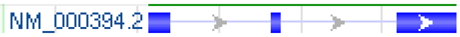
1 Feature Rendering

This section covers features that use generic feature rendering settings. The features (SNPs, structural variants, clone placements, and segmental duplications) that require special handling are covered in the later sections.

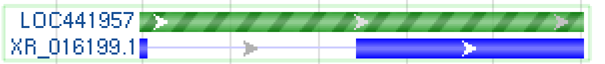
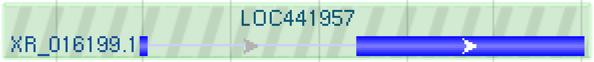
1.1 Feature Color Code

Feature type	Color	Visual Examples
Gene	Green	
RNA	Blue	
Coding region	Red	
All other features	Black	

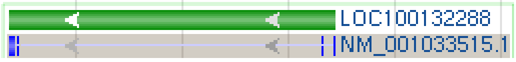
1.2 Special Rendering Styles

For features with special attributes, special rendering will be applied.

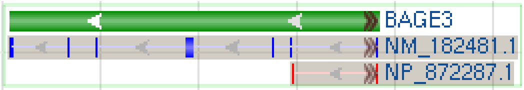
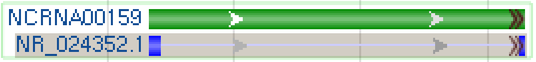
1.2.1 Genes marked as pseudo

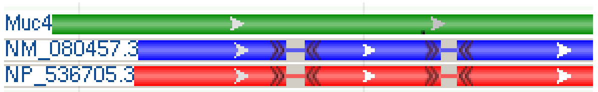
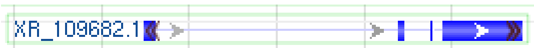
Settings	Visual Effect	Visual Examples
All features are shown in one gene group	Stripes over green gene bar	
Gene bar is hidden	Green stripe background	
Gene bar and gene label are hidden	Green stripe background	

1.2.2 Features with exception text

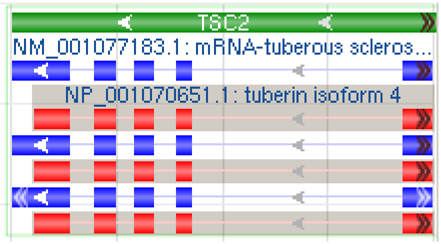
Exception Example	Visual Effect	Visual Examples
Mismatch in transcription	Shaded background	
Mismatch for translation	Shaded background	
Unclassified transcription discrepancy	Shaded background	

1.2.3 Feature location marked as partial

Example	Visual Effect	Visual Examples
Partial start	Black “<<” or “>>” at 5’ end	
Partial stop	Black “<<” or “>>”at 3’ end	

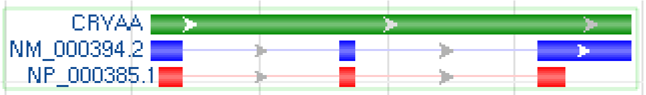
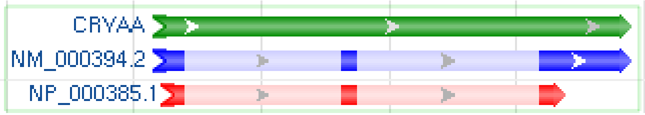
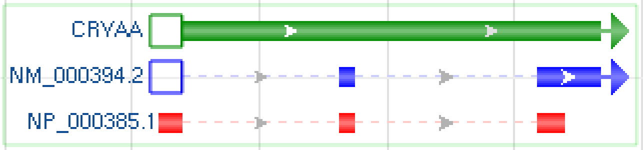
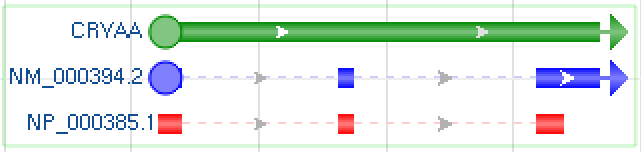
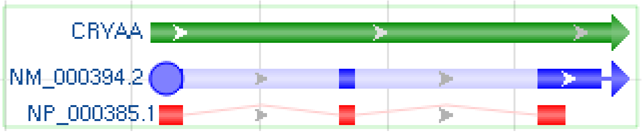
Partial start and stop for internal fuzz on mRNAs/CDSs	Black “<<” and “>>” at both ends	
Partial start and stop	Black “<<” and “>>” at both ends	

1.2.4 Features marked as partial

Example Cases	Visual Effect	Visual Examples
Example includes a partial feature (marked as white “<<” and “>>”) and features with partial stop (marked as black “>>”)	White “<<” and “>>” at both ends	

1.3 Feature Decorations

Different feature decoration styles are solely for offering different ways of visualization. The exact same feature can be rendered using each of the five existing styles.

