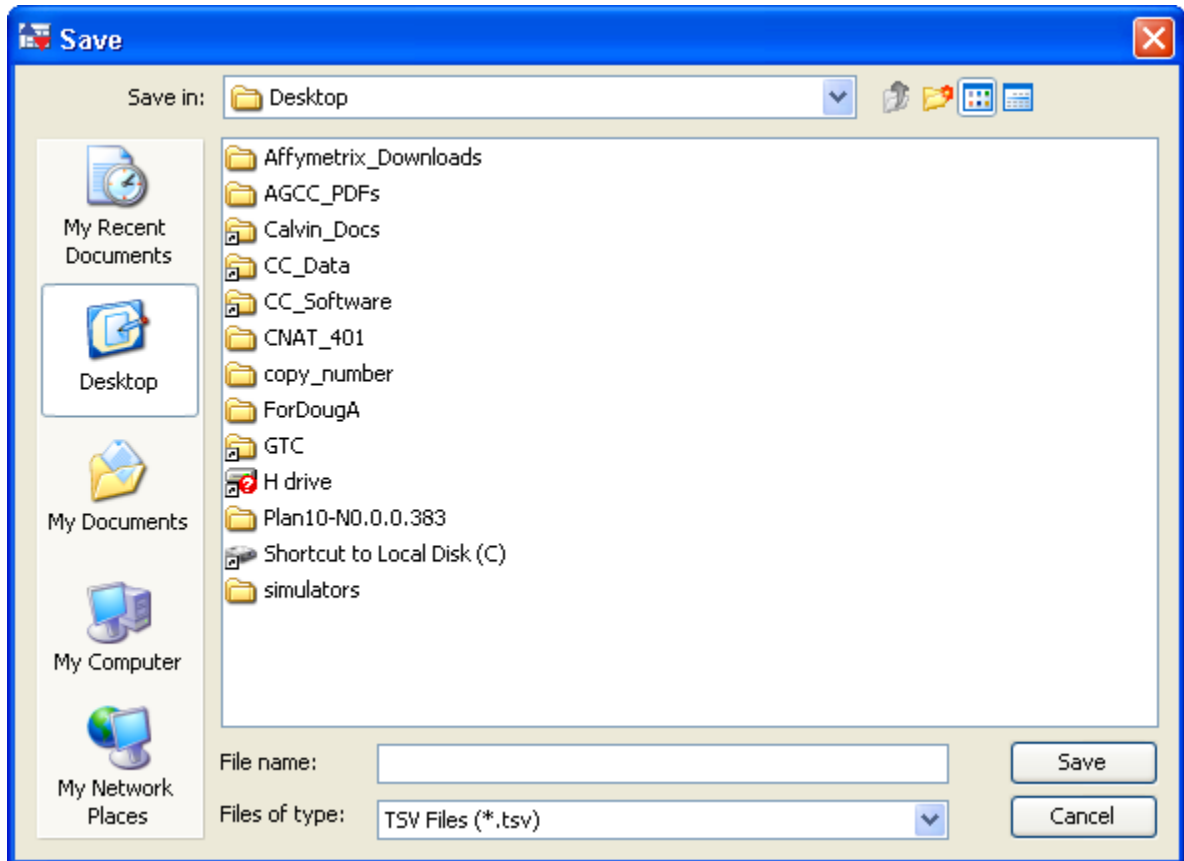


Figure 46. Save dialog box.

2. Select a save location and enter a file name for the TSV file.
3. Click **Save**.

If you do not add a “.tsv” extension to the file name, this will be done the first time the **Save** button is clicked; the **Save** button must then be clicked again to save the .tsv file.

The TSV file is saved in the specified location.

To export to BED format:

1. From the Tools menu, select **Export All to BED...**; or

Click the **BED button**  in the toolbar.

The Save dialog box opens.

2. Select a save location and enter a file name for the BED file.


3. Click **Save**.

If you do not add a “.bed” extension to the file name, this will be done the first time the **Save** button is clicked; the **Save** button must then be clicked again to save the .bed file.

The BED file is saved in the specified location.

To copy data to the clipboard:


1. Select the data you want to copy in the table.
2. From the Tools menu, select **Copy Selections**; or

Click the **Copy Selections button**  in the toolbar.

The selected data is copied to the clipboard and can be pasted into other files.

To search the annotations:

1. From the Tools menu, select **Find Annotations...**; or

Click the **Find Annotations button**  in the toolbar.

The Search dialog box opens (Figure 47).

