## Celera Genome Browser

## User Guide

June 2007 Open Source Release 5.0 © 2002 Celera Genomics, All rights reserved.

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## ABOUT THIS GUIDE

Welcome to Celera's Genome Browser. This guide describes the Genome Browser's features and functions, and provides step-by-step instructions for using the product.

As new features are added to the Genome Browser, we will update the user guide accordingly. This guide is available as an Acrobat PDF from the Genome Browser help menu. Select HelplUser Manual from the Main Menu.

This guide was developed for new Genome Browser users.

Product Release Notes

The Genome Browser release notes describe new features, known issues with the product, and additional helpful information. To access the release notes, select HelplRelease Notes from the Genome Browser Main Menu.

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## CONVENTIONS USED IN THIS GUIDE

This guide uses the following typefaces and symbols for presentation purposes:

Bold Text Indicates text that you should type. Brackets surround user-specific text. For example, [search string] means that you should type the text for which to search. You do not type the brackets. For short text strings, we also use quotation marks to surround the text you type. You do not type the quotation marks.

Italic text Indicates words that require special emphasis.

NOTE Highlights important information or warnings.

TIP Describes shortcuts.

# FINDING INFORMATION IN THIS GUIDE

The following table describes where to find information in this guide:

Chapter <u>1.0</u> Genome Browser Overview	Describes the features and functions of the Genome Browser. Also describes the system requirements.
Chapter 2.0 Getting Started	Explains how to access the Genome Browser and describes the Main Window. Explains how to set preferences.
Chapter 3.0 Genome Browser Data Overview	Describes the Celera data that appears in the Genome Browser. Also describes the Celera Exchange (XML) documents.
Chapter <u>4.0</u> Accessing and Viewing Data	Explains how to use the Genome Browser to access and view genomic information.
Chapter <u>5.0</u> Creating and Modifying Annotation Data	Explains how to use the Genome Browser to create, modify, and export annotation data.
Appendix A Exchange File Format	Describes the Celera Exchange File format that you use to create XML files for importing into the Genome Browser. Includes the Document Type Definition.
Appendix B Troubleshooting	Explains how to troubleshoot problems you may experience with the Genome Browser.

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## TECHNICAL SUPPORT

Currently, there is no technical support for the open source release.

Genome Browser

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### INTRODUCTION

Genome Browser is a visualization tool that allows you to:

- graphically view Celera genome sequence information and annotation data
- view multiple genome versions at the same time
- overlay and view your mapped feature data with Celera data
- create and save annotation

The Genome Browser is designed to support a fully functional 3-tiered application consisting of:

- an interactive GUI client front-end
- an application server in the middle tier
- a relational database back-end

Currently, the open source release only supports the stand-alone single-tier client functionality Future versions may provide a standard data server or access to industry standard data protocols. This would allow the user to open the Genome Browser client and then connect it to a variety of different data servers, as well as open local feature files and temporary workspace files for viewing and annotating data on top of the server assembly data.

NOTE: All file import and export functionality takes place on the client side of the application, whether or not the Genome Browser is connected to an application server.

The current, stand-alone release allows you to load genomic data directly into the client using the Celera version of the GAME XML file format known as the Celera Exchange File Format.

XML Features

The Celera Exchange (XML) File Format allows you to import and export data from the Genome Browser in a flexible manner that supports a variety of different workflows and data sources. Using the Celera Exchange File Format with the Genome Browser, you can:

- Specify assembly files
- Import feature files
- Annotate and save workspace files

The following sections provide more detail about these features.

Specify Assembly Files

See Appendix A for the detailed Celera
Exchange File Format Specification and
Document Type Definition. Also, see page
3-4 for more details about the XML
assembly files.

For 3-tier operation, assembly data is stored in a database, which you access via data server.

However, the open-source release only supports stand-alone operation, so assembly Sequence information must be specified in an assembly file, usually in the form of a FASTA file.

See page <u>3-4</u> for more details about XML feature files.

Import Feature Files

You can import either precomputed or human-annotated features as multiple feature files directly into the Genome Browser client as long as they are specified in relation to an assembly Genomic Axis. Since the feature files do not contain any Genomic Axes themselves – only references to them – you may partition feature data among files in any convenient manner. Multiple feature files may contain data on the same Genomic Axis, or a single feature file may contain data on multiple Genomic Axes.

See page <u>3-5</u> for more details about XML workspace files.

Annotate and Save Workspace Files

The Genome Browser performs all interactive annotation operations in local memory in a data structure called the workspace. You can export the annotation data in the workspace (modified and newly created genes, transcripts, exons, start codons, and evidence relationships) to a single workspace (.gbw) file, which you can reload at a later time to continue annotating.

If you wish to commit your annotation data to your database, you can then feed the workspace file into a promotion utility (not provided by the open-source Genome Browser release).

## SYSTEM REQUIREMENTS

Before you access the program, ensure your machine has the following installed:

- Java Runtime Environment (JRE) 1.3.0\_
- At least 256 MB RAM
- Adequate virtual memory settings

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# ACCESSING THE GENOME BROWSER

To access the Genome Browser:

- 1 Make sure that a Java virtual machine is installed on your computer
- 2 From your desktop, double-click the Genome Browser icon.

The Genome Browser Main Window appears:

Main Menu

Outline View

Property Inspector View

Genome Axis Annotation View

Left Frame

Right Frame

Memory Meter

NOTE: The Genome Browser enables only those menu and sub-menu items that are valid in a given context. If an item is greyed out, it is unavailable at that time.

When you access the Genome Browser, the window is the same size and layout and in the same location as when you last closed the application.

#### About the Genome Browser Main Window

The following table describes the contents of the Genome Browser Main Window and how to navigate within the window.

#### Genome Browser Main Window

Main Menu

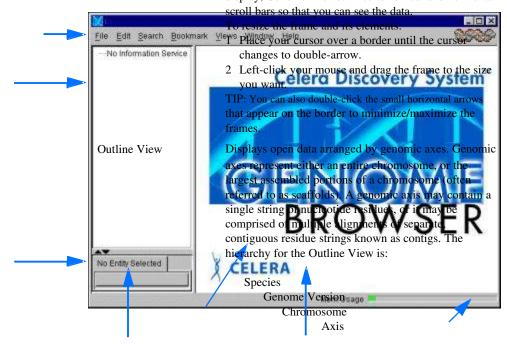
Displays the menu options that are available to you. The menu options vary, depending on the data you have open. This guide describes the available options in greater detail when explaining how to perform tasks.



Contains two elements:

- Outline View
- Property Inspector View

If the Outline View contains more data than it can display, Genome Browser inserts vertical and horizontal



- Click the + sign to expand a folder or folder name.
- Click the sign to collapse a folder or folder name.

NOTE: When you first open the Genome Browser, the Outline View displays "No Information Service."

Property Inspector View	Displays the properties associated with the entity currently highlighted in the outline view or in the annotation view.  NOTE: When you first open the Genome Browser, or if you do not have an object selected, the Property Inspector View displays "No Entity Selected."
Right Frame	Contains the Genomic Axis Annotation View.  If you enable the SubView view, it will appear in the bottom portion of the right frame area.  You can resize this frame, too.
Memory Usage Meter	Roughly indicates the amount of available memory while you are using the application. The meter color changes to a yellow line if 70% of available memory is used and a red line if 90% of available memory is used. Depending upon what the application is doing, this meter may fluctuate.  To find out exactly how much memory the Genome Browser client is using:  1 Double-click the memory meter.  The Memory Usage dialog box appears, which

Total Memory — the total memory that the Genome Browser requires, up to the maximum it is

Free Memory — how much memory remains available for the Genome Browser client to use.

Used Memory — how much memory the Genome

Browser is consuming at the moment.

indicates:

allowed to consume.

Genome Browser Main Window (cont'd)

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### Compacting Memory

Double-click the Memory Usage Meter in the lower right of the Genome Browser Main Window.

The Memory Usage dialog box appears:

Click Yes.

Status Messages from the Genome Browser

only if you have the Genome Browser open and are connected to a Celera database.

NOTE: You will receive this message If the Genome Browser has downtime scheduled, Celera will broadcast a Status Message with the appropriate information across the server to the Genome Browser client.

## SETTING PREFERENCES

Before you begin using the Genome Browser, you should review and set or change, if necessary, the Genome Browser Preferences.

Setting Viewing Preferences

The following sections explain how to:

- Create a preferences file and specify the location for group preference files
- Modify the Genomic Axis Annotation View Settings

Creating a Preferences File

You may save changes you make to the View settings in a file to use for yourself or to share with a group of users.

- 1 From the Main Menu, select Edit|Preferences|Genomic Axis Annotation View.
- 2 Click the View Preference File tab.

3 Click New File.

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The Create New View Setting File dialog box appears.

- 4 Type a name in the text box.
- 5 Click OK.

The new preference becomes the default.

Deleting a Preferences File

To remove a preferences file:

- 1 From the Main Menu, select EditlPreferenceslGenomic Axis Annotation View.
- 2 Click the View Preference File tab.
  The Preferences: Genomic Axis Annotation View dialog box appears.
- 3 Click Delete File.
- 4 In the Delete File dialog box, select the file you want to delete and click OK.

The Deleting file dialog box appears.

5 Click Yes to delete the file or No to retain it.

To remove any changes (e.g., color choices) from the current session, click Reload.

To reset to default, click Reset to Default.

Specifying a User- or Site-Specific Preference File

You may wish to specify a preference file other than the default, or your site may wish to create and his Asia Augustice Year settings are the same across users. If that is the case, you need to tell the Genome Browser to use the desired preference file and specify the location for the new preferences file. No Description. 1 From the Main Menu, select EditlPreferenceslGenomic Axis Annotation View. 2 Click the View Preference File tabag File 3 Click Change Active Settings File. Delete File 4 Browse to locate the directory's folder that contains your XML files, and Reset To Default select the folder. OK Cancel Apply

5 Click Select File.

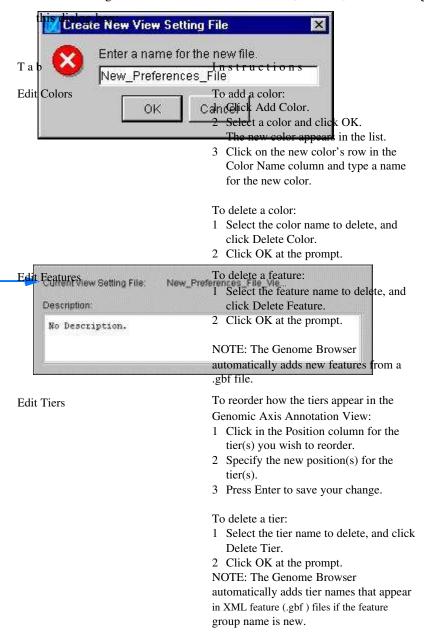
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Modifying Genomic Axis Annotation View Settings

The Genome Browser provides a variety of color schemes to help you view genomic data. You modify default settings in the Preferences: Genomic Axis Annotation View dialog box.

