

1 Generic 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

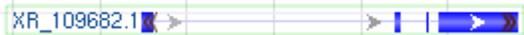
Settings	Visual Effect	Visual Examples
All features are shown in one gene group	Stripes over green gene bar	
Gene bar is hidden	Green stripe background	
Gene bar and gene label are hidden	Green stripe background	

1.2.2 Features with exception text

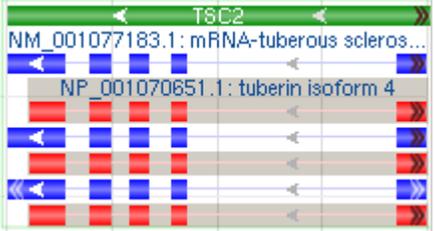
Exception Example	Visual Effect	Visual Examples
Mismatch in transcription	Shaded background	
Mismatch for translation	Shaded background	
Unclassified transcription discrepancy	Shaded background	

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	

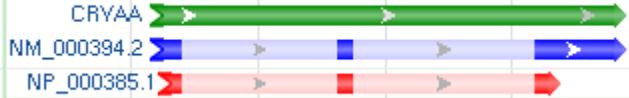
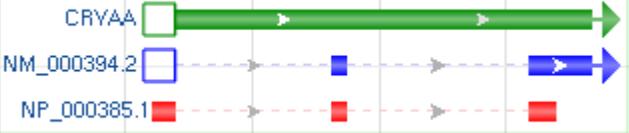
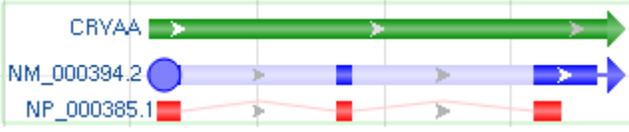
Partial start and stop	Black "<<" and ">>" at both ends	
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1.2.4 Features marked as partial

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

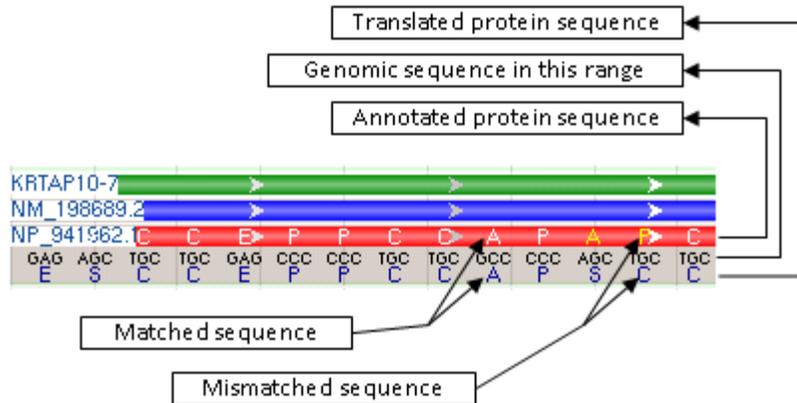
A Gene Model is a feature group that contains four main features: gene, mRNA, CDS and Exon. The group may also contain SNP and other features that are projected from mRNA and CDS products.

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	

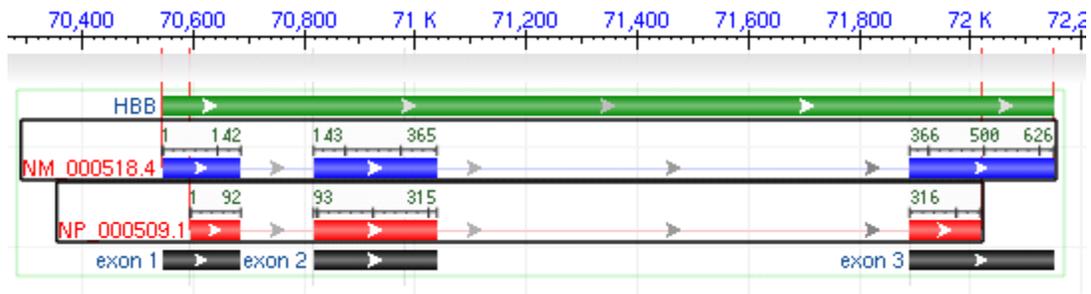
2.2 Special Rendering for CDS Features

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 shown with the feature's local coordinate.