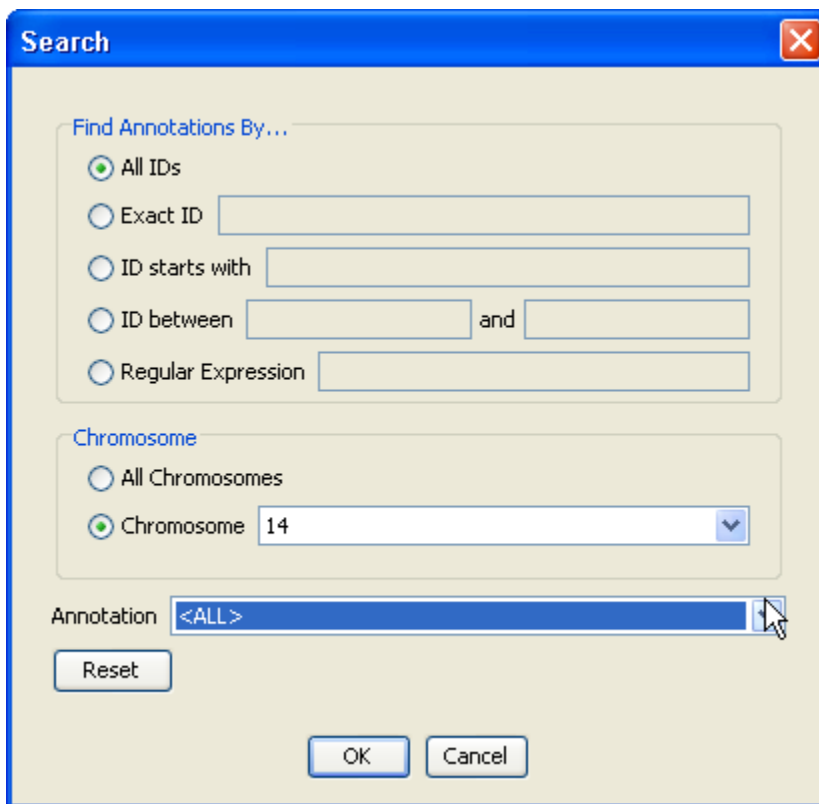


Figure 47. Search dialog box

The Search dialog box allows you to search the annotation files for all or part of the names of an annotation. You can search one or many annotation types for an ID.

2. Select the Find Annotations By option you wish to use and enter a value, if necessary:

All IDs Return all annotations that meet the Chromosome and Annotation type selections.

- Exact ID** Return annotations that match the entered value exactly.
- ID starts with** Return annotations that begin with the entered value.
- ID between** IDs alphabetically between the given values. Example: “hoxa” to “hoxc” would give all homeobox genes with names starting with “hoxa” and “hoxb”, but not “hoxc” or “hoxd”.
- Regular Expression** Allows for wildcard ID searches. A single wild character is represented by a period. Multiple wild characters in a row by period-star: “.*”

3. Limit the results returned by chromosome location:

- All Chromosomes** Return matches on all chromosomes.
- Chromosome number** Return matches only on selected chromosome.

4. Select the annotation file to search from the drop-down list; or
 Select <ALL> to search all annotation files.

Click **Reset** to clear the entries; or

5. Click **OK** to perform the search.

The List View displays a list of annotations that match the search terms (Figure 48).

Chromosome	Min	Max	Strand	ID	Type
chr1	885,829	890,958	+	klhl17	refseq
chr1	1,874,611	1,925,136	-	kiaa1751	refseq
chr1	3,642,407	3,653,746	-	kiaa0495	refseq
chr1	3,721,203	3,763,657	-	kiaa0562	refseq
chr1	6,008,966	6,083,110	+	kcnab2	refseq
chr1	6,008,966	6,083,110	+	kcnab2	refseq
chr1	6,573,371	6,585,516	-	klhl21	refseq
chr1	10,193,417	10,289,402	+	kif1b	refseq
chr1	10,193,417	10,364,241	+	kif1b	refseq
chr1	11,902,711	11,909,067	-	kiaa2013	refseq
chr1	14,797,799	15,264,879	+	kiaa1026	refseq
chr1	15,123,212	15,265,159	+	kiaa1026	refseq
chr1	15,128,882	15,264,879	+	kiaa1026	refseq
chr1	15,145,001	15,264,879	+	kiaa1026	refseq
chr1	18,680,010	18,685,065	+	klhdc7a	refseq
chr1	19,417,171	19,450,633	-	kiaa0090	refseq
chr1	20,863,095	20,917,097	-	kif17	refseq
chr1	22,252,077	22,282,058	-	klhdc1	refseq

846 results

Figure 48. List View

Click in the row for an annotation to display that segment of the genome in the Browser.

Chapter 4: Graphs

Graphs indicate scores or other numeric values as a function of genomic position. Graphs are generally displayed as some form of plot (x,y-plot, bar plot, etc.). The results from GeneChip mapping arrays and from chromosome copy number analysis are generally represented as graphs. Simple graphs represent values for individual genomic positions. Interval graphs represent values for ranges of genomic positions.

There are two main features of the graphical view:

- In all graphical views, the X-axis corresponds to a physical position along the chromosome, and the Y-axis is the value of the metric displayed (except for heatmaps, where the color indicates the value of the metric).
- The graph name (displayed above each graph) indicates the *.cnt or *.cnchp or *.lohchp filename followed by the metric graphed. For example, SK-BR-3_NSP_CN.brImm.CN:Log2Ratio indicates that the Log2Ratio metric of the sample file SK-BR-3_NSP_CN.brImm.CN.CNT is displayed in the first chart.