1 From the Main Menu, select EditlPreferenceslGenomic Axis Annotation View.

The Preferences: Genomic Axis Annotation View dialog box appears.



The following table describes how to edit colors, features, and tiers using

#### Specifying SubView Preferences

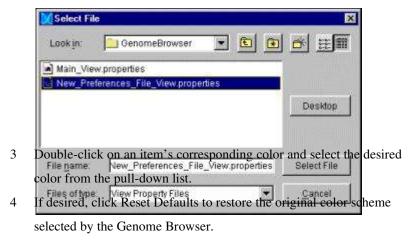
The following sections explain how to:

- Edit Alignment View Settings
- Edit Consensus Sequence View Settings
- Edit Transcript Translation View Settings

Editing Alignment View Settings

You can change the Alignment View settings to accommodate your color preferences.

- 1 From the Main Menu, select EditlPreferences|SubViews.
- 2 Click the Edit Query Alignment View Settings tab:

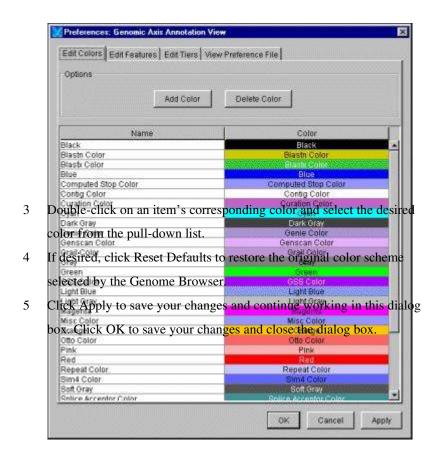


5 Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the dialog box.

#### Editing Consensus Sequence View Settings

You can change the Consensus Sequence View settings to accomodate your color preferences.

- 1 From the Main Menu, select EditlPreferences|SubViews.
- 2 Click Edit Consensus Sequence View Settings.



Editing Transcript Translation View Settings

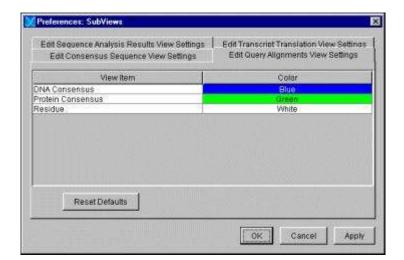
You can change the Transcript Translation View settings to accommodate your color preferences.

- 1 From the Main Menu, click EditlPreferences|SubViews.
- 2 Click Edit Transcript Translation View Settings:

- 3 Double-click on an item's corresponding color and select the desired color from the pull-down list.
- 4 If desired, click Reset Defaults to restore the original color scheme selected by the Genome Browser.
- 5 Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the dialog box.

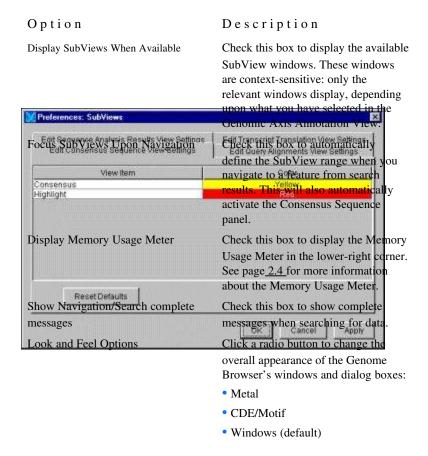
### Specifying System Preferences

- 1 From the Main Menu, select EditlPreferenceslSystem .
- 2 Click the Applications Settings tab:



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3 Select the desired options. Use the following table to guide you through your choices.



Specifying the Data Source Settings

You must specify the data source settings before you can access any data in the Genome Browser. Depending on the data you will access, you may do one or more of the following:

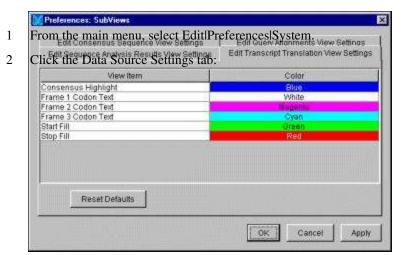
- Set login information
- Specify XML directories
- Specify XML Service URLs

### Setting Login Information

Celera database, you do not need to login.

NOTE: If you are not connecting to a You will need to set your login information before you may connect to a database. You do this from the Preferences: System dialog box.

NOTE: When you select FilelOpen Genome, the Preferences dialog box appears automatically if you have never set your login information.



- In the User Name text box, type your database user name.
- Type your database Password in the text box.
- If desired, select the "Save Login Information" box. If you do not check this box, you will need to login each time you connect to a database.

6 Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the dialog box.

Adding XML Directories

Before you may open an XML assembly or load an XML feature file's data, you need to tell the Genome Browser where to look for the files.

- 1 From the Main Menu, select EditlPreferenceslSystem.
- 2 Click the Data Source Settings tab.
- Click Add to Current Directories.The Select Directory dialog box appears:

- 4 Browse to locate the directory's folder that contains your XML files, and select the folder.
- 5 Click Select Directory.
- 6 Repeat steps 3 through 5 for each directory that you wish to add.
- 7 If desired, specify a validation option.
- 8 Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the dialog box.

NOTE: To delete an XML directory, select the directory to delete and click Remove Selected Directory.

Specifying XML Service URLs

NOTE: For more information about the Genome Browser XML Service, refer to the Celera Programmer Reference Guide for the Genome Browser.

NOTE: To delete a URL, select the URL you want to delete in the Current URLs box, and click Remove Selected URL.

If your site takes advantage of the Genome Browser's XML Service capability, you should specify the URLs in the Preferences dialog box.

- 1 From the Main Menu, select EditlPreferenceslSystem .
- 2 Click the Data Source Settings tab.
- 3 Type the URL provided by your site programmer in the New URL text box, and click Add to Current URLs.
- Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the dialog box.

Celera provides splice profiles for Human, Mouse and Drosophila data. You can override these default splice profiles and specify different "Acceptor," "Donor," and "Neither" splice profile files you have stored on your computer.

Setting Splice Profiles

2

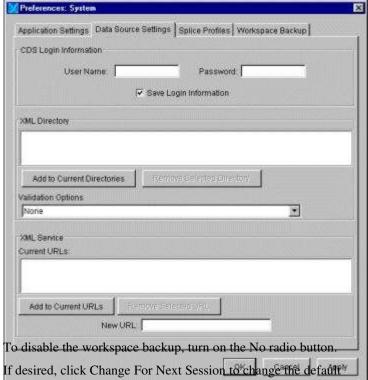
If desired, check the Override Default Splice Profiles box. This will activate the Choose buttons that allow you to upload Donor, Acceptor and Neither splice files.

3 Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the System Preferences dialog box.

### Setting Workspace Backup Options

The Genome Browser is set to periodically backup your Workspace. If you like, you can disable this feature.

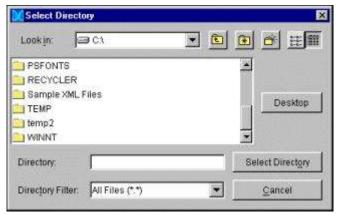
- From the Main Menu, select EditlPreferenceslSystem .
- Click the Workspace Backup tab..



- 3
- 4
  - directory and/or file name for the workspace backup file..
- Click Apply to save your changes and continue working in this dialog box. Click OK to save your changes and close the dialog box.

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# GENOME BROWSER DATA CLASSES

The Genome Browser supports these major data classes:

See <u>page 3.6 f</u>or a diagram that illustrates the Genome Browser data object flow.

- Assembly
- Features, comprised of precomputes and annotations

### Assembly

Assembly is the raw nucleotide residue data, stored as sequences of A, T, C and G, which have been ordered together to form single base-pair coordinate systems known as Genomic Axes. Genomic Axes represent either an entire chromosome, or the largest assembled portions of a chromosome (often referred to as scaffolds). A Genomic Axis may also represent a set of scaffolds that are ordered and oriented relative to one another. A Genomic Axis may contain either a single string of nucleotide residues, or it may be comprised of multiple, separate, contiguous-residue strings known as contigs, ordered and oriented within a scaffold.

#### Fe a t u re s

Features are comprised of precomputes and annotations.

### Precomputes

Precomputes are features, aligned to a sequence, that have been determined by some algorithmic process (e.g., BLAST). Precomputes may be either:

- simple form —single ranges along a genomic axis, with descriptive information (e.g., HSP)
- compound form adjacent simple features that are grouped by the analysis program (e.g., BlastHits)

#### Annotations

Annotations are features, aligned to a sequence, that have been created by a human or automated curation process. The Genome Browser supports three annotation feature types:

- Exon a single range on a nucleotide sequence, together with
  descriptive information and zero or more evidence relationships.
   Evidence relationships are references to precompute objects used as
  evidence of the existence of an exon.
- Transcript a sequence of exons separated by intron regions, translated into a protein sequence. A transcript may also have a Start\_Codon or a Translation\_Start\_Position (if the Start\_Codon can not be determined), together with an optional Stop\_Codon to indicate the translation frame of the transcript.
- Gene one or more transcripts that are associated with a single gene.

# CELERA EXCHANGE (XML) DOCUMENTS

The Genome Browser supports the following types of XML exchange documents:

NOTE: You may open only Celerasigned XML assembly files.

- Assembly
- Feature
- Workspace

These files all use the same data file format, but they contain different kinds of data. They are distinguished by using different file name extensions to indicate the type of data they contain.

The following sections provide more detail.

XML Assembly Files

Assembly files (.gba extension) are the only files that can contain genomic assembly data. An assembly file may contain features as well, but for large data sets it is more practical to break the features into multiple feature (.gbf) files that you can load on an as-needed basis. Celera-signed assembly files do not contain features.

