Using the genome browser in DECIPHER

The Genoverse browser (http://genoverse.org/) is built into DECIPHER and offers interactive visualisation of patient genomic variants on the reference genome, with additional tracks that display information to assist variant interpretation. These additional tracks include information about affected genes, overlapping variants from population databases such as gnomAD, overlapping variants from other DECIPHER patients and overlapping variants from other variant disease databases such as ClinVar. This short guide will help you understand the basic functions in the browser.

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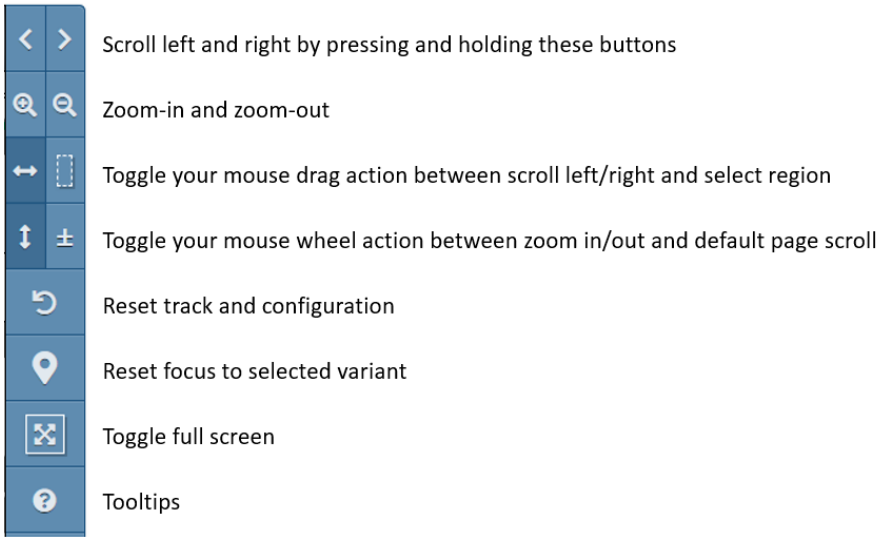
Navigating the browser

There is a navigation menu on the right-hand side of the browser. These controls are shown below.



The "?" button shows tooltips, which provide help on the buttons and layout of the browser.

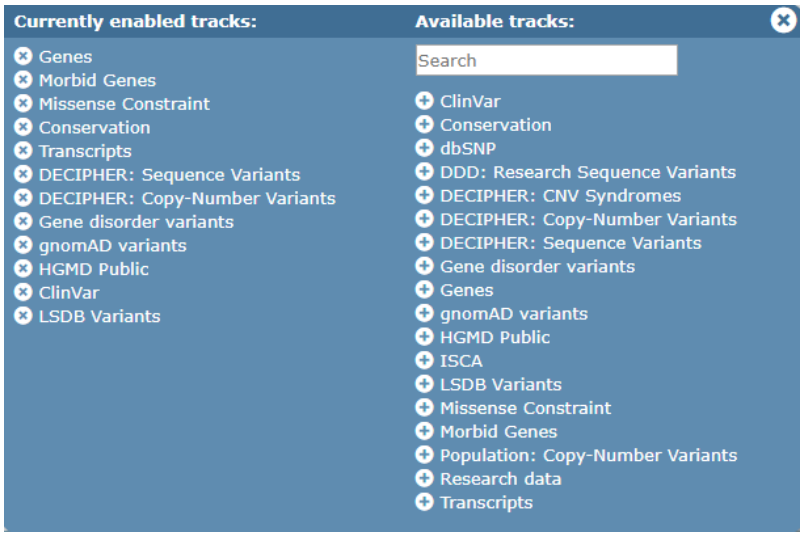
Customising the Browser

The browser has many data tracks available. Different tracks are shown by default for different variant classes.