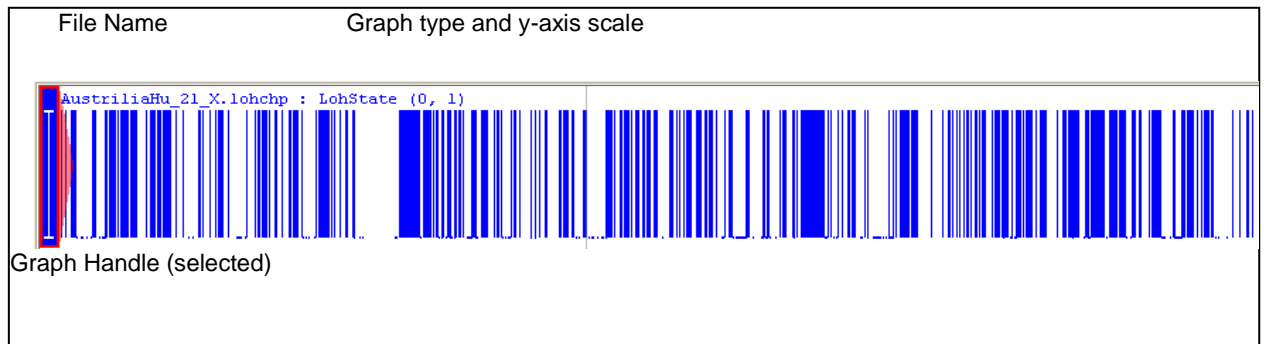


Figure 49. Loss of heterozygosity (LOH) graph with graph handle selected

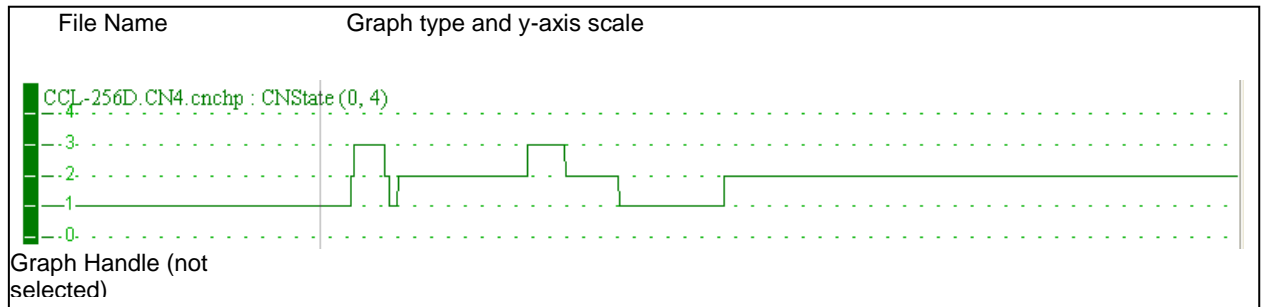


Figure 50. Copy Number graph (zoomed in)

Click on the Graph handle at the left end of the graph bar (Figure 49) to:

- Drag the graph to a new position.
- Select the graph for modification (see Changing graph appearance).

This section explains:

- *How to select graphs types for display* (page 46)
- *Changing graph appearance* (page 50)

Selecting graph types for Display

To select files for display:

- Select or deselect from the Graph Types list on the left side of the screen (Figure 51).

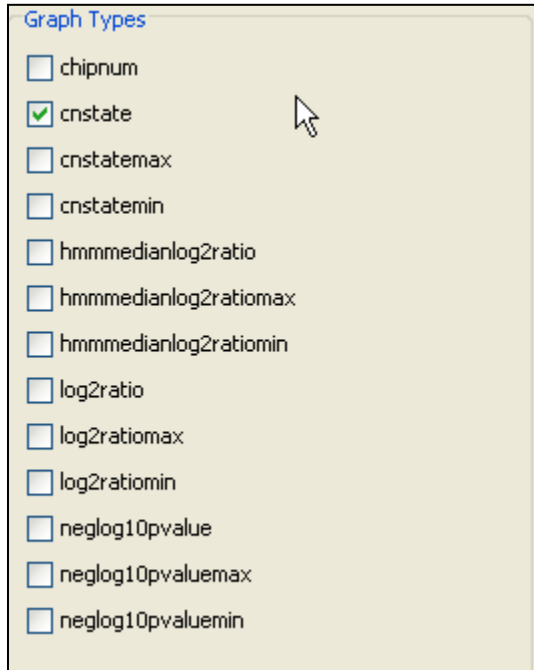


Figure 51. Graph Types list

The types of graphs available differ, depending upon the type of array and the type of analysis performed.

Different types of data can be displayed for:

- *100K/500K array data*
- *SNP 6.0 array data*

Types of Data Displayed for 100K/500K

Copy Number Graphs

- 📌 Values in the chart below that are labeled *paired analysis only* require that the check box next to *Generate Allele-Specific Copy Number (Advanced Options page)* be selected.