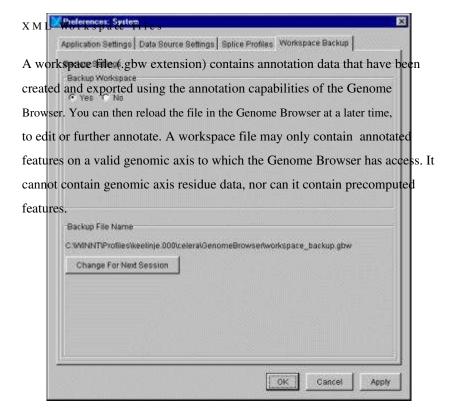
XML Feature Files

NOTE: Although there is no constraint on how to organize features in feature files, your computer may experience memory problems if a feature file is too large.

Feature files (.gbf extension) contain either precomputed or humanannotated features calculated along a genomic axis. However, they are not allowed to contain assembly (i.e., genomic axis residue) data. There is no limit to how many or what type of features can be contained in a feature file, as long as they each reference a valid genomic axis to which the Genome Browser has access. You can open any number of feature files in the Genome Browser at one time, and a single feature file may contain features on many different genomic axes.

Human-curated features contained in the feature files show up in the "Promoted" tier of the browser.

NOTE: You may load only one workspace file in to the Genome Browser at a time.



The following diagram illustrates the Genome Browser data object flow.

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## ACCESSING THE DATA

To access data in the Genome Browser, you may either:

- Search for a specific ID and navigate to its region on the genome
- Open one or more genome versions and drill down to a region on a genome

Searching for a Specific ID

1 From the Main Menu, select Search|Features...

The Search Known Features dialog box appears:

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NOTE: If you are not already connected to an information service, the first query type is the only available option. 2 Select a query type:

- Search the available Genome Version(s) Searches all genome versions to which you have access, whether you are connected to a genome version or not.
- Search the loaded Genome Version(s) Searches only the genome versions to which you are connected. This type is unavailable if you do not have a genome version loaded.
- Search the current Genome Version Searches only the genome version you have selected in the Outline View. This type is unavailable if you do not have a folder selected in the Outline View.

NOTE: The last ID type used will be the topmost available option in the list. 3 Select one of the following ID types from the pull-down list:

- Transcript (Celera Accession)
- Gene (Celera Accession)
- Genomic Axis Name
- Protein (Celera Accession)
- Feature ID
- Unknown ID
- Chromosome
- Celera Variant (SNP)
- External ID (gi or accession)
- STS Name
- BAC accession or clone name
- Subject Sequence ID
- Gene Ontology
- Gene Family
- Regulatory Region Accession
- Conserved Segment Accession
- Gene Index Accession

NOTE: Open New Browser is unchecked by default. Check this box to display the data in a separate browser window. 4 Type or paste the ID for which to search in the blank text box and click OK. With the exception of GA names, this search is not case-sensitive.

5 Select the desired result and click Navigate to view the data, or Bookmark to mark the results for later use.

Searching for Celera Discovery System Sequence Data

This section describes how to search for a GA (Genomic Axis) number in the Genome Browser:

- From the Main Menu, select Search|Features...
   The Search Known Features dialog box appears.
- 2 Select a query type.
- 3 Select Genomic Axis Name from the pull-down list, and type or paste the desired GA number (e.g., GA\_x9V1BB6) in the blank text box.
  TIP: Right-click to paste the GA number in the text box.

See <u>page 4.4</u> for more information about the available query types.

4 Click OK.

If more than one result appears, select the desired GA ID.

5 Click Navigate.

The information for this genomic axis appears in the Property Inspector View.

From the Main Menu, select ViewslGenomic Axis Annotation

View to see the data in the Genomic Axis Annotation View or doubleclick on the GA number in the Outline View.

Genome Browser

Searching for an XML Feature ID

You can also search for a Feature ID that appears in a XML Feature file (.gbf).

From the Main Menu, select Search|Features...
 The Search Known Features dialog box appears.

See <u>page 4.4</u> for more information about the query types.

- 2 Select a query type.
- 3 Select Feature ID from the pull-down list and type (or paste) the unique Feature ID in the blank text box.
- 4 Click OK.

The Choose Path dialog box appears with the feature selected or a list of feature IDs. If more than one appears, select the desired ID.

5 Click Navigate.
The information for this feature appears in the Property Inspector View.

6 From the Main Menu, select ViewslGenomic Axis Annotation to see the data in the Genomic Axis Annotation View.

Opening a Genome Version

You may open one or more genome versions to drill down to the assembly and feature data that you want to view. You can either:

- Connect to a Celera database to view an assembly and its feature data
- Open an XML assembly (.gba) file

To open a genome version:

specify the Data Source Settings from
the Preferences dialog box. Refer to

"Setting Preferences" on page 2.6 for
list of

From the Main Menu, select FilelOpen Genome Version.

The Selection Species/Assembly dialog box appears, displaying a list of available genome versions.

instructions.

NOTE: You may open only Celera-

signed XML assembly files. Also, before you can open a .gba file, you must

2 Select the desired assembly from displayed list, and click OK, or doubleclick on the selected assembly.

TIP: To open more than one genome version at a time, use Ctrl-click to select multiple genome versions and then click OK.

The Outline View now displays the unexpanded species folder. For example:

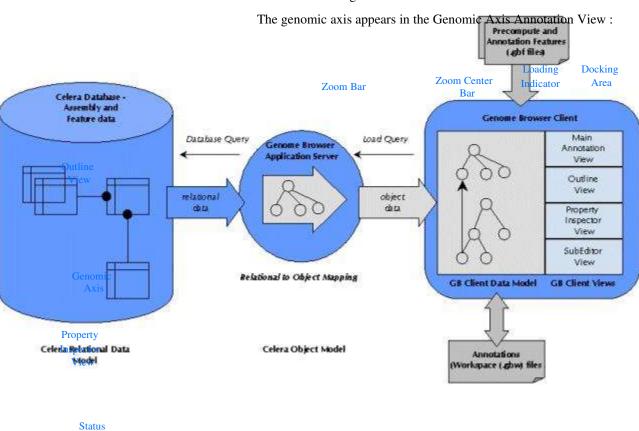
### Drilling Down to a Genomic Axis

- 1 Connect to the Celera database or open an assembly file.
- 2 Expand the species folder and the sub-folders to open the folder that contains desired axis.

TIP: Right-click your mouse on the genomic axis, chromosome, or scaffold position on the chromosome; and select a sort option for viewing by size:

3 Select the genomic axis' folder.

The axis information appears in the Property Inspector View.



4 Double-click the genomic axis' folder.

Scroll Bar

SubView

Genomic Axis Annotation View

- To center around a specific region before you zoom in or out, position the vertical Zoom Center Bar in the desired region.
- To zoom out or in, slide the zoom bar up or down.
- To pan across the genomic axis, slide the horizontal scroll bar left or right.

Bar

Opening an XML Workspace (.gbw) File

You can open your own workspace file, as long as this file was created using the Celera Exchange File Format. The Genome Browser displays the data with the Celera data, allowing you to create or modify annotations on your data.

See Appendix A for the detailed Exchange File Format Specification and Document Type Definition.

You may work with only one XML workspace file at a time.

- 1 Ensure that the file was created using the Celera Exchange File format.
- 2 From the Main Menu, select FilelOpen Workspace File.
  If you have never opened an XML workspace file, the Open dialog box appears automatically. Browse to locate the desired file, and click Open.

TIP: If you have opened one or more XML workspace files, the submenu allows you to access the Open dialog box or to select from a list of recently opened files. Select a file from this list to open it, or select Open File... to access the Open dialog box, browse to locate the desired file, and click Open.

Viewing an XML Workspace or Feature File

- 1 Open the desired XML workspace file.
- 2 From the Main Menu, select FilelView Celera Exchange File.
  The Celera Exchange File Viewer displays.

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3 Drill down to the desired object in the viewer to display its attributes and properties:

Navigating to a Feature in an XML Workspace File

- 1 Open the desired XML workspace file.
- 2 From the Main Menu, select FilelView Celera Exchange File. The Celera Exchange File Viewer displays.
- 3 Drill down to the desired feature and select it.
- 4 From the Viewer's menu, select Genome BrowserlNavigate.
  The Genome Browser displays the selected feature in the Genomic Axis Annotation View and the Property Inspector View.

## Displaying Open Data Sources

You can easily find out which assembly, feature, and workspace files you have open.

- 1 From the Main Menu, select FilelList Open Data Sources.
  A dialog box appears, listing all data sources that the Genome Browser has open now.
- 2 Close the dialog box.





# LOADING PRECOMPUTE AND ANNOTATION FEATURES

To conserve system resources, Genome Browser does not automatically load contig or feature information when you navigate to an axis. You must select a portion of the genomic axis to view, and then load the features for the selected portion.

See <u>"Adding XML Directories" on page 2.18</u> for instructions.

- If you wish to load features that you created using the Celera Exchange File Format, ensure that you have specified the XML directory that contains the feature file(s).
- 2 Before you select a region, you should use the zoom and horizontal scroll bars to zoom in on the desired region.
- 3 Left-click and drag your mouse to select a 500Kb or less region on the axis. The Genome Browser allows you to select a larger region, but it will load data from the start of the selected region up to the maximum range.

To select the entire visible region, select Options|Select Visible Region from the Main Menu.

- 4 From the Main Menu, select Data ManipulationlLoad Data, or rightclick on the axis and select Load Data from the sub-menu.
- 5 Select the annotation and precompute feature types to load from the menu.

TIP: Loading predicted splice site, start, and stop locations can consume a great deal of memory. We suggest that you select a smaller region from a genomic axis when loading these feature sets.

NOTE: Mouse over the Loading Indicator to see the Progress Meter.

The following table shows the Celera feature types (Your feature file may contain different feature types).