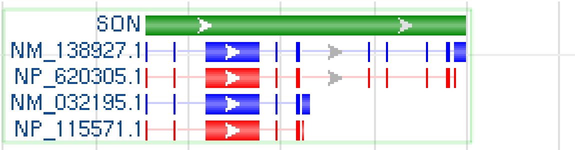
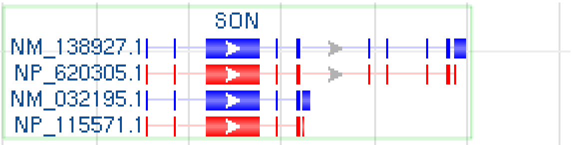
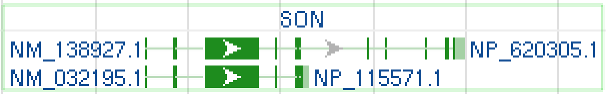
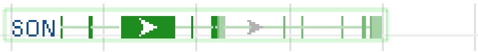
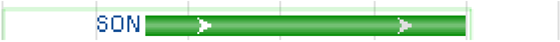
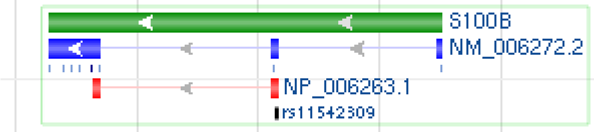
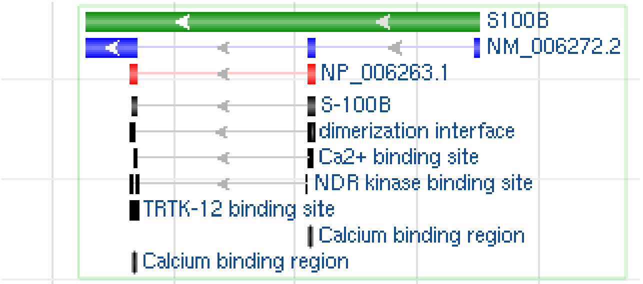
Décor Styles	Visual Effect	Visual Examples
Default	Solid bars for feature intervals or exons, and solid lines for introns	
Arrows	Arrows at both ends showing the strand, and lighten bars for introns	
Square anchor	Square for feature start, arrow for feature stop, dash lines for introns	
Circle anchor	Circle for feature start, arrow for feature stop, dash lines for introns	
Fancy	Circle for mRNA start only, square for other features start except for gene and CDS, arrow for feature stop, lighten bars for mRNA introns, and canted lines for CDS introns	

Note: the glyphs representing feature start and stop will be rendered only when their sizes can fit into the first and last intervals.

2 Gene Model Features

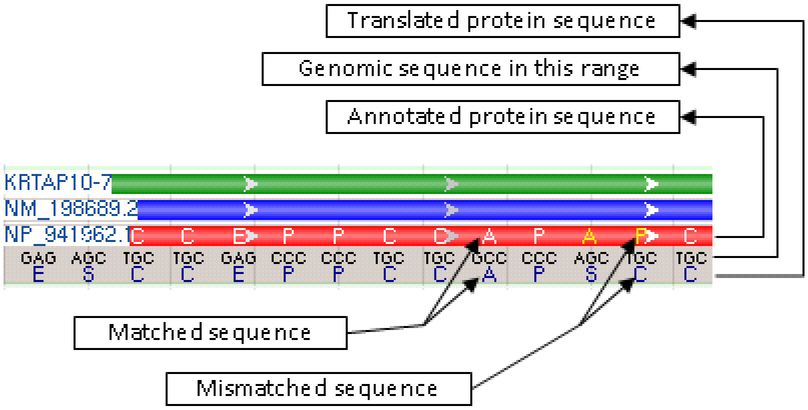
A Gene Model is a feature group that contains four main features: gene, mRNA, CDS and Exon. The group may also contain SNP and other features that are projected from mRNA and CDS products.

2.1 Gene Model Rendering

Rendering Options	Visual Examples
Show all	
Show all transcripts and CDSs, no gene bar	
Merge transcript and CDS pairs, no gene bar	
Merge all transcripts and CDSs, no gene bar	
Show on single line with exon structure	
Gene bar only	
With SNP features projected from mRNA and CDS products	
With other features projected from mRNA and CDS products	

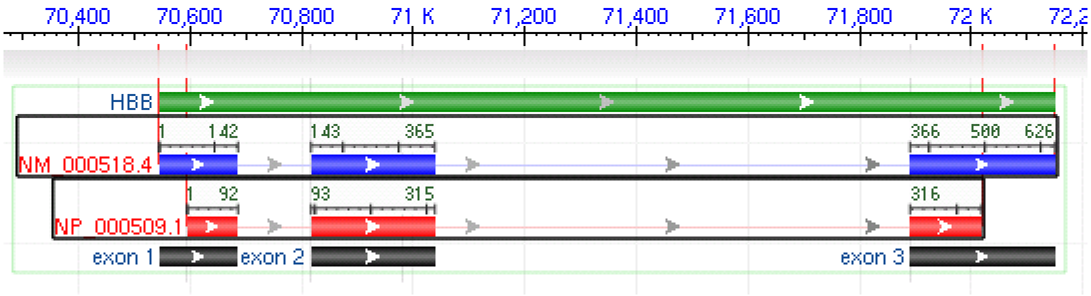
2.2 Special Rendering for CDS Features

When zoomed into the sequence level, both annotated protein sequence and translated protein sequence are shown for CDS features.



2.3 Feature Ruler

For a selected RNA or CDS feature, a feature ruler will be show with the feature's local coordinate.



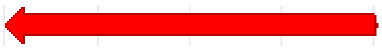
3 Clone Placement Features

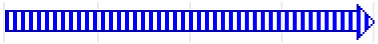
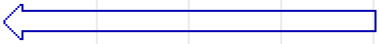
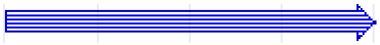
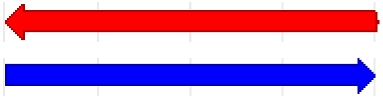
3.1 Communicated attributes

Graphical renderings for all clone placement features convey following attributes:

- Concordancy
- Uniqueness
- Clone end confidence
- Directionality, and
- Supporting evidence

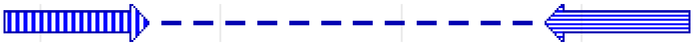
3.2 Visual examples for the conveyed attribute

Attribute	Possible values	Rendering	Visual Example
Concordancy	Concordant	Color: blue	
	Discordant	Color: red	
	Concordancy not set	Color: grey	
Uniqueness	Unique	Connecting line: solid	
	Multiple	Connecting line: Dotted	

	Uniqueness not set	Connecting line: dashed	
Clone end confidence	Unique	Fill: solid color	
	Multiple	Fill: vertical bars	
	Virtual	Fill: empty	
	Other/Not set	Fill: horizontal bars	
Directionality	Forward or backward	Arrow	
Supporting evidence	All non-prototype ends are 'supporting'	With no shaded background	
	Not all non-prototype ends are 'supporting'	With shaded background	

3.3 Rendering examples for various attribute combinations

The rendering is able to handle any combination of the five attributes shown above. Below are some rendering examples with various attribute combination.