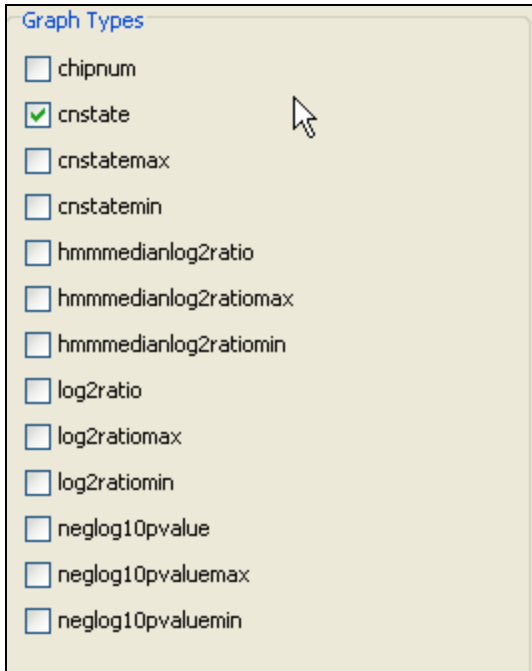


Figure 52. Copy Number Graphs for 100K/500K data

The following types of copy number data (Figure 52) can be displayed:

Data type	Definition
chipnum	1 or 2. Only for the 100K or 500K two-chip SNP assay.
CNState	HMM copy number state.
CNStateMax	HMM copy number state of the allele with the higher signal intensity (<i>paired analysis only</i>).
CNStateMin	HMM copy number state of the allele with the lower signal intensity (<i>paired analysis only</i>).
HmmMedianLog2Ratio	Median Log2Ratio value of all contiguous SNPs in the given HMM copy number state segment.
HmmMedianLog2RatioMax	Median Log2 Ratio value of all the contiguous SNPs in the given HMM copy number state segment of the allele with the higher signal intensity (<i>paired analysis only</i>).
HmmMedianLog2RatioMin	Median Log2 ratio value of all the contiguous SNPs in the given HMM copy number state segment of the allele with the lower signal intensity (<i>paired analysis only</i>).

Data type	Definition
Log2Ratio	Smoothed Log2Ratio value.
Log2RatioMax	Smoothed Log2 ratio value for the allele with the higher signal intensity (<i>paired analysis only</i>).
Log2RatioMin	Smoothed Log2 ratio value for the allele with the lower signal intensity (<i>paired analysis only</i>).
NegLog10PValue	Negative Log10 p-value indicating how different the median Log2Ratio of the HMM state is from the normal state (CN State 2) for that particular sample.
NegLog10PValueMax	Negative Log10 p-value indicating how different the median Log2 ratio of the HMM state of the allele with the higher signal intensity is from the normal state (CN State 2) for that particular sample (<i>paired analysis only</i>).
NegLog10PValueMin	Negative Log10 p-value indicating how different the median Log2 ratio of the HMM state of the allele with the lower signal intensity is from the normal state (CN State 2) for that particular sample (<i>paired analysis only</i>).

LOH Data Displayed

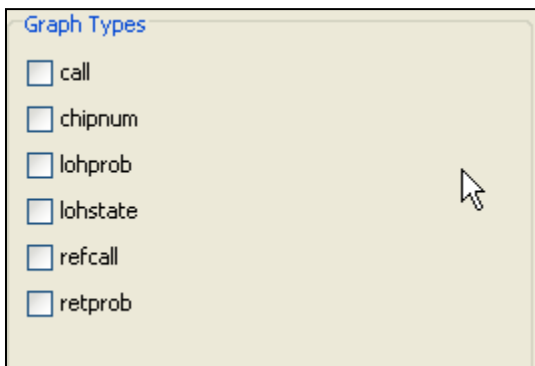


Figure 53. LOH graphs for 100K/500K data

The following types of LOH data (Figure 53) can be displayed:

Data Type	Definition
Call	Genotype call for the tumor/test sample.
chipnum	1 or 2. Only for the 100K or 500K two-chip SNP assay.

Data Type	Definition
LOHProb	Likelihood that a SNP is in LOH state (closer to 1 indicates a stronger likelihood of LOH).
LOHState	1 = LOH and 0 = Retention.
RefCall	Genotype call for the paired reference sample (paired analysis only).
RetProb	Likelihood that an SNP is in Retention state (closer to 1 indicates a strong likelihood of Retention).

Types of data displayed for SNP 6.0

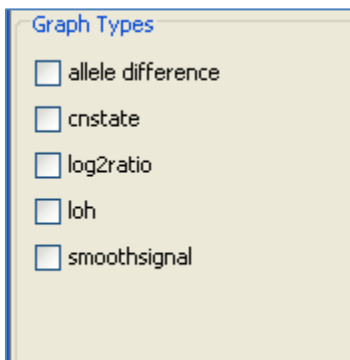


Figure 54. Graphs for SNP 6.0 data

The following types of data (Figure 54) can be displayed for SNP 6.0.