### Celera Feature Types

celera Feature Types	
Feature Type	Options
<ul><li> Human Curated Features</li><li> High Pri Computed Features</li><li> Low Pri Computed Features</li><li> Contigs</li></ul>	Select any or none to load information, including underlying contig data, computational analysis results, and annotation (automatic and human-reviewed).
	NOTE: The Genome Browser loads all pre-computed features aligned to the genomic axis' selected region from the relevant feature (.gbf) file(s) when you select High Pri Computed Features.
	TIP: The high-priority computed features are computationally determined to be the most important subset of computed features, and have undergone redundancy filtering. If your system experiences memory problems, you may choose to not load the low-priority computed features.
Predicted Splice Sites	Calculates splice site locations. You can load splice sites on either genomic axis' strand or on both strands. Select one or both of the following:  • Forward
	10111414
Predicted Start Codons	Reverse  Calculates start codon location. Select
Predicted Start Codons	one or both of the following:
	• Forward
	• Reverse
Predicted Stop Codons	Calculates stop codon location. Select

As you load the features, the Genomic Axis Annotation View displays them. The features on the forward strand are above the axis, and those on the reverse are below the axis. The exception would be if the axis was reverse-complemented.

Forward • Reverse

one or both of the following:

Genome Browser

See <u>"Showing or Hiding Data Tiers" on page 4.14 for more information.</u>

Each set of features appears on a "tier." Genome Browser only displays tier labels for those feature sets relevant to the selected region. The tiers are collapsed by default.

When you click on a feature in the Genomic Axis Annotation View, the information for the selected feature appears in the Property Inspector View.

Showing or Hiding Data Tiers

Depending on the number of available feature sets, you may decide to hide some tiers to better see the data that most interests you. Genome Browser defaults to having all tiers available.

To hide a tier, right click on a tier label and select Hide to remove the tier from the display.

NOTE: You should always expand a data tier when you are performing annotation.

To expand or collapse a data tier, right click on the tier's label and select Expand or Collapse. You can single-click on a tier name to toggle between the Expand and Collapse options.

To see long tier names, adjust the tier column by pulling the window margin to the right, as if to widen the window.

To move a particular feature close to the axis, right click on the feature and select Dock. To move a feature (e.g., consensus) away from the axis, right click on the feature and select Undock.

To move the axis tier up and down over precomputed tiers, click and hold the top of the axis tier up/down, or until the double-arrow becomes visible. Clicking a double-arrow at the top of the axis will move the axis to the top of the View. Clicking the double-arrow at the bottom of the axis moves the axis to the bottom of the View.

To change the order of the tiers, click on a tier name and drag it up or down.

To magnify results, select the area to magnify. Press the Alt key and rightclick at the same time, and you will see the image magnified in the View.

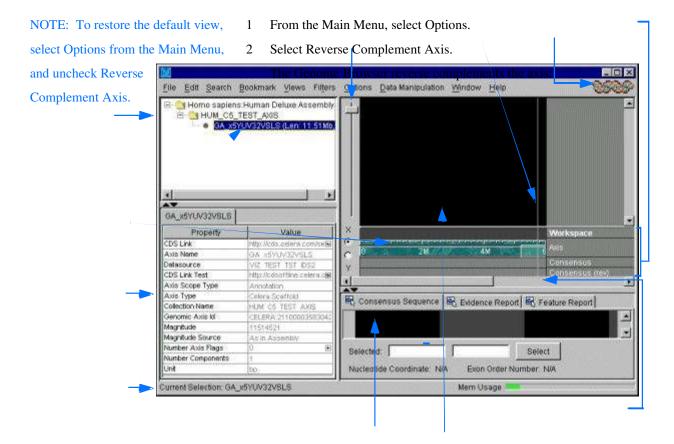


By default, the Genome Browser displays the forward strand from left to right above the axis, and the reverse strand from right to left below the axis:



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When you are reviewing or annotating the reverse strand, it may be easier to flip the display. You can reverse complement the axis to display the reverse strand from left to right on the top of the Genomic Axis Annotation View:



# DISPLAYING A FEATURE'S PROPERTIES

To display any feature's properties, you use your mouse to select it in the Genomic Axis Annotation View.

To select a simple feature, click on the rectangle that represents the feature in the Genomic Axis Annotation View.

A red box appears around the selected feature, and its properties appear in the Property Inspector View.

To select a compound feature, shift-click anywhere on the feature.

- If the feature is a transcript, then simply click on an intron region to select it.
- If the feature is a precompute, click the line linking the glyphs.

A yellow box appears around the feature, and its properties appear in the Property Inspector View.

To select a gene, Ctrl-click on a transcript.

A green box appears around the gene, and its properties appear in the Property Inspector View.

Zooming to a Selected Feature

To zoom in on a selected feature:

From the Main Menu, double-click on the feature, or select Options|Zoom to Axis Selection or Options|Zoom to Subview Selection.

The Genome Browser centers and zooms in on the feature.

Highlighting Evidence

The Genome Browser allows you to highlight precompute features that were used to support a promoted feature. To display this information:

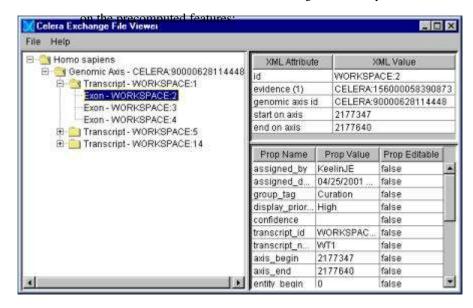
- 1 From the Main Menu, select Options.
- 2 Select Highlight Evidence.

To see an annotated feature's evidence, click the feature in the Promoted tier. The Genome Browser puts a red box around the evidence for that feature:

Showing Edge Matches

The Genome Browser allows you to highlight a promoted feature's edge matches to precomputed features. To display this information:

- 1 From the Main Menu, select Options.
- 2 Select Show Edge Matches.



To see the edge matches for a promoted feature, click the feature in the Promoted tier. The Genome Browser shows edge matches as yellow bars

Showing Evidence Edge Mismatches

The Genome Browser allows you to highlight where the evidence is different than the transcript feature. To display this information:

- 1 From the Main Menu, select Options.
- 2 Select Show Evidence Edge Mismatches .

Showing non-GT/AG Splices Matches

The Genome Browser allows you to highlight exon edges that are non-canonical in blue. To display this information:

- 1 From the Main Menu, select Options.
- 2 Select Show non-GT/AG Splices Matches.

Finding a Feature's Matching Subject Sequence

The Genome Browser allows you to find features that have the same subject sequence on the selected portion of the genomic axis with features loaded. This can be useful when you are evaluating LAP, Genewise, or SIM4 features. For example, you can select a LAP feature and use this function to locate the corresponding BLASTx feature.

- In the Genomic Axis Annotation View, select a range on the genomic axis.
- TIP: If you have the SubView open, click the Subject Sequence
  Report tab to review matching subject sequence information for a selected feature.
- 2 Press <Shift> and click to select the feature.
  A yellow box appears around the feature.
- From the Main Menu, select Options|Find Matching Subject Sequence.

The Genome Browser locates features with the same subject sequence and places a box around these features.

Displaying Consensus Sequence

You can display a region's consensus sequence:

- in the Genomic Axis Annotation View
- in the SubView

To display a region's consensus sequence in the Genomic Axis Annotation View:

- In the Genomic Axis Annotation View, center the Vertical Zoom Center Bar around the region.
- 2 Slide the Zoom Bar, which appears on the left side of the Genomic Axis Annotation View, down until the consensus residues appear:

To display a region's consensus sequence in the SubView:

- 1 If the SubView is not displayed, select Views|Display SubViews When Available from the Main Menu.
- NOTE: If you move the blue glyp, the 2 vertical scroll in the Consensus Sequence window will move in unison.
- When the SubView displays, select a Genomic Axis Range Right-click over the axis and select Select SubView Range. The SubView Range will be initially set to the Annotation view range. The range will be highlighted by a yellow bar, and a blue glyph will appear that allows you to scroll across the consensus sequence. The underlying genomic

	sequence for the axis' selected region appears in the Consensus
	Sequence window:
re the blue glyph to	
l across the	
ence	
erlying genomic	
ence for the axis'	
	Vertical scroll
	vertical scroll
	3 If desired, click on a feature in the Genomic Axis Annotation View.

A yellow box indicates the selected feature, and the color of the feature's sequence in the Consensus Sequence window changes to the same color that appears in the Main Annotation:

Yellow box indicates the selected feature

Color of the feature's sequence matches the same color in the Main Annotation

Displaying the Translations or Reverse Complement

To do this from the SubView:

- 1 Right-click on the Consensus Sequence tab and select Show.
- 2 Click the desired translation and/or Reverse Complement from the submenu.

A check appears next to your selection.

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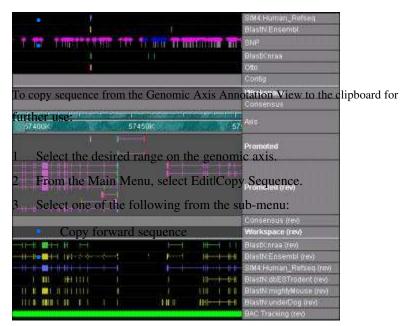
#### Copying Sequence to the Clipboard

To copy consensus residues from the SubView to the clipboard for further use:

1 Select the desired region in the SubView. You can either click and drag to



- 2 Right-click on the Consensus Sequence tab and click Copy Selected Sequence to Clipboard or Copy Selected Sequence to Sequence Analysis.
- 3 Select one of the following from the sub-menu:
  - Nucleotide Sequence
  - Amino Acid Sequence
- 4 If you select Amino Acid Sequence, select either:
  - +1 Translation



#### Setting Color Rules For Feature Display

You can use color rules to change some or all features' color intensity based on the value of a specific feature property.



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NOTE: The Save and Delete buttons are active only if you have already defined a color rule. Also, the Apply Rule and Reset View buttons are active only if you have a region selected on the genomic axis. 2 Click New.

The Color Rule SettingslCreate a New Color Rule dialog box appears:

3 Use the following table to complete this dialog box:

#### Step Instructions

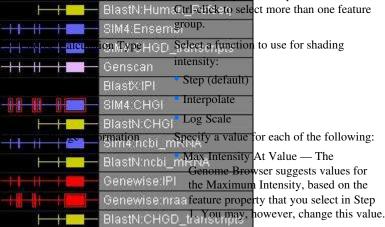
 Select Property to Trigger From Select a Feature Property from the pulldown list. The Genome Browser will apply the color rule based on this feature's property value.

2 Select Effected Feature Groups

Check one of the following boxes:

- Apply To All All features that have the selected feature property will display in the Genomic Axis Annotation View using this color rule.
- Apply To Specific Only the feature group that you select will display using this color rule.