Display	**Description**
	Unique, concordant, unique ends
	Multiple, concordant, one unique end, one multiple end
	Uniqueness-not-set, concordant, one multiple end, one confidence-not-set end
	Unique, concordant, one confidence-not-set end, one virtual end
	Unique, discordant, unique ends
	Multiple, discordant, one unique end, one multiple end
	Uniqueness-not-set, discordant, one multiple end, one confidence-

	not-set end
	Unique, discordant, one confidence-not-set end, one virtual end
	Unique, concordancy-not-set, unique ends
	Multiple, concordancy-not-set, one unique end, one multiple end
	Uniqueness-not-set, concordancy-not-set, one multiple end, one confidence-not-set end
	Unique, concordancy-not-set, one confidence-not-set end, one virtual end
	Multiple, discordant, one unique end, one multiple end, not all non-prototype ends are ‘supporting’
	Uniqueness-not-set, concordancy-not-set, one multiple end, one confidence-not-set end, not all non-prototype ends are ‘supporting’
	Unique, concordant, unique ends, not all non-prototype ends are ‘supporting’
	Clone with no end, no strand
	Clone with no end, plus strand
	Clone with no end, no strand, multiple, concordant not set
	Clone with no end, plus strand, discordant, unique not set

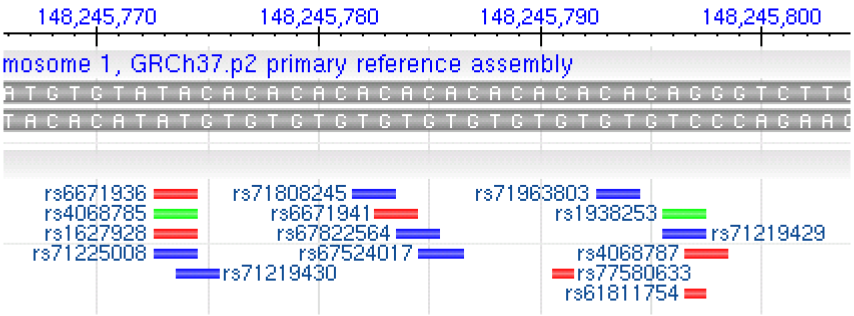
4 SNP Features

4.1 Color Code

Variation Type	**Color**

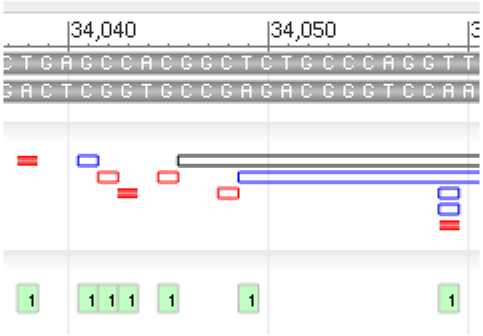
Single Nucleotide Polymorphism	Red
Deletion/Insertion Polymorphism	Blue
Heterozygous Variation, undefined at nucleotide level	Golden
Short Tandem Repeat (microsatellite) Polymorphism	Yellow
Named Variation (insertion/deletion polymorphism of named repetitive element)	Hunter Green
Sequence Scanned for Variation, but none observed	Black
Mixed Variation (cluster contains submissions from 2 or more allelic classes)	Green
MNP (multiple nucleotide polymorphism with alleles of common length > 1)	Gray

4.1.1 Visual Examples



4.2 Shape Code

A SNP can be represented by either a hollow or a solid rectangle. A solid rectangle means that this particular SNP has a weight of 1, and a hollow rectangle indicates a weight of 2 or more.



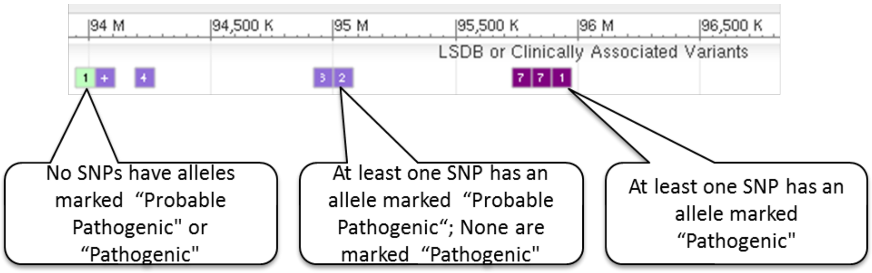
SNP Map weight info (the number of times a SNP maps to the genome contig (1-10))

1	hits genome once (on the same chromosome), annotated on NT_ contigs
2	hits genome twice, annotated on NT_ contigs with warning
3	hits genome 3-9 times, not annotated
10	hits 10+ times on genome, not annotated

(taken from SNP documentation at http://www.ncbi.nlm.nih.gov/snp)