Data type	Definition
AlleleDifference	Difference of A signal and B signal each standardized with respect to their median values in the reference.
CNState	HMM copy number state.
Log2Ratio	Smoothed Log2 ratio value.
LOH	LOH State; 1=LOH, 0=Retention.
smoothsignal	Smoothed log2 ratios or smoothed log2 ratios calibrated to Copy Number and anti-logged (depending on the options setting during copy number/LOH analysis)

Changing graph appearance

You can modify many properties of the graphs, including:

- The height of the graph
- The style used to display the data
- The color of the graph
- The scale of the graph

Settings and adjustments that are specific for graphs can be made using the **Graph Properties** dialog box.

To change the settings for selected graphs:

1. Select the graph(s) to change by doing one of the following:
 - To change a single graph, click the colored bar at the left side of a graph (the graph handle) or the track label to select it.
 - Shift-click to select additional graphs.
2. Right-click on the graph handle and choose **Properties...** from the menu (Figure 55).

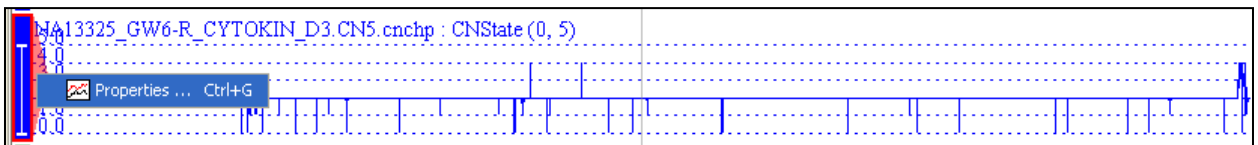


Figure 55. Selecting a graph

The Graph Properties dialog box opens (Figure 56).

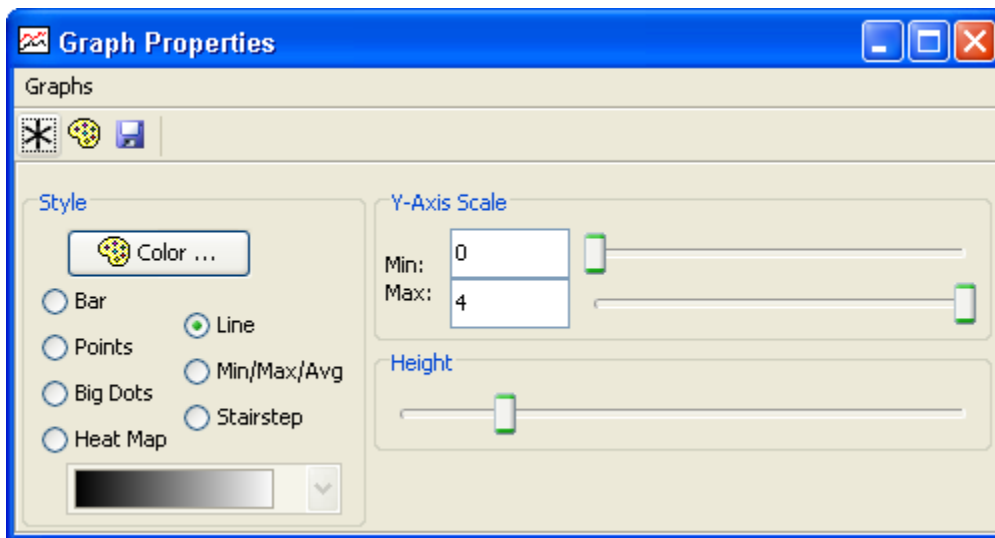


Figure 56. Graph Properties dialog box

 You can also open the Graph Properties dialog box by clicking the Properties button in the toolbar.

3. Make changes to the graph settings by typing in new values or by operating sliders in the Graph Adjuster panel. For details, see:
 - Change the height of a graph

- Change graph style
- Change graph color
- Change scale

Any changes you make to the values in the **Graph Properties dialog box** will apply to all currently selected graphs.

To change the settings for all graphs:

1. Click the Properties button in the Browser toolbar.
The **Graph Properties** dialog box opens.
2. From the Graphs menu, select **Select All Graphs**; or
Click the **Select All Graphs** button in the dialog box toolbar.

Change the height of a graph

There are two ways to change the vertical height of a graph:

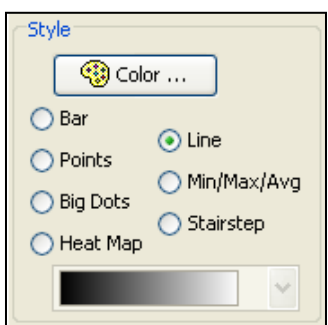
- Select the graph(s) you want to change by (shift+)clicking on their handles, press the Graph Properties icon, then drag the Height slide bar in the Graph Adjuster tab.
- Use the Stretch Slider on the left side of the Display to stretch all Graphs and Annotations simultaneously in the vertical direction

Change graph style

Graphs can be shown in various representational styles. The type of graph that is most appropriate depends on the type of question being asked about the data. For example, when comparing trends and patterns, it is very useful to use the line graph display method. The number of expression intervals being shown also can affect the graph display choice. The user is encouraged to experiment with the different display types to find out which method works best for specific purposes and at specific zoom in magnifications.