If you check Apply To Specific, select a feature group from the pull-down list box.



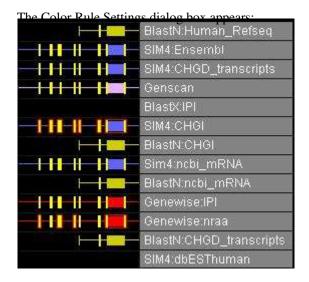
- Min Intensity At Value The Genome Browser suggests values for the Minimum Intensity, based on the feature property that you select in Step 1. You may, however, change this value.
- Ranges Specify the number of ranges. This is applicable for the Step and Interpolate functions only. The Log Scale function uses the extreme values.
- 5 Enter The Rule Name

Type a name for the color rule in the text box.

- 6 When you have completed the information in this dialog box, click OK.
- 7 Click OK to exit the Color Rule Settings dialog box.

#### Editing an Existing Color Rule

1 From the Main Menu, select EditlColor Rules.



Select a Rule to edit from the pull-down list.

Change the existing preferences as desired. The following table describes the settings you may change:

Color Rule Setting

#### Instructions

Rule Settings Group

Select a function to use for shading intensity:

- Step (default)
- Interpolate
- Log Scale

Then, check one of the following

- Apply To All All features that have the selected feature property will display in the Genomic Axis Annotation View using this color rule.
- Apply To Specific Only the feature group that you select will display using this color rule.

If you check Apply To Specific, select a feature group from the pull-

down list box.

Finally, select a Feature Property from the pull-down list. The Genome Browser will apply the color rule based on this feature's property

value.

Data Points Group

To edit the Value or Intensity for a specific data point, click in the row to edit, type your changes, and then press <Enter>.

To add a new data point, complete the Value and Intensity text boxes

and click Add Row.

To delete a data point, click in the row to delete and click Remove

Row.

Select Example Color

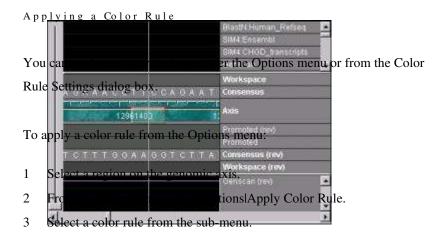
To use a different example color, click Select Example Color and then select a new color. The new color will appear in the Example column.

- 3 When you have made the desired changes, click Save.
- Click OK to exit the Color Rule Settings dialog box.

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#### Deleting an Existing Color Rule

- 1 From the Main Menu, select EditlColor Rules.
- 2 Select a Rule to delete from the pull-down list.
- 3 Click Delete.
- 4 When the Rule Deletion confirmation message appears, click Yes.



To apply a color rule from the Color Rule Settings dialog box:

- 1 Select a region on the genomic axis.
- 2 From the Main Menu, select EditlColor Rules.
- 3 Click Apply Rule.
- 4 Click OK to exit the dialog box.

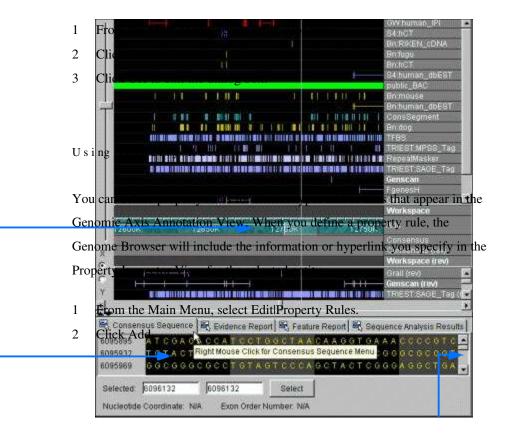
Resetting the View

You can reset the color intensity to 100% from either the Options menu or from the Color Rule Settings dialog box.

To reset the color intensity to 100% from the Options menu:

- 1 From the Main Menu, select OptionslApply Color Rule.
- 2 Select Reset Intensities from the sub-menu.

To reset the color intensity to 100% from the Color Rule Settings dialog box:



3 The Add Property Rule dialog box appears:

The following table explains how to complete this dialog box:

Section

Selection	This displays the current selection's ID,
	Entity Type (feature type), and Discovery Environment (tier name).
Rule Name = text box	Type a name for the rule you are creating in this text box. You may use spaces.
Conditions	1 Select one or more feature types from the When Entity Type = section.
	2 Select one or more tier names from

section.

button.

Instructions

the When Discovery Env =

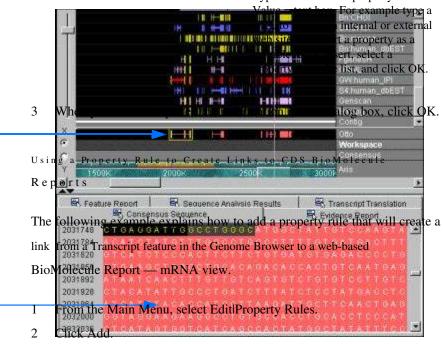
3 Click either the And or the Or radio

#### Section

#### Create Property

#### Instructions

- 1 Type a name for the property in the Name = text box. Do not type spaces or hyphens.
- 2 Type a value for the property in the



- 3 In the Rule Name = text box, type a name for the rule; for example, "BioMolecule Report link".
- 4 In the When Entity Type = section, select "Transcript".
- 5 In the Name = text box, type a property name; for example, "BMR mRNA View".
- 6 In the Value = text box, type the URL to use as follows:

http://myserver/servlet/
com.celera.gateway.GatewayURL?&page=bmr
&species=Homo+sapiens&id=<transcript\_accession>

	4.34	Disp	laying	a Fea	ture's	Proper	ties
--	------	------	--------	-------	--------	--------	------

When you have finished completing the dialog box, it should appear similar to this:

7 Click OK.

Double-click link to access the HTML BioMolecule Report for this transcript.

Displaying Predicted Features in the Transcript Translation View

To review a predicted or promoted feature in the Transcript Translation View:

1 If the SubView is not displayed, select Views|Display SubViews when available from the Main Menu.



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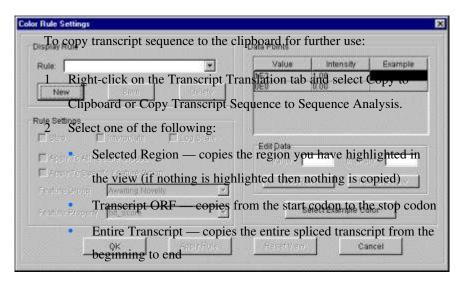
Changing the Translations Display

#### If you wish to change the display:

1 Right-click on the Transcript Translation tab and select Show.

- 2 Select or deselect a menu item.
- 3 Repeat steps 1 through 2 until the display is as you wish.

Copying Transcript Sequence to the Clipboard



- 3 Select one of the following from the sub-menu:
  - Nucleotide Sequence
  - Amino Acid Sequence

- 4 If you select Amino Acid Sequence, select one of the following:
  - +1 ORF
  - +2 ORF



5. Enter The Rule Name

The Genome Browser allows you to view the alignment between the Celera consensus sequence and the sequence for any homology feature. This is particularly useful for BLAST, SIM4, Genewise, and LAP.

- In the Genomic Axis Annotation View, select a region from the axis that is larger than the feature you want to view.
- 2 If the SubView is not displayed, select Views Display SubViews when available from the Main Menu.
- 3 Click on a homology feature in the Genomic Axis Annotation View.

  The Query Sequence Alignments tab appears in the SubView.
- 4 The Query Sequence Alignments includes a zoom and zoom center bar. Zoom in the Query Sequence Alignments to review the sequence homologies.

TIP: If a feature has more than one exon, the Genome Browser will automatically zoom in on the first exon. To zoom in on the second or other exon, click on it before you zoom in.

Depending upon the analysis program for the selected feature, the window displays either nucleotides or amino acids (i.e., a BLASTN feature displays DNA sequence, and a BLASTX feature displays protein sequence). The Celera sequence is on top, and the subject sequence is

NOTE: You must select a feature that 3 is completely within the range you have selected on the axis. The doublearrow in the upper left of the the Subview allows you to expand or collapse the View.

NOTE: To review an HTML report, right-click on the DbAlignView tab and select HTML Report! Show HTML Report.

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below. The frames are relative to the entire sequence of the genomic axis.

The Query Sequence Alignments indicates homology by varying shades of color. For protein sequences, bright green indicates a perfect match, and for nucleotide sequences, bright blue indicates a perfect match.

- 5 Select another feature, if desired.
- 6 To delete the contents of the Query Sequence Alignments window, right-click on the Query Sequence Alignments tab and select Clear.

Displaying the Feature Report

NOTE: The Feature Report displays information for only the data within the selected SubView range on the axis.

- In the Genomic Axis Annotation View, select a region from the axis that contains the features you wish to view in the report, and load the features for the range.
- 2 If the SubView is not displayed, select ViewslDisplay SubViews when available from the Main Menu.
- 3 In the SubView, click the Feature Report tab.
- 4 Select Options|Populate.

NOTE: Clicking on a feature in the Genomic Axis Annotation View highlights the corresponding row in the Feature Table (if selected). The Genome Browser populates the Feature Report. The following table explains how to find information in this report.

To: Do this:

Sort data by a column heading in descending order.

heading.

Sort data by a column heading in ascending order.

Shift-click on the column

Click on the column

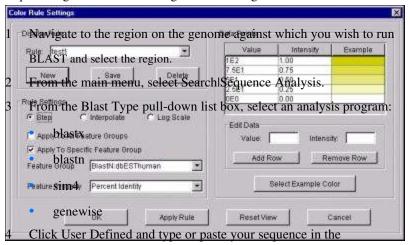
heading.

Highlight report data in the

Click in the row that you Genomic Axis Annotation View. want to highlight to select an individual feature, or shift-click to select the composite feature. A colored box appears around the feature in the Genomic Axis Annotation View.

### RUNNING BLAST SEARCHES

The Genome Browser allows you to compare a public or proprietary sequence against a selected range on a Celera genomic axis.



Sequence text box. It does not matter whether you include the definition line.

- Click Search.When the Search button becomes active again, the search is complete.
- 6 Click the Sequence Analysis Results tab in the SubView to review your results.