3 Clone Placement Features

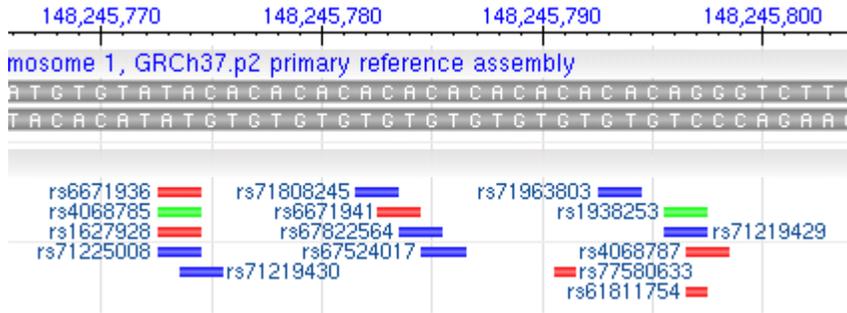


4 SNP Features

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

4.2 Visual Examples



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 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 Types

Variant Type	Color	Comment	Visual Examples
CNV	Black	Four common cases, plus CNV with all insertion SSVs, CNV with all Loss SSVs, CNV with mixed SSVs	
Other	Black with pattern	Four common cases	

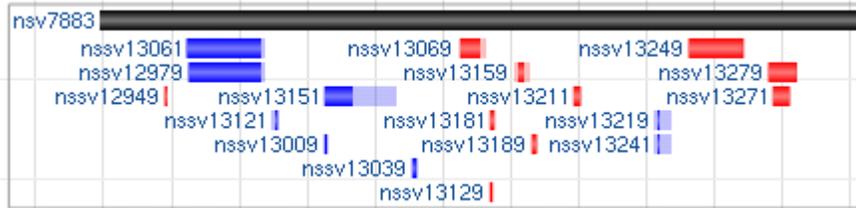
5.3 Allele Types

Note: the first variant shown in all screenshots in this section is the parent variant (either CNV SV or Other SV)

Allele Type	Variant Type	Color	Visual Examples
Gain	CNV	Red	
Loss	CNV	Blue Note: The last one is a Loss variant with allele_length	
Complex	CNV/Other	Light azure	
Unknown	CNV	Grey	
Insertion	CNV	Tangerine yellow	
Inversion	Other	Light violet with pattern	
Loss of Heterozygosity	Other	Blue with pattern	
Everted	Other	Deep brown with pattern	
Uniparental Disomy	Other	Cyan with pattern	
Translocation	Other	Light indigo with pattern	

5.4 Rendering Styles for Linked Structural Variants Group

5.4.1 Default rendering with both parent and children shown

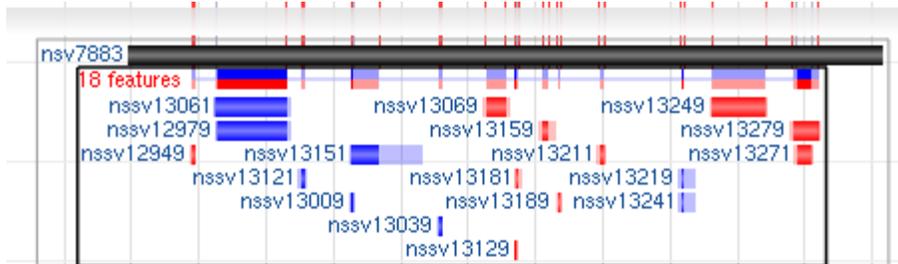


5.4.2 Rendering with supporting variants in a packed form

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.

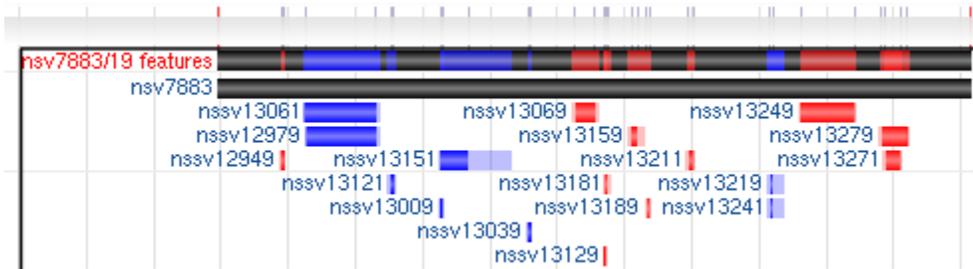


5.4.3 Superimpose all supporting variants over the parent variant

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.



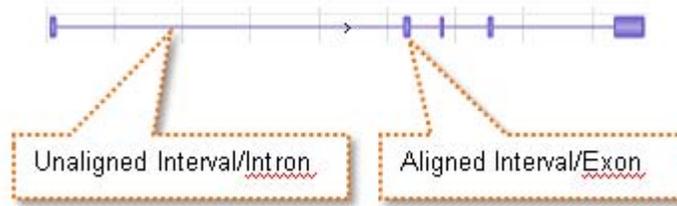
6 Segmental Duplications

Identity Attribute	Color	Example
> 99.0	Orange	$\hat{identity}=99.19$
> 98.0	Yellow	$\hat{identity}=98.20$
> 90.0	Grey	$\hat{identity}=91.19$
≤ 90.0	Black	$\hat{identity}=0.00$