To change the graph style:

1. SHIFT + Click the colored bar at the left end of the graph.
2. Click the **Graph Properties** tab.
3. In the **Style** section choose one of the options.



The following graph styles are available in GTC Browser:

- **Bar** (Figure 57)– Individual values are shown as vertical bars that are one base wide for position graphs and of variable width for interval graphs.

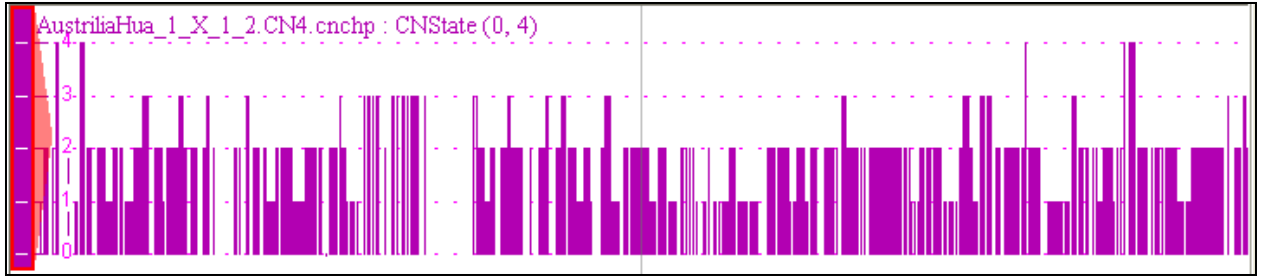


Figure 57. Bar

- **Line** (Figure 58) – Subsequent values are linked with a line. Even if the input file was not sorted, the values will be connected in order along the genomic coordinate axis.

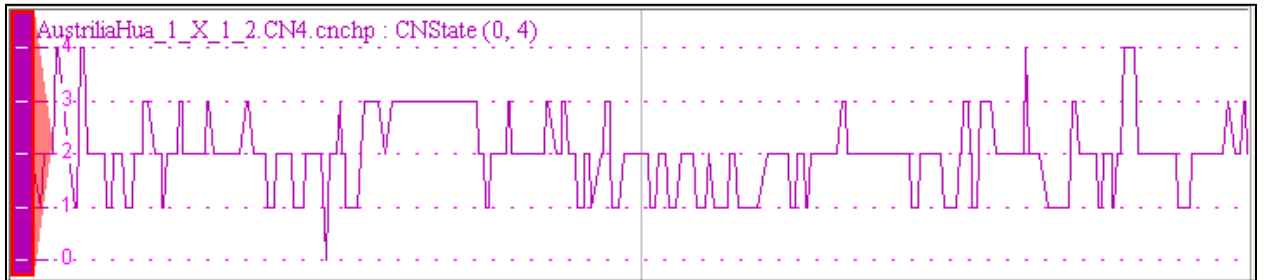


Figure 58. Line

- **Points** (Figure 59) – Shows a single dot for each data value. For interval graphs, horizontal lines will be connected to the start and end points.



Figure 59. Points

- **Big dots** (Figure 60) – Shows a single big dot for each data value.



Figure 60. Big dots

- **Min/Max/Avg** (Figure 61) – This style is especially useful for showing very densely populated graphs with data points for large numbers of positions.

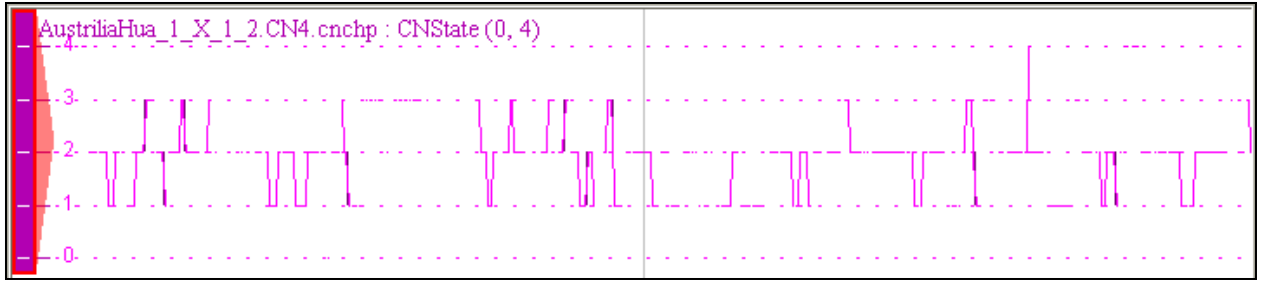


Figure 61. Min/Max/Average

When GTC Browser is zoomed all the way in, the display is equivalent to the **Line** style. When zooming out, GTC Browser starts to summarize values. When the scale of the display reaches the point where individual x-values are associated with multiple score values, GTC Browser picks the maximum and minimum values and draws a vertical bar between them. In addition, GTC Browser draws lines through the average of all the data points represented at each x value.