# SEARCHING FOR A NUCLEOTIDE OR PROTEIN SEQUENCE STRING

You can search for a nucleotide or amino acid sequence string in either the Consensus Sequence or Transcript Translation window.

- If the SubView is not displayed, select Views|Display SubViews when available.
- 2 Select either the Consensus Sequence or Transcript Translation tab.
- 3 Right-click your mouse and select Find... or press <Ctrl-F>.
- 4 Type or paste a search string in the Find what box.

  TIP: You can use keyboard shortcuts for copying (<Ctrl-C>) and pasting in this box (<Ctrl-V>).
- 5 Select an option from the Look in pull-down list:
  - Nucleotide
  - Nucleotide Reverse Complement
  - ORF +1
  - ORF +2
  - ORF +3
  - All ORF's

NOTE: If you select Nucleotide
Reverse Complement or an Amino
Acid option, you do not need to be
displaying that type of data in the
Consensus Residues or Transcript
Translation window. For example, the
Genome Browser will find the reverse
complement of ATG, even if the
window is not displaying the
Nucleotide Reverse Complement
when you search.

- 6 If you selected an Amino Acid option from the Look in pull-down list, select either one-letter or three-letter from the Translation box.
- If you click Find Next, the Genome Browser will return the next occurrence of the search string.

If you click Find All, the Genome Browser will return a table that displays all occurrences of the search string, with the corresponding locations.

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Click Find Next or Find All.

### USING BOOKMARKS

Genome Browser allows you to create and edit bookmarks. You can bookmark contigs and precomputed and annotated features.

#### Creating

- 1 Click the item you want to bookmark to select it.
- 2 From the Main Menu, select Bookmark|Bookmark Current Selection.

#### Opening a Bookmark

- 1 Ensure that you have an information service open.
- 2 From the Main Menu, select Bookmark|Select Bookmark.
- 3 Select your bookmark from the sub-menu.

#### Editing or Deleting a Bookmark

- 1 From the Main Menu, select Bookmark/Select Bookmark.
- 2 Select the bookmark that you wish to delete or modify.
- Select Delete or Edit.

  If you select Delete, the bookmark disappears from the window. If you select Edit, the Bookmark Properties box appears, and you can type over the existing name.
- 4 Click Close when you are done.

# DISPLAYING MULTIPLE BROWSER WINDOWS

Genome Browser allows you to display more than one browser window at a time. This allows you to review multiple views of the same genome.

- 1 From the Main Menu, select Window.
- 2 Select New Browser Window.

resources, displaying multiple browser windows may consume more memory than is acceptable for your computer.

NOTE: Depending on your system's

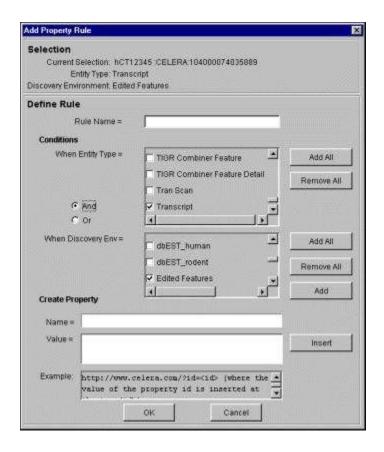
Genome Browser displays a duplicate of the window that you already have open. You can either review different features for the same data, or you can select different data to review.

Closing a Browser Window

- 1 Click in the browser window you want to close to activate it.
- 2 From the Main Menu, select Window.
- 3 Select Close Browser Window.

### PRINTING A PAGE

To print any screen while you're in Genome Browser, select FilelPrint Screen.



	<u>5.2</u>
Introduction	<u>5.2</u>
Edit Commands	<u>5.3</u>
Annotation History	<u>5.4</u>
Creating a Transcript or Exon Using a Computed Feature	<u>5.5</u>
Reviewing and Editing Comments	<u>5.7</u>
Adding Evidence to a Transcript or Exon	<u>5.7</u>
Removing Evidence from a Transcript or Exon	<u>5.7</u>
Splitting a Transcript	<u>5.8</u>
Merging Transcripts	<u>5.9</u>
Adjusting Exon Boundaries	
Using the Consensus Sequence Window to Modify an Exon	<u>5.9</u>
Using Evidence to Modify an Exon	<u>5.10</u>
Using a Splice Site to Modify an Exon	<u>5.10</u>
Adding or Deleting Bases in the SubView	<u>5.11</u>
Finding a Long Open Reading Frame	<u>5.11</u>
Evaluating Subject Sequence Alignments	<u>5.12</u>
Deleting a Feature from the Workspace	<u>5.12</u>
Exporting Annotation Data	<u>5.13</u>

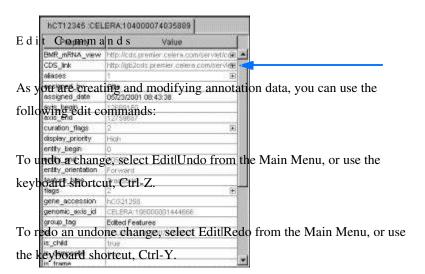
### INTRODUCTION

As your eview the need to create, modify, and export annotation data. When you enable curation, the Genome Browser allows you to create new features or modify existing features in the Workspace area: transcripts Rule Name = BioMolecule Report Link Conditions genes When Entity Type = ▼ Transcript Add All Transcription Factor Site (P) exons Remove All Translation Start Position start and stop codons Virtual Exon evidence relationships Awaiting Novelty Add All BAC Tracking By default, the Genome Browser enables curation. If you want to create or modify fearthes, you need to enable curation, which you do from the Preferences dialog box. Value = .celera.gateway.GatewayURL?spage=bmr aspecies=Homo+sapiensaid=<transcript acce Before you begin annotating you should where the due of the property id is inserted Enable the Highlight Evidence and Show Edge Matches features OK Cancel (default)

See "Specifying System Preferences" on page 2.15 if you do not know how to disable curation.

See <u>page 4.18</u> to learn how to highlight evidence and <u>page 4.18</u> to learn how to show edge matches. See <u>page 4.14</u> to learn how to expand the data tiers.

- Expand the data tiers with which you are working.
- Open the SubView.

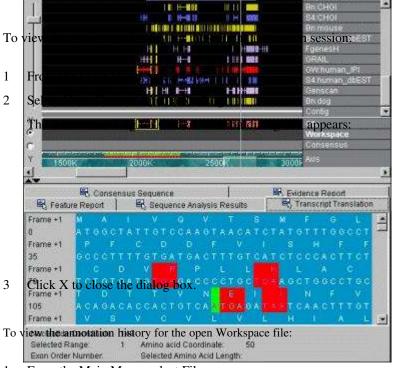


To copy information from the Property Inspector View to the clipboard, select the information and use the keyboard shortcut, Ctrl-C. To paste the contents of the clipboard, position your cursor in the appropriate place, and use the keyboard shortcut, Ctrl-V.

#### Annotation History

The Genome Browser tracks the annotation that you create and modify. You can review the annotation history for either:

- the current annotation session
- the open Workspace file



- 1 From the Main Menu, select File.
- Select View Open Workspace Annotation Log.The Workspace Log dialog box appears:

#### <u>TBA</u>

3 Click X to close the dialog box.

# CREATING A TRANSCRIPT OR EXON USING A COMPUTED FEATURE

NOTE: To select a transcript, click on 3 an intron region, or shift-click on one of its exons.

Click the new feature to select it.

The Property Inspector View displays the information for this feature.

The Genome Browser automatically sets the assigned by and assigned date fields in the Property Inspector View. The Genome Browser uses your system login id for the modified by value. Also, if you drag a transcript from the Promoted tier into the Workspace, the modified by and modified date are automatically set, too. You can modify the release status, reviewed by, and reviewed date fields.

# REVIEWING AND EDITING COMMENTS

You may create and edit comments for transcripts and exons in the workspace. Precompute and promoted feature comments are read-only.

NOTE: If a feature does not have comments to display, the comments field does not appear.

To access comments for any feature, select it in the Genomic Axis Annotation View and click the + that appears in the comments field value column in the Property Inspector view. You may need to scroll down to see it.

If the comments are read-only, the following dialog box appears:

If the comments are not read-only, the following dialog box appears:

The following table describes this dialog box.

Element Description/Instructions

New Comments Allows you to to add comments to this feature:

Type the information in the New Comments text box, and

click Add Comment.

You can add multiple comments in this text box; simply

click Add Comment after each item.

Existing Comments

Displays existing comments for this gene.

### ADDING EVIDENCE TO A TRANSCRIPT OR EXON

To add computed evidence to a transcript or exon that appears in the Workspace:

NOTE: The Genome Browser will not 1 add the evidence to the transcript if 2 you place the evidence on an intron 3 region. You must drag it on top of an exon.

- Click the evidence in the Genomic Axis Annotation View to select it.
- 2 Drag it down to the Workspace on top of an exon.
- 3 Release your mouse button.

# REMOVING EVIDENCE FROM A TRANSCRIPT OR EXON

#### To remove evidence:

- 1 Click the exon in the Workspace.
- 2 Right-click on the evidence to display the Evidence Options menu, and select Remove as evidence for current curation.

### SPLITTING A TRANSCRIPT

To split a transcript in the Workspace:

- 1 Click on the intron region in the area where you wish to split the transcript.
- 2 Right-click your mouse and select Split Transcript.
  The Genome Browser splits the transcript in the location where you right-clicked.

### MERGING TRANSCRIPTS

First, you need to ensure that the transcripts are attached to the same gene:

- 1 Click the transcript in the Workspace to select it.
- 2 Right-click on an intron region and select Create New Gene from the context menu.
- 3 Ctrl-click to select the newly created gene to which the transcript will be attached.
- 4 Right-click your mouse on the transcript to attach and then click Attach
  Transcript to Selected Gene.
- 5 Ctrl-click on either feature, and a green box appears around both.

Next, you can merge the two transcripts in the Workspace:

- 1 Select the first transcript in the Workspace.
- 2 Right-click on an intron region in the second transcript.
  The context menu appears:

Select Merge with Selected Transcript.
 The Genome Browser merges the transcripts in the Workspace.

### ADJUSTING EXON BOUNDARIES

You can modify exon boundaries using:

- the Consensus Sequence window
- Evidence
- Splice sites

The following sections provide instructions for each method.