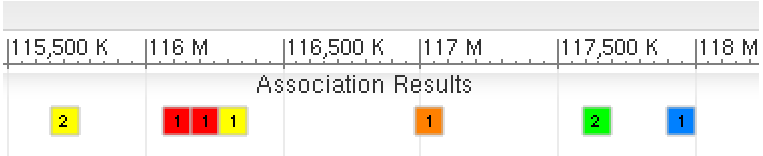
4.3 SNP Bins For Clinical Associations

Color	Description
Light Green	No SNPs in this bin have an allele marked “Probable Pathogenic” or “Pathogenic”
Light Purple	At least one SNP in this bin has an allele marked “Probable Pathogenic”; none are “Pathogenic”
Purple	At least one SNP in this bin has an allele marked “Pathogenic”



4.4 SNP Bins for Association Results

The color represents the highest p-value in that bin.



p-Value Range	<2	2-3	3-4	4-5	5-6	6-7	>7
Color	Teal	Sky Blue	Blue	Green	Yellow	Orange	Red

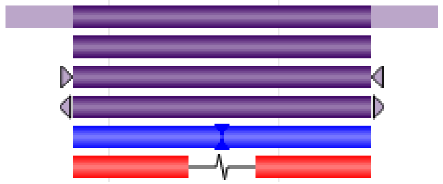
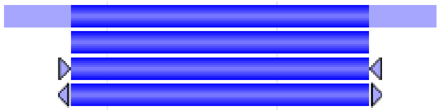
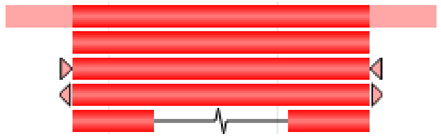
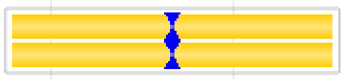
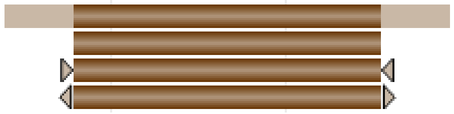
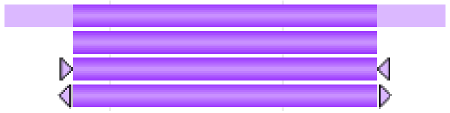
5 Structural Variants

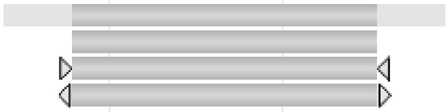
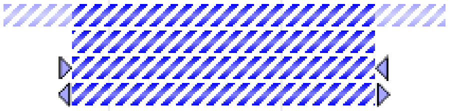
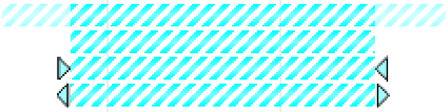
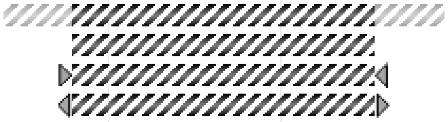
5.1 Common Rendering

There are four common scenarios for most variants (either SVs or SSVs) as shown in the table below. However, mixed cases with a defined breakpoint at one end and an undefined breakpoint range at the other end are possible as well. Here, we use copy number variants (CNV SV) as examples:

Breakpoint Type	**Rendering**	**Visual Examples**
With breakpoint resolution	Fully saturated color	
With defined breakpoint range	Transparent color for breakpoint ranges	
With undefined breakpoint, but known outer bound	Triangles pointing toward each other	
With undefined breakpoint, but known inner bound	Triangles pointing away from each other	

5.2 Variant Call Types (SSV) and Region Types (SV)

Type	**Comment**	**Visual Examples**
Copy number variation	Color: violet Four common cases, plus CNV with length of insertion, CNV with length of deletion (CNV SV)	
Copy number gain or Duplication	Color: blue (Gain SV or SSV)	
Copy number loss or Deletion	Color: red The last one is a loss variant with length of deletion (Loss SV or SSV)	
Mobile element insertion or Novel sequence insertion	Color: tangerine yellow (Insertion SV or SSV)	
Tandem duplication	Color: deep brown (Eversion SV or SSV)	
Inversion	Color: light violet (Inversion SV or SSV)	
Translocation	Color: light indigo with pattern (Translocation SV or SSV)	

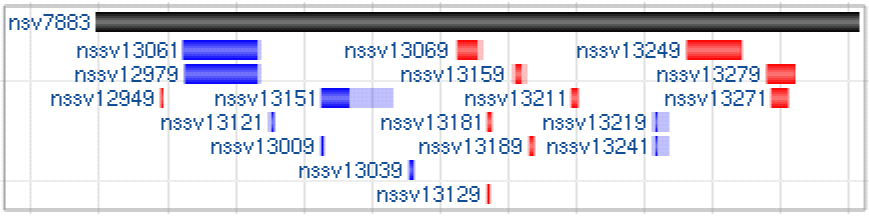
Complex	Color: black (Complex SV)	
Complex	Color: light azure (Complex SSV)	
Unknown	Color: grey (Unknown SV or SSV)	
Loss of Heterozygozity	Color: blue with pattern (LOH SV or SSV)	
Uniparental Disomy	Color: cyan with pattern (UPD SV or SSV)	
Other	Color: black with pattern (All other SV or SSV)	

Note:

1. SV Type “Copy number variation” can only have children of SSV Types “Copy number gain” and/or “Copy number loss” - in any combination. SV color remains the same (violet) in all such cases.
2. SV Type “Complex” can have either:
   - children all of SSV Type “Complex,” or
   - children of two or more SSV Types, in any combination (except “Copy number gain” and “Copy number loss,” which are covered above)