7 Alignments

7.1 Alignment in Different Mode

7.1.1 With Score Coloration Disabled



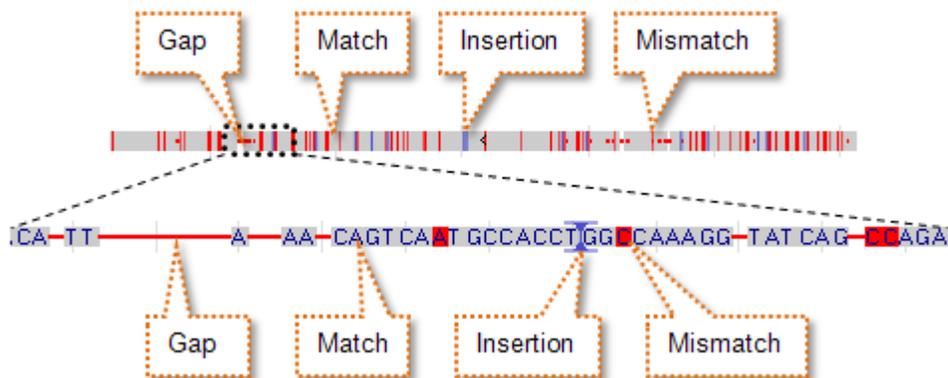
7.1.2 With Score Coloration Enabled



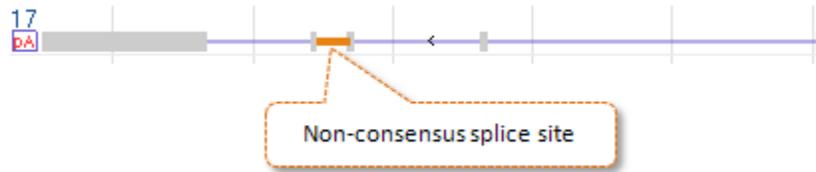
7.2 Alignment Score Coloration

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 below, 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

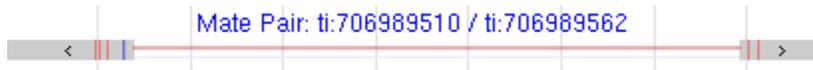


7.4 Unaligned tails

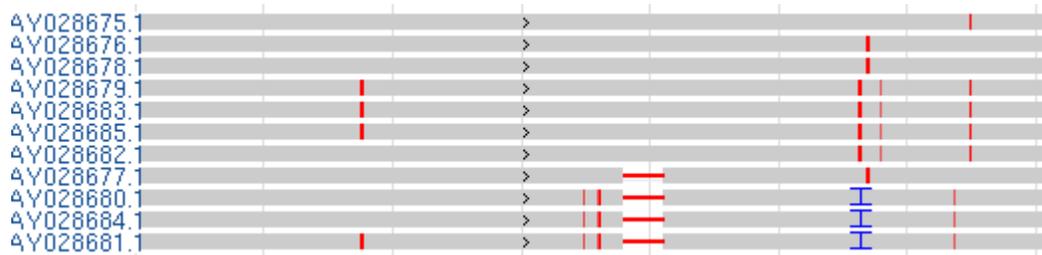
The numbers above the box show the number of unaligned bases.

Unaligned Tail Type	Visual Examples
PolyA	
Other	

7.5 Mate Pair Alignment



7.6 Multi-alignment



7.7 Smearred Alignment



8 Sequence Track

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

[Sequence NC_000001.9: Homo sapiens chromosome 1, reference assembly, complete sequence](#)

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

[Sequence NC_000001.9: Homo sapiens chromosome 1, reference assembly, complete sequence](#)

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AGGTCCCTCTGGCCAGTGGGCAGGGACAGGGGCAGCTAAAGCCAGGGAGATCCTACTAAC
TCCAGGAGACCCTCACCCTCCCTCTCCCGTCGATTTCCGGTCCCTCTAGGATGATTG

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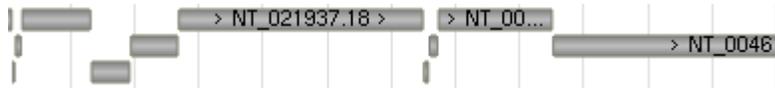
9 Segment Map

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

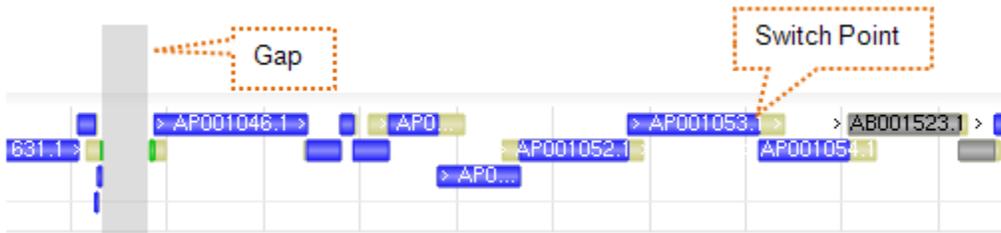
9.1 Segment Color Code

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 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.

10.1 Side Label vs. Strand

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

10.2 Examples

Label Placement	Visual Examples
Default	Alignment (top):
	Component (inside):
	Features (side):
Side Label	
Top Label	
No Label	