Using the Consensus Sequence Window to Modify an Exon

- 1 If the SubView is not displayed, select Views|Display SubViews when available from the Main Menu.
- 2 Select a range along the axis. A red bar appear marking the range you selected.

NOTE: The SubView Range should be 3 greater or equal to the Parent Feature range to view the colored nt's. 4

- Select the Subview range by selecting Data Manipulation|SubView Range. A yellow bar marks the range you selected.
- 4 Select the exon to modify from the Axis.
- 5 Click the Consensus Residues tab, if it is not on top.

  The underlying genomic sequence information appears in the box, in the area of the selected feature. A yellow bar on the Genomic Axis indicates the relative position. Exon sequence appears as purple, and intron sequence appears as yellow.

NOTE: You must begin with the first/ 6 last base of the exon, or the boundary will not change. You can not move exon boundaries past a Start or Stop codon.

Starting at the exon's edge, drag the edge to the desired position.

The boundary change is reflected on the axis.

Using Evidence to Modify an Exon

If you have added evidence to a transcript in the Workspace, you can tell the Genome Browser to use the evidence to modify an exon:

NOTE: The Genome Browser will not 1 add the evidence if you drag it to an 2 intron region. You must drag it on top 3 of an exon.

- Click the evidence in the Genomic Axis Annotation View to select it.
- 2 Drag it down to the Workspace on top of the exon.
- 3 Click the exon to modify in the Workspace to select it.
- 4 Right-click on the evidence to display the Evidence Options menu:

NOTE: To undo a change, select FilelEditlUndo... from the Main Menu.

5 Select an option from the menu.
You can set the left and right edge of a current curation, as well as remove any evidence.

Using a Splice Site to Modify an Exon

- Load the Predicted Splice Sites data for the selected axis range.

  TIP: Loading predicted splice site locations can consume a great deal of memory. We suggest that you select a smaller region from the genomic axis before you load splice sites.
- 2 Click on the splice site to use.
- 3 Drag it down, and place your mouse pointer over the exon.
- 4 Release your mouse button.

# ADDING OR DELETING BASES IN THE SUBVIEW

You can add or delete bases to a transcript in the SubView to see the effect on a translation.

NOTE: You can not save these changes.

- 1 If the SubView is not displayed, select ViewslDisplay SubViews when available from the Main Menu.
- 2 In the Genomic Axis Annotation View, click on a feature.
- 3 Select the Transcript Translation tab in the SubView.
- 4 In the SubView, type or delete bases in the sequence.

# FINDING A LONG OPEN READING FRAME

- 1 If the SubView is not displayed, select Views|Display SubViews when available from the Main Menu.
- 2 In the Genomic Axis Annotation View, click on a feature in the workspace to select it.
- 3 Select the Transcript Translation tab in the SubView.
- 4 In the SubView, right-click your mouse any where in the sequence.

  The Curation Options sub-menu appears:

NOTE: The changes may not be apparent in the Genomic Axis Annotation View.

### 5 Select one of the following:

- Set Longest Open Reading Frame calculates the longest open reading frame from Stop to Stop codon.
- Set Longest ATG to Stop calculates the longest open reading frame from Start to Stop codon.
- Stop Codon

# EVALUATING SUBJECT SEQUENCE ALIGNMENTS

- 1 If the SubView is not displayed, select Views|Display SubViews when available.
- 2 In the Genomic Axis Annotation View, click on a feature to select it.

NOTE: Query Sequence Alignments View allows you to see nt-nt HSPs and aa-aa HSPs in color. Select the Subject Sequence Alignments tab.
The Genome Browser displays the Celera data hits along the subject sequence's axis.

### DELETING A FEATURE FROM THE WORKSPACE

To delete a transcript, exon, and gene from the Workspace:

- 1 Click the feature to select it, and right-click your mouse.
- 2 Select Delete Transcript (or Exon) from Workspace.

# EXPORTING ANNOTATION DATA

When you export the data, the Genome Browser saves the Workspace file as XML in the Celera Exchange File Format. You can then reload this file at a later time to modify the annotation data.

- 1 From the Main Menu, select FilelSave Workspace.
- 2 In the Save dialog box, browse to locate the desired folder.
- Type a file name, and click Save.A dialog box appears asking whether you want to clear the workspace.

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# Appendix A. Exchange File Format

Introduction	<u>A.2</u>
XML Markup Element Descriptions	<u>A.3</u>
Document Type Definition	<u>A.10</u>
Example Feature File	<u>A.13</u>

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# INTRODUCTION

The Celera Data Model is based on the concept of data objects with identity. This means that every piece of genomic data has a permanent independent existence and can be referenced via a unique object id, known as a UID. Data objects come in various types, which can form relationships with other data objects via references to their UIDs.

UIDs are alphanumeric strings that can uniquely identify any data object created. Since they are by definition unique over all data sources and over all time, UIDs allow a data object to reference any other data object, even when they are located in separate data storage media or created at different times.

Not all data providers have access to a common source of unique identifiers, so the UID format is broken into parts separated by a colon: an alphanumeric prefix, called the UID Space, and a decimal suffix which represents an integer less than 264 power. The prefix, which is case-insensitive, identifies the UID space and represents the authority guaranteeing the uniqueness of the suffix integer. UIDs generated by the Celera UID server, for example, all have the "CELERA" prefix. For example:

#### CELERA:nnnnn

You can open data objects from other sources with unique identifiers into the Genome Browser without fear of UID collisions, simply by picking a unique UID space prefix. The Genome Browser can open data objects with up to 2<sub>16</sub> distinct UID spaces simultaneously. This UID specification is consistent with the XML attribute definitions of ids and idrefs that can fail to pass a validating parser if they are simple numeric strings.

# XML MARKUP ELEMENT DESCRIPTIONS

The Celera Exchange File Format is based on a subset of the GAME DTD, with a small number of minor extensions. The following table describes the syntax of each XML element, or tag, and any file (.gbf, or .gbw) restrictions.

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# Element and Description

# Attributes and Elements

#### <game>

The root node, or document element, for a valid Celera Exchange Format file.

# Required

# ATTRIBUTES:

version — the version of game DTD to which this document conforms.

# Optional ATTRIBUTES:

assembly\_version — the version of the assembly data in the database. This attribute is for feature (.gbf) and workspace (.gbw) files that import data on top of the database.

taxon — the scientific name of the species of organism with which the data

is associated.
ELEMENTS:
<annotation>

<computational\_analysis>

<feature\_set>

**ELEMENTS:** 

<seq>— a single feature (.gbf) or workspace (.gbw) file may contain any number of elements of type "SUBJECT".

#### <annotation>

This corresponds directly to a Gene data object, and is used to represent a human-curated gene.

#### ATTRIBUTES:

id — the UID for the gene. It must appear once and only once.

**ELEMENTS:** 

<feature\_set> — represents

Transcript data objects. This element may occur more such as URL may occur more train of the followed by a decimal integer. This element may occur more than once.

Transcript data objects. This element may occur more such as URL may occur more than once.

The following optional element is for curated gene. For Celera-annotated Workspace (.gbw) files, only:

Transcript data objects. This element may occur more than once.

Workspace (.gbw) files, only:

The following optional element is for curated gene. For Celera-annotated workspace (.gbw) files, only:

Transcript data objects. This element may occur more than once.

The following optional element is for curated gene. For Celera-annotated workspace (.gbw) files, only:

The following optional element is for curated gene. This objects that are intended to be modified.

Annotations saved out in (.gbw) files have <name> elements with "WG" prefix followed by decimal integer, representing workspace genes generated on the client. The Genome Browser automatically generates these elements.

# Element and Description

### <computational analysis> This represents a group of precomputed features generated by a particular algorithm or analysis program.

# <feature set>

This corresponds directly to a Transcript data object and is used to describe a human-curated transcript. Feature sets contained within <annotation> elements correspond to Transcript data objects that have been assigned to a specific Gene data object. However, <feature\_set> elements can also exist directly under a <game> element, in which case they describe Transcript data objects not assigned to any Gene data object.

#### Attributes and Elements

#### Required

#### ATTRIBUTES:

evidence relationships that refer to an entire computational algorithm, rather than an individual result set. **ELEMENTS:** <result\_set> —Represents the precomputed features. cprogram> —This must contain a name, which is used by the Genome

Browser to establish a horizontal tier for displaying the precomputed features.

id — stores a UID, which can be used by

#### ATTRIBUTES:

id —the UID for the transcript. **ELEMENTS:** 

<type> — must contain the string "transcript" and must occur once and only once.

<name> — an alphanumeric string containing the accession number for the curated transcript. For Celera-annotated transcripts, this should have a "CT" prefix followed by a decimal integer. Feature\_sets saved out in (.gbw) files have <name> elements with "WT" prefix followed by decimal integer, representing workspace transcripts generated on the client. <feature\_span> —represents the Exon, Start\_Codon, Stop\_Codon, and Translation Start Position data objects that make up the transcript. This element may occur more than once.

#### Optional

#### **ELEMENTS:**

<date> — indicates when the program was run. <database> — indicates against which database the analysis program <parameter> — represents the type of algorithm (e.g. blastx, blastn) used by

#### **ELEMENTS:**

each run of the program.

<replaced> — refers to the promoted Transcript data objects that are intended to be modified. This element is for Workspace (.gbw) files only, and it may occur more than once.

#### Element and Description

This represents a piece of nucleotide

#### <seq>

sequence against which <result\_span> and <feature\_span> elements may be aligned. In a feature (.gbf) or a workspace (.gbw) • AA file, the <seq> element may represent some other subject sequence against

which a feature is aligned.

#### Attributes and Elements

#### Required

#### ATTRIBUTES:

id — The UID for the data object. type — Use one of the following values:

- DNA
- RNA

# Optional

#### ATTRIBUTES:

length — the number of nucleotides in the sequence.

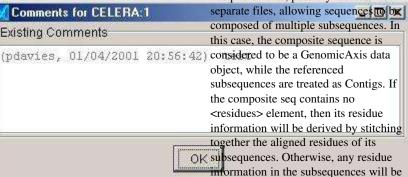
#### **ELEMENTS:**

<residues> —the raw sequence data stored as characters.

<dbxref> — the database source for this sequence data.

<description> — general information. The following optional may be used more than one time:

<seq\_alignment> — refers to other <seq> elements, possibly located in



# Comments for CELERA:1

#### **Existing Comments**

<feature\_span>

This describes simple human-curated features that can be combined to make a feature set.