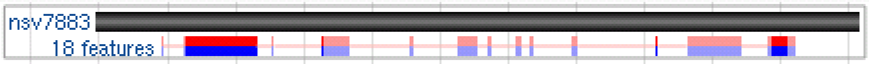
5.3 Rendering Styles for Linked Structural Variants Group

5.3.1 Default rendering with both parent and children shown

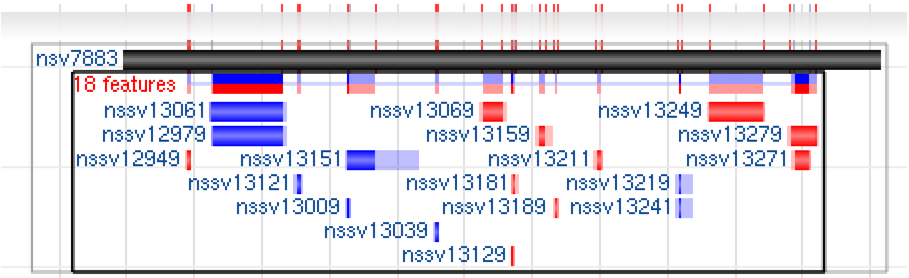


5.3.2 Rendering with supporting variants in a packed form

If there are multiple types in the supporting variants, multiple colors will be used to reflect the corresponding allele type.



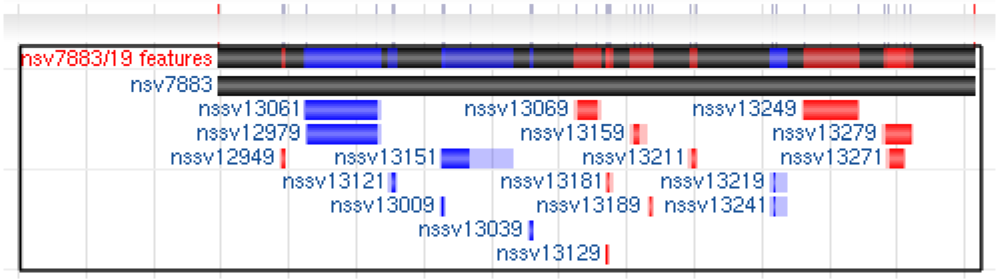
Click and select the packed feature bar to show all the supporting variants.



5.3.3 Superimpose all supporting variants over the parent variant

The supporting variants are superimposed on top of the parent variant with the shortest variants on the top. The colors reflect the corresponding allele type.

Click and select the packed feature bar to show all variants.



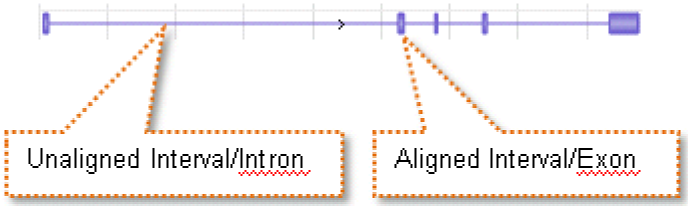
6 Segmental Duplications

Identity Attribute	Color	Example
> 99.0	Orange	
> 98.0	Yellow	
> 90.0	Grey	
<= 90.0	Black	

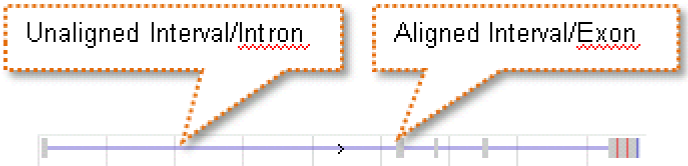
7 Alignments

7.1 Alignment in Different Mode

7.1.1 With Score Coloration Disabled



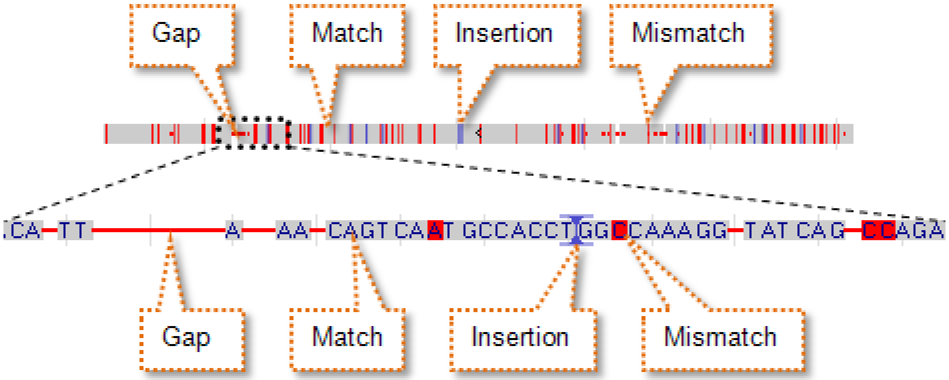
7.1.2 With Score Coloration Enabled



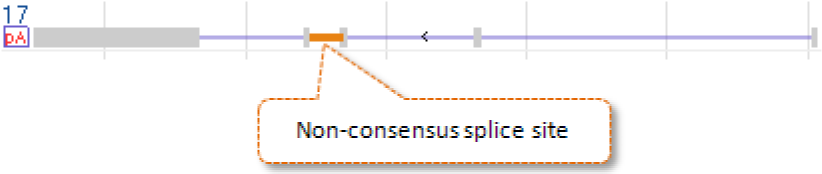
7.2 Alignment Score Coloration

There are four possible aligned-types: match, mismatch, gap (deletion), and insertion. The visual representations for all four different glyphs are illustrated in the table blow, and as well as in the screenshots.