- **Stairstep** (Figure 62) – Similar to the bar graph style, except that bar widths along the horizontal axis are stair-stepped.

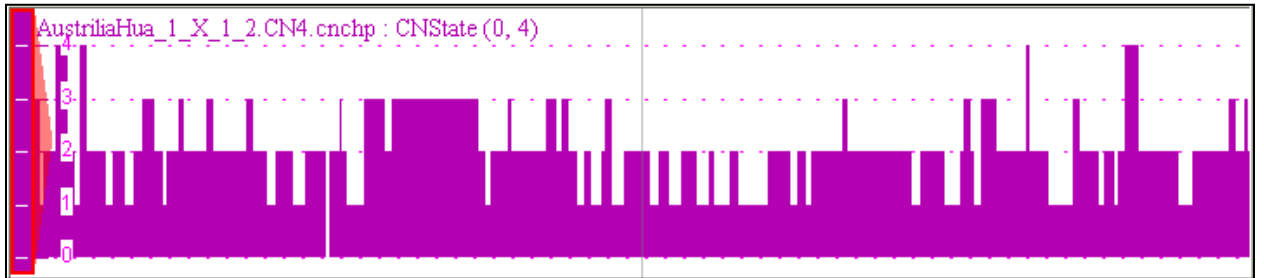


Figure 62. Stairstep

For example, if position 100 has a value of 50 and position 200 has a value of 75 and there are no values in between, then GTC BROWSER will draw a bar of height 50 that starts at position 100 and stops at position 200. Then, at position 200, GTC BROWSER will draw a new bar of height 75 that terminates at the next location with a value.

This style is particularly useful for viewing .egr and .sgr files, ESTs, or other high-density data.

- **Heat map** (Figure 63) – Instead of showing relative intensity via the height of the line at each pixel or coordinate as in most other graph styles, a heat map shows expression levels via color or brightness of the line at each pixel or coordinate. This graph style is useful if you want areas of greater values to jump out at you. If a graph does not render or is hard to see, adjust the visible bounds of the graph until features are readily visible. Several heat map color schemes are available to choose from.

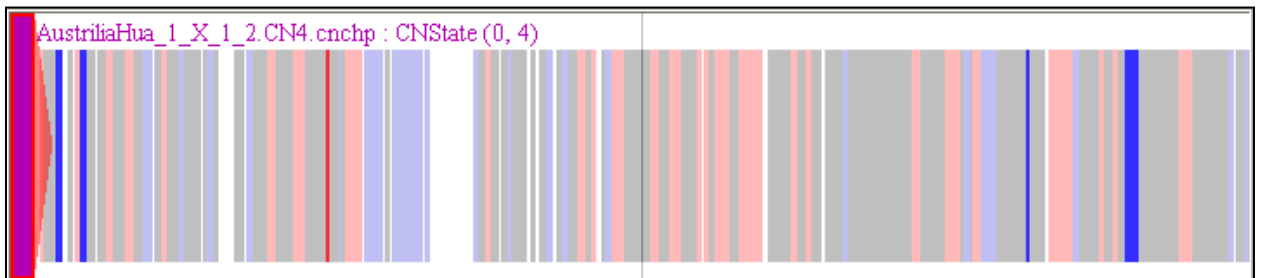


Figure 63. Heat map

Change graph color

To change graph color:

- Click the graph handle on the left end of the graph to select the graph.
- Click the Graph Properties button. In the Style section, click the Color button.
- Choose the desired color, and then click OK.

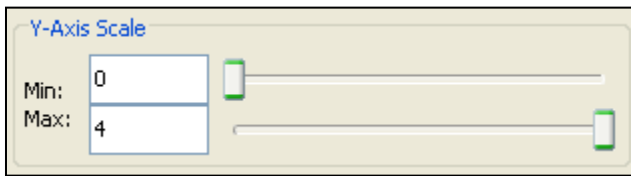
For graphs in the heat map style, the chosen color will only be used on graph labels and handles. The data colors come from the chosen heat map.

Change scale

Changing the visible bounds involves changing the scale of the graph by setting the maximum and minimum values to be displayed.

To set these visible bounds, use the **Y-axis scale** section of the **Graph Properties** dialog:

- Select the graph(s) to change: Click the graph handle at the left end of each graph to change. (Shift-click to select more than one.)
- Click the **Graph Properties** button.
- Adjust the visible graph bounds:



To set specific minimum and maximum values, use the sliders, or type in values to the boxes, these values will be applied to each selected graph. You are free to set maximum and minimum values that cover a range smaller or larger than the actual range of your data.

Chapter 5: Exporting Data

GTC Copy Number Browser can export:

- *Images for use in other documentation*
- *Data from graphs*

Other data export functions are described in the sections of the different information boxes (Selection Details, List View, Segment Report).