The <feature\_span> element may not stand alone. It must be contained within a <feature\_set> element.

#### ATTRIBUTES:

id — the UID for the data object. **ELEMENTS:** 

<seq\_relationship> — indicates the <feature\_span> element's location on an assembly sequence. <type> — contains one of the following values:

- exon
- · start-codon
- stop-codon
- frame

These values correspond to the Exon, Start\_Codon, Stop\_Codon and Translation\_Start\_Position data objects.

#### **ELEMENTS:**

ignored.

<evidence> — refers to precomputed results on which the feature span is

<replaced> — refers to the promoted data objects that are intended to be modified. This element is for workspace (.gbw) files only, and it may occur more than once.

### Element and Description

<seq\_relationship>

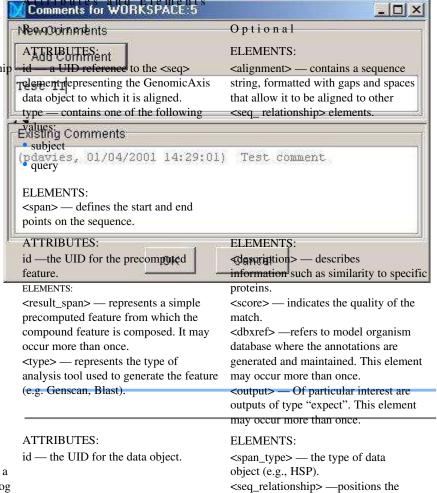
This describes the geometric relationship between a <feature\_span> or <result\_span> element and its <seq> element.

#### <result\_set>

This represents a compoundprecomputed feature data object, and is a precomputed analog to the <feature set> element for humancurated features.

#### <result\_span>

This describes simple precomputed features that can be combined to make a feature set, and is a precomputed analog of the <feature\_span> element. The <result span> element may not stand alone. It must be contained within a <result\_set> element.



<result\_span> on a <seq> element. Seq relationships of type "subject" and "query" refer to spans on a subject and query sequences respectively <score> — indicates the quality of the match. This element may occur more than once. <output> — This takes sub-elements <type> and <value>, when type is "Expect" <value> is the expect value. This element may occur more than once.

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# Element and Description

# <seq\_alignment>

This indicates that the containing <seq> should derive a portion of its sequence from a referenced <seq> element.

#### Attributes and Elements

# Required

#### ATTRIBUTES:

seq\_id — references the UID of the subsequence <seq> element.

### Optional

### ATTRIBUTES:

start and end — indicate the range and orientation on the composite sequence where the subsequence is to be aligned entity\_start and entity\_end — the corresponding range and orientation on the subsequence. file\_path — the path string for an assembly (.gba) file that contains the subsequence. If no file is specified, the subsequence is assumed to be in the

same file as the composite sequence.

# Element and Description

#### cproperty>

This stores a name-value attribute pair representing some piece of information attached to a specific instance of a data object (e.g., a URL).

The properties are displayed in the Genome Browser in the Property Panel, and properties containing valid URLs can be used to hyperlink via a web browser.

#### <comments>

This stores a single comment text. An <annotation> element may contain multiple comments.

#### <replaced>

This is used only by human-curated features in workspace (.gbw) files.

#### Attributes and Elements

### Required

#### ATTRIBUTES:

name value

#### Optional

#### ATTRIBUTES:

editable — indicates whether the attribute can be modified by the user, and is by default "false". Only humancurated features (e.g., Gene, Transcript) may have modifiable properties.

**ELEMENTS:** 

property> — enables hierarchies of property> elements to any depth.

### ATTRIBUTES:

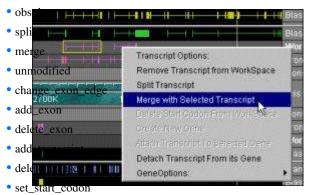
author date

#### ATTRIBUTES:

id — the UID of the reference to a promoted human-curated feature, that is intended to be modified.

type — indicates the nature of the modification. Use one of the following values:

• update



- remove\_start\_codon
- changed\_properties
- · added\_comment

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# DOCUMENT TYPE DEFINITION

```
<?xml encoding='UTF-8' ?>
<!ELEMENT game ( (annotation | computational_analysis |</pre>
feature_set | seq )* )>
<!ATTLIST game version
                                CDATA #IMPLIED
                assembly_version CDATA #IMPLIED
                taxon
                                CDATA #IMPLIED >
<!ELEMENT annotation (name , description? ,
annotation_source? , comments* , feature_set+ , replaced* ,
property* )>
<!ATTLIST annotation id CDATA #REQUIRED >
<!ELEMENT computational_analysis (date? , program ,</pre>
version? , parameter* , database? , result_set+ )>
<!ATTLIST computational_analysis id CDATA #REQUIRED >
<!ELEMENT feature_set (name , type , annotation_source? ,</pre>
author? , creation_date? , version? , replaced* , property*
, feature_span+ )>
<!ATTLIST feature_set id CDATA #REQUIRED >
<!ELEMENT seq (dbxref? , description , residues? ,
seq_alignment* )>
<!ATTLIST seq length CDATA #IMPLIED
               id CDATA #REQUIRED
               type CDATA #REQUIRED >
<!ELEMENT name (#PCDATA )>
<!ELEMENT description
                      (#PCDATA )>
<!ELEMENT date EMPTY>
<!ATTLIST date day CDATA #REQUIRED
               year CDATA #REQUIRED
               month CDATA
                            #REQUIRED >
<!ELEMENT progra • ( #PCDATA ) >
<!ELEMENT version (#PCDATA )>
<!ELEMENT parameter (type , value )>
<!ELEMENT database (name , date? , version? )>
<!ATTLIST database
                   id CDATA #IMPLIED >
<!ELEMENT result_set (description? , type? , score? ,
dbxref* , output* , result_span* , property* )>
<!ATTLIST result_set id CDATA #REQUIRED >
<!ELEMENT type (#PCDATA )>
<!ELEMENT author (#PCDATA )>
<!ELEMENT creation_date (#PCDATA )>
<!ELEMENT feature_span (type , annotation_source? ,
seq_relationship , property* , replaced* , evidence* )>
<!ATTLIST feature_span id CDATA #REQUIRED >
<!ELEMENT dbxref (xref_db , db_xref_id , version? )>
```

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```
<!ELEMENT value (#PCDATA )>
<!ELEMENT score
                                       (#PCDATA )>
<!ELEMENT output (type , value )>
<!ELEMENT result_span
                                                        (span_type? , score? , output* ,
value? , seq_relationship* , property* )>
<!ATTLIST result_span id CDATA #REQUIRED >
<!ELEMENT seq_relationship (span , alignment? )>
                                                                     id CDATA #REQUIRED
<!ATTLIST seq_relationship
                                                                    type (query | sbjct ) #IMPLIED >
<!ELEMENT evidence (#PCDATA )>
                                                                         (homology | gene_prediction )
<!ATTLIST evidence
                                                    type
#IME #IME
                              Evidence Options:
                                                                                            IMPLIED >
                         crefsed set left #PCDATATest curation
           1 1 1 1
                          Use to set right edge of current curation
                         ab Reference evidence of the Reference of the second secon
           1 11 111 11
<!EI
<!ELEMENT span (start , end )
<!ELEMENT alignment (#PCDATA
<!ELEMENT start
<!ELEMENT end (#PCDATA )>
<!ELEMENT replaced (#PCDATA )>
<!ATTLIST replaced type (unmodified |
                                                                     modified
                                                                      obsolete
                                                                      new
                                                                      split
                                                                      merge
                                                                      deep-modified ) #REQUIRED
                                                    id
                                                             CDATA #REQUIRED >
                                                   (property* )>
<!ELEMENT property
                                                    name CDATA #REQUIRED
<!ATTLIST property
                                                    value CDATA #REQUIRED
                                                    editable (true | false )
                                                                                                                           #IMPLIED >
<!ELEMENT seq_alignment (#PCDATA )>
                                                                                                  CDATA #IMPLIED
<!ATTLIST seq_alignment
                                                                 start
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                                                                 entity_start CDATA #IMPLIED
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                                                                 file_path
                                                                                                  CDATA #IMPLIED
                                                                                                  CDATA #REQUIRED >
                                                                 seq_id
<!ELEMENT residues
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                                                   (#PCDATA )>
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```

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<!ELEMENT annotation\_source (#PCDATA )>

Curation Options:
Set Start Codon
Set Translation Start
Set Longest Open Reading Frame
Set Longest ATG to Stop
Decks Start Codon
Decks Start Codon
Decks Start Codon Fram Optiobase
Set Stop Codon to Calculate ORF upstream

# EXAMPLE FEATURE FILE

This section provides an example Celera Exchange Format feature (.gbf) file.

There are additional example files on the Genome Browser download page:

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                 Test
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given none given A=3
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 ${\tt TGACACACCAGGTTAATATCTTTAATATATACCGAGCTCATACAAATTTATCTGACCC}$ 

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<alignment>

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Troubleshooting	<u>B.2</u>
Memory Problems	<u>B.2</u>
Data Access Problems	<u>B.2</u>
Exception Errors	<u>B.3</u>

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# TROUBLESHOOTING

This appendix contains answers to some common troubleshooting questions.

If after reviewing this appendix, you still can't solve your issue, you should contact your Celera Technical Support Specialist.

Memory Problems

In general, you can solve many issues by ensuring that you have allocated adequate memory for the Genome Browser. For instructions, please refer to "Troubleshooting Performance Issues" on page 1-5.

Data Access Problems

If you are not properly logged in, you will receive the following message when you attempt to open an information service:

You will need to restart the Genome Browser and re-enter your login information. For instructions, please refer to "Setting Preferences" on page 2-6.

# Exception Errors

If the Genome Browser experiences an exception error, an error dialog box will appear that allows you to send a message to the Celera support staff. If this happens, you should:

- 1 Click Send email to Celera Support.
- 2 Type your e-mail address in the box provided, and click OK.

NOTE: If you prefer to be contacted by telephone, you should include your name and telephone number in the description box.

- Type a description of what you were doing when the exception occurred in the box provided, and click OK.

  A message appears indicating that your problem was sent to the Celera Support staff.
- 4 Click OK.

A Celera Support representative will contact you.

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B.4 Troubleshooting			