Align-type	**Zoomed-out View**	**Zoomed-in View**
Match	Grey bar	Grey background
Mismatch	Red vertical bar	Red background
Gap	Red thin horizontal bar	Red thin horizontal bar
Insertion	Blue vertical bar	Blue hourglass with a bar on both top and bottom proportional to insertion bases



7.3 Non-consensus splice site

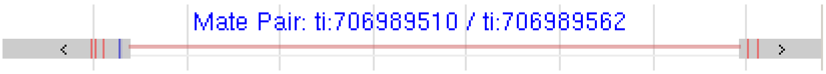


7.4 Unaligned tails

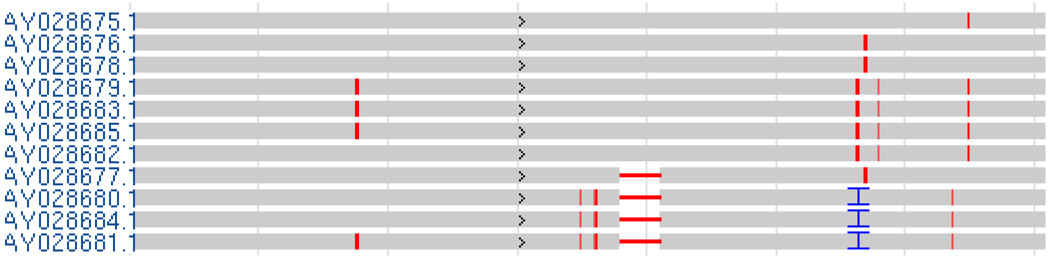
The numbers above the box show the number of unaligned bases.