Exporting Images

Graph images can be exported to share with other researchers in the following formats:

- PDF
- PNG: can be inserted in other documentation.

 You can export graphics from both Karyoview and Chromosome view.

To export as a PNG graphic:

1. Set up the display the way you want it.
2. From the Graphs menu, select **Export PNG**.

The Save dialog box opens (Figure 64).

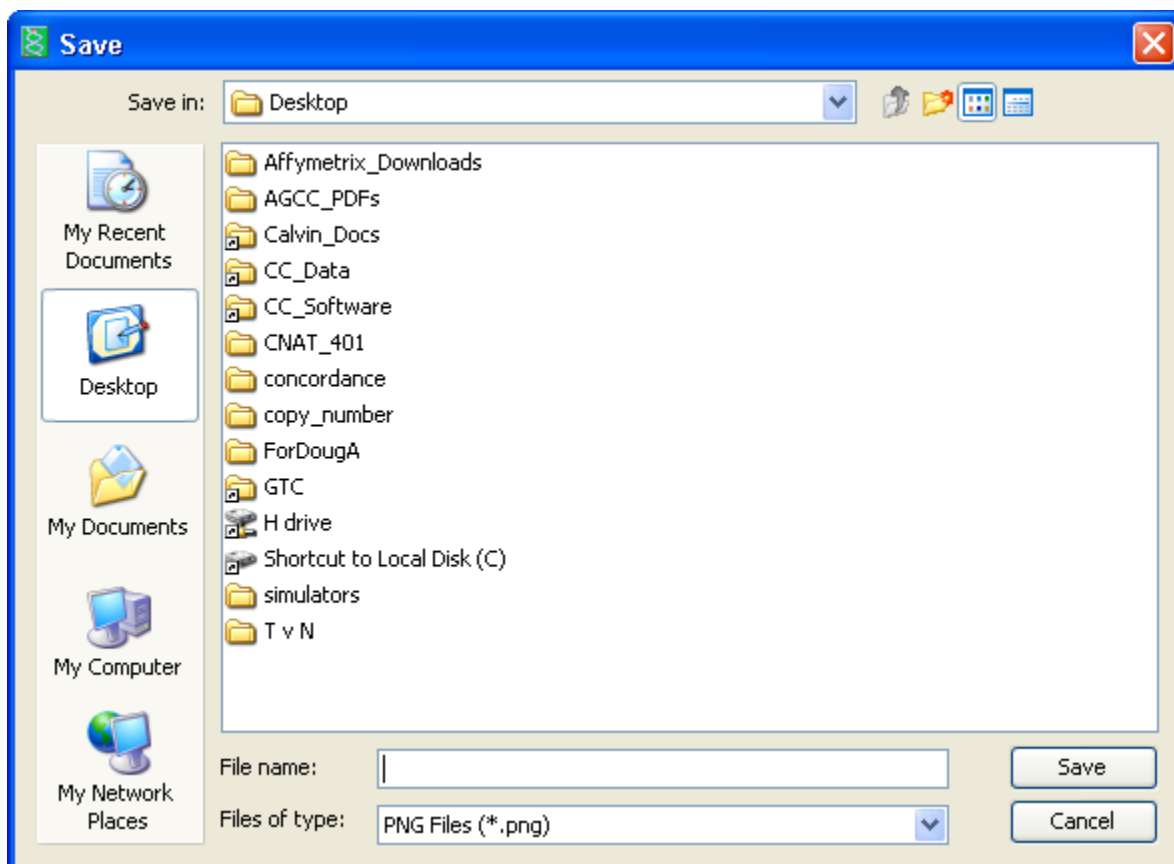


Figure 64. Save dialog box for PNG file

3. Use the dialog box controls to select a location and enter a file name.
4. Click **Save**.

If you do not add a “.png” extension to the file name, this will be done the first time the **Save** button is clicked; the **Save** button must then be clicked again to save the .png file.

A graphic in PNG format is saved.

You can send the graphic to another researcher or use it in a document.

To export as a PDF file:

1. Set up the display the way you want it.
2. From the View menu, select **Export PDF**.

The Save dialog box opens.

3. Use the dialog box controls to select a location and enter a file name.
4. Click **Save**.

If you do not add a “.pdf” extension to the file name, this will be done the first time the **Save** button is clicked; the **Save** button must then be clicked again to save the .pdf file.

A PDF file of the graphic saved.

You can send the graphic to another researcher.

Exporting Graph Data

 **You can export graph data only from the Chromosome view.**

You can export graph data in wiggle (WIG) format for viewing in other genome browsers.

 **The wiggle (WIG) format allows display of continuous-valued data in a track format. For more information, see www.genome.ucsc.edu.**

To export graph data:

1. From the Graph menu, select Export Displayed Graphs.
The Save dialog box opens.
2. Select the File name and location where you want the file to be saved.
3. Save the file.

If you do not add a “.wig” extension to the file name, this will be done the first time the Save button is clicked; the Save button must then be clicked again to save the .wig file.