Unaligned Tail Type	**Visual Examples**
PolyA	
Other	

7.5 Mate Pair Alignment



7.6 Multi-alignment

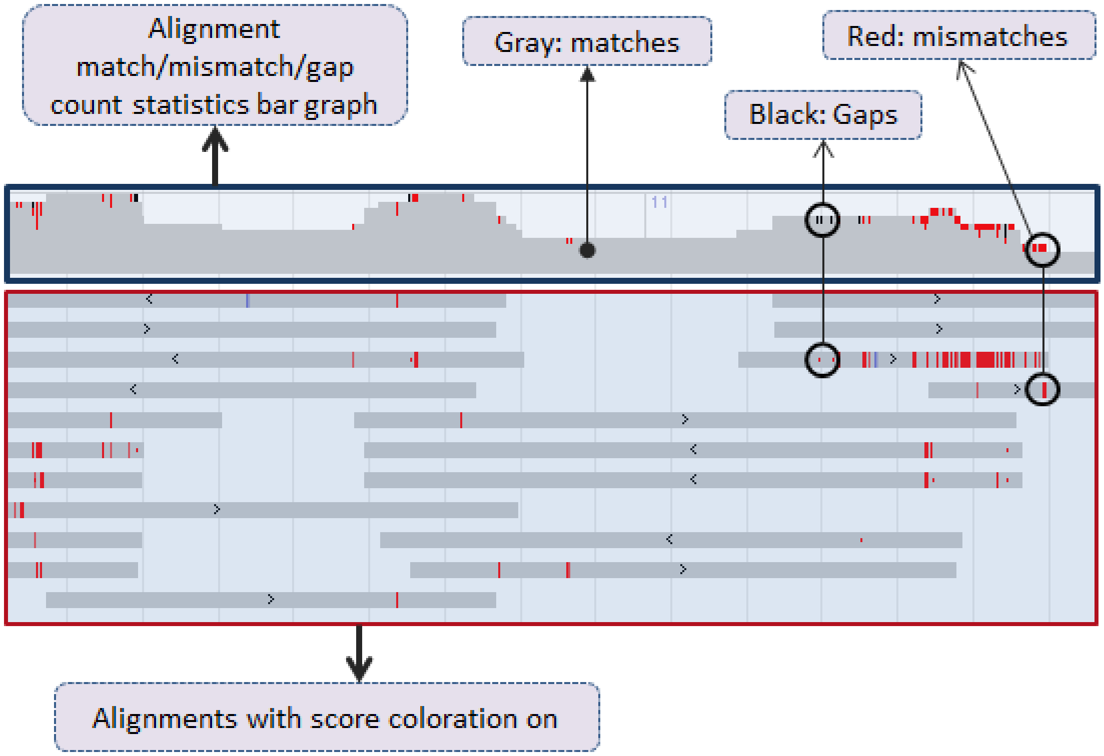


7.7 Smeared Alignment

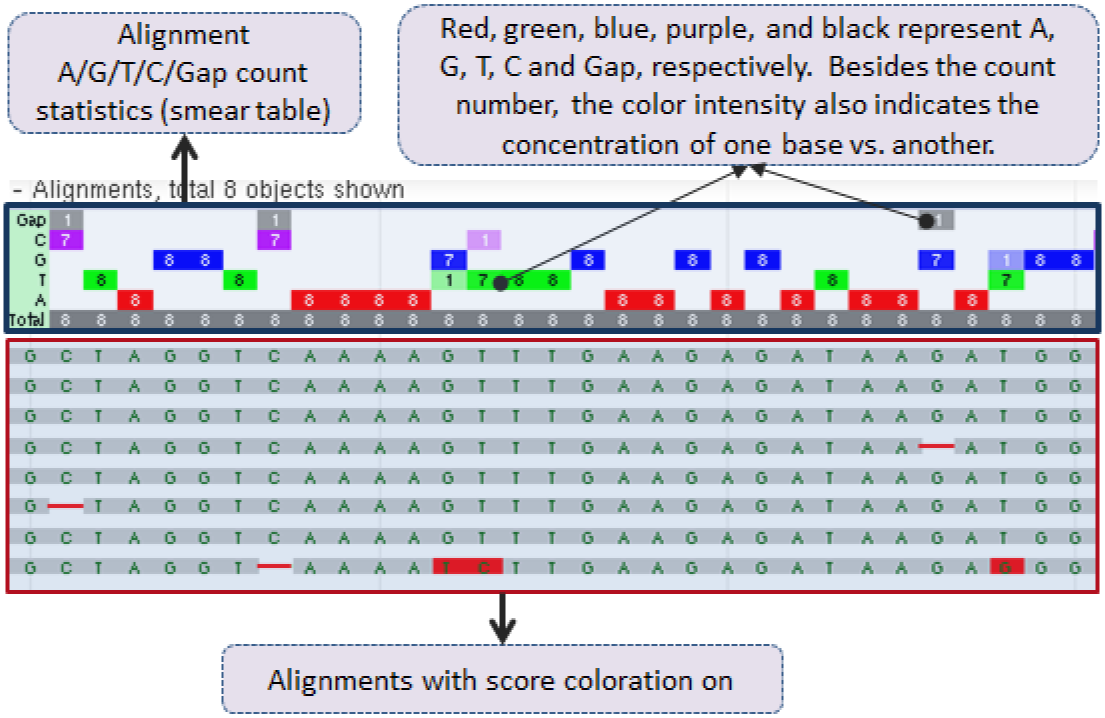
7.8 Alignment statistics

This option displays statistics at a given base position across all related alignments that have coverage at that base. The statistics include individual counts for A, G, T, C and Gap. Several other statistics can be derived with a known reference sequence base, including: matches, mismatches and total alignment count. There are eight different combinations of rendering with these three settings: Content (A/T/G/C/Gaps or Matches/Mismatches/Gaps), Data type (Count or Percentage), and Display (Bar graph or Smear table). Here are several examples.

7.8.1 Zoomed-out view of a bar graph with match/mismatch/gap count



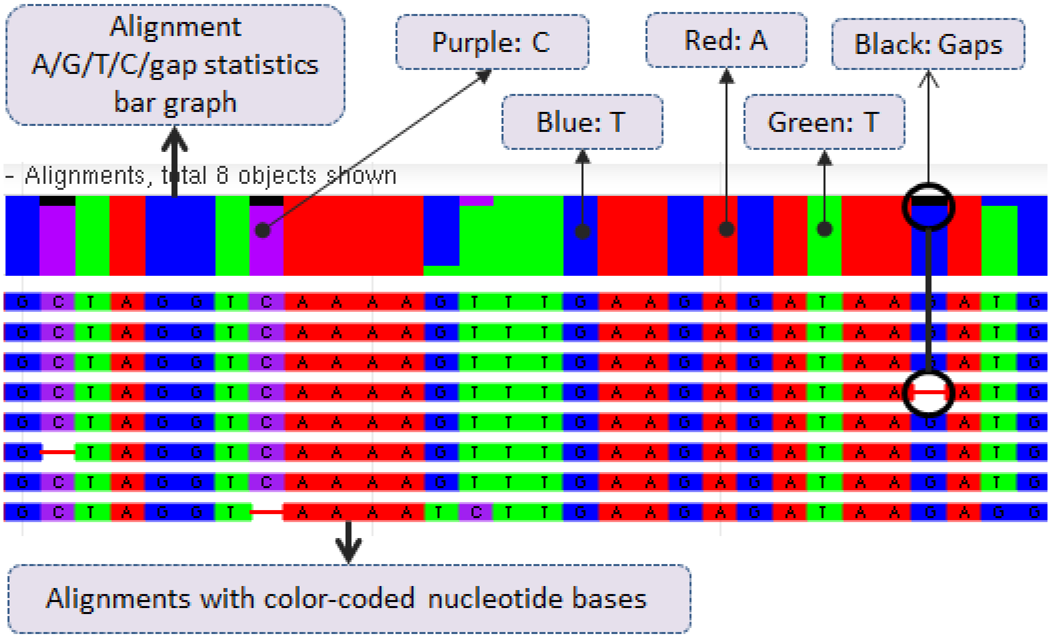
7.8.2 Zoomed-in view of a smear table with A/G/T/C/Gap count


Alignment A/G/T/C/Gap count statistics (smear table)

Red, green, blue, purple, and black represent A, G, T, C and Gap, respectively. Besides the count number, the color intensity also indicates the concentration of one base vs. another.

Alignments with score coloration on

7.8.3 Zoomed-in view of a bar graph with A/G/T/C/Gap count


Alignment A/G/T/C/gap statistics bar graph

Purple: C

Red: A

Black: Gaps

Blue: T

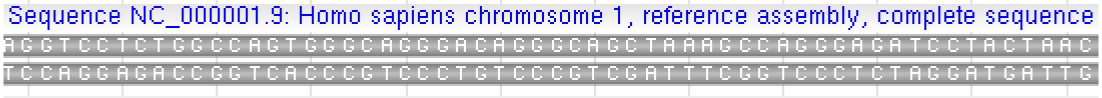
Green: T

Alignments with color-coded nucleotide bases

8 Sequence Track

The grey bar represents the sequence track in zoomed-out view

In zoomed-in view, both original sequence (top) and the complementary sequence (bottom) are shown.



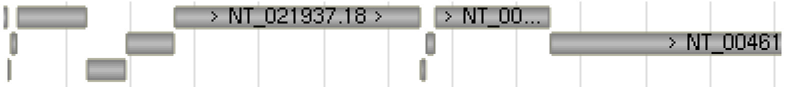
9 Segment Map

Depending on the sequence type, a sequence may have scaffold (contig) map, and/or tiling path (component map).

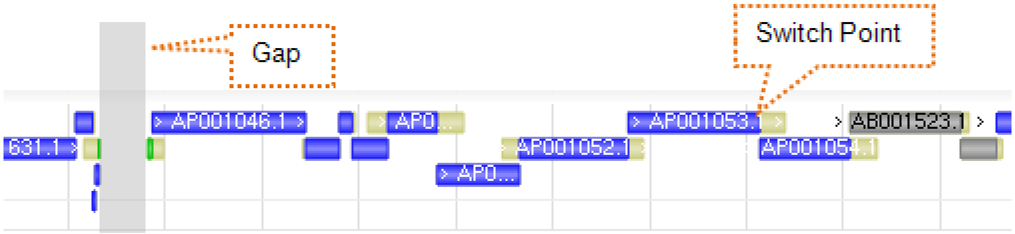
9.1 Segment Color Code

Segment Type	Finished	Draft	WGS	Other	Gap
Color	Blue	Orange	Green	Grey	Black

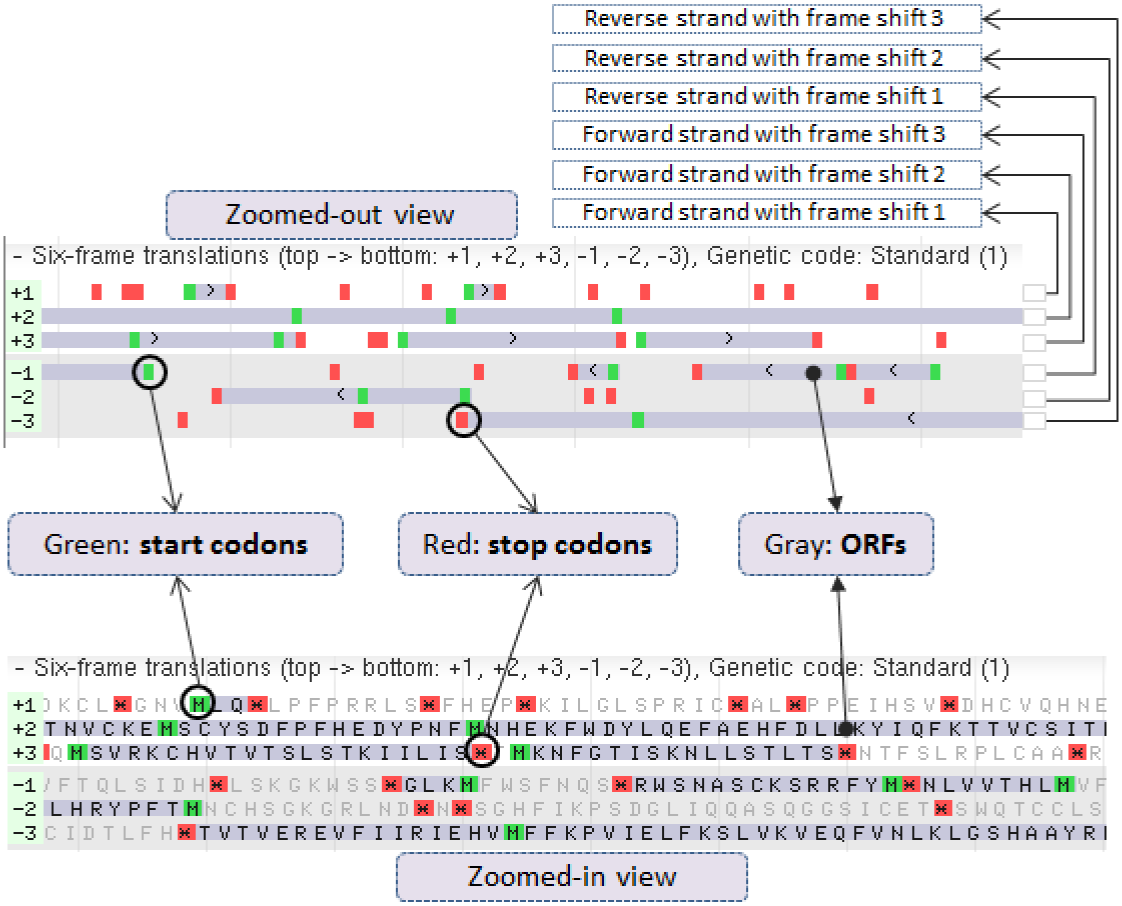
9.2 Scaffold Map Example



9.3 Tiling Path (Component Map) Example



10 Six-frame translations



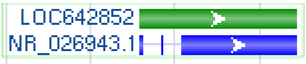
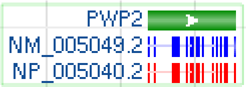
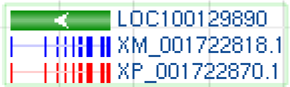
11 Label Placement

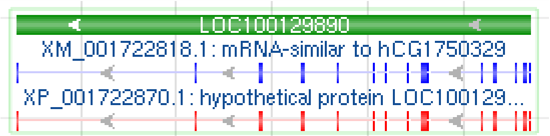
There are four global options regarding label placement: default, side label, top label, and no label. ‘Default’ may mean different settings for different objects. For example, default label placement for alignments is top labeling, but default setting for features is side labeling.

11.1 Side Label vs. Strand

In side labeling mode, the label is always placed at object’s 5’ side.

11.2 Examples

Label Placement	Visual Examples	
Default	Alignment (top):	
	Component (inside):	
	Features (side):	
Side Label		

Top Label	
No Label