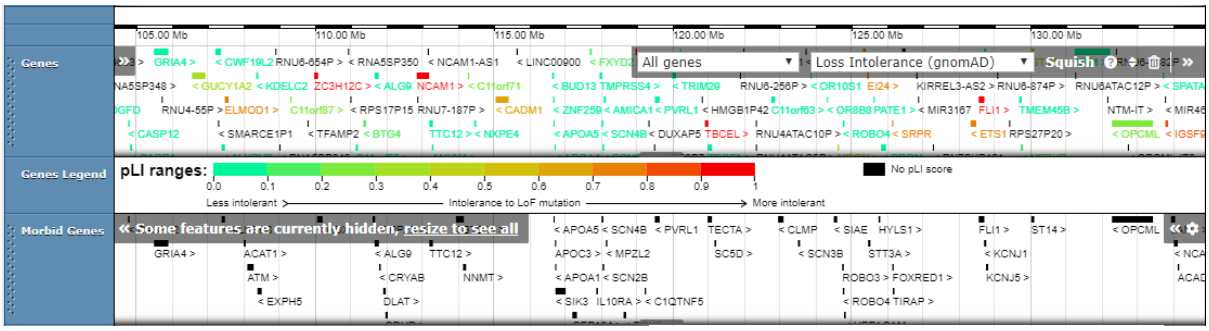
It is possible for users to customize the browser. For registered users that are logged into DECIPHER, the customised view is saved. To see all the tracks that are available, click on the "Tracks" button at the top left-hand side of the genome browser (shown in red below).

A popup will appear that allows the customisation of the browser (as shown below). On the left-hand side are the currently enabled tracks, on the right-hand side are the available tracks. To delete tracks, click on the "x" icon next to the relevant track in the currently enabled tracks list. To add tracks, click on the "+" icon next to the relevant track in the available tracks list. It is possible to have

duplicated tracks. This is useful to view the same subset of data using different filters, for example, duplicating the Genes track will allow developmental disorder genes and genes coloured by pLI scores to be visualised at the same time. It is also possible to reorder the currently enabled tracks in this list by dragging and dropping the track in the preferred location.



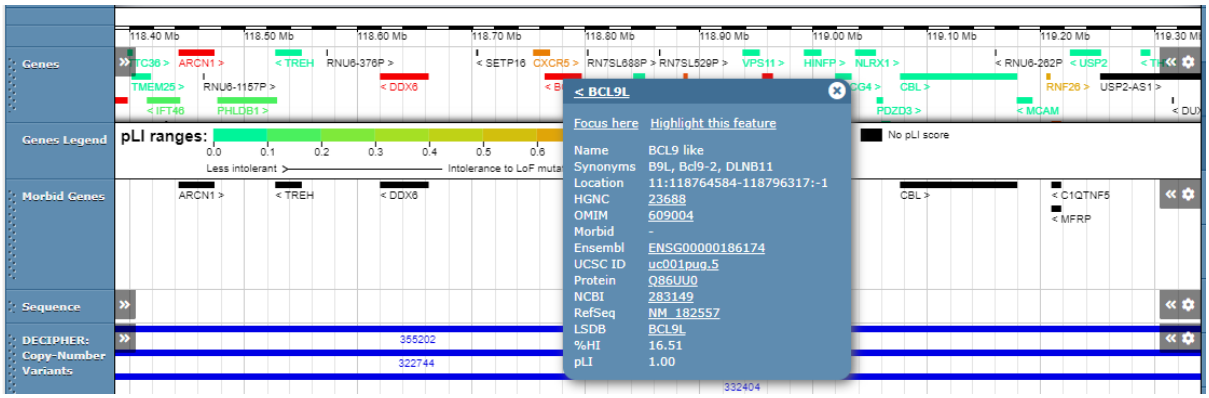
The left and right edges of the tracks provide information regarding the tracks (as shown below). The left edge is used for informational messages such as “Some features are currently hidden, resize to see all”. These messages may be dismissed by clicking on the “<<”. The right edge used to provide information about the tracks and track filters. Clicking on the “<<” allows information about the track and filters to be seen. The screenshot below shows the filters available for the “Genes” track. Information about the track can be seen by clicking on the “?” icon. The track can be removed from view by clicking on the rubbish bin icon. The up-down arrow icon can be used to set the track to a fixed height, or to auto-adjust the height of the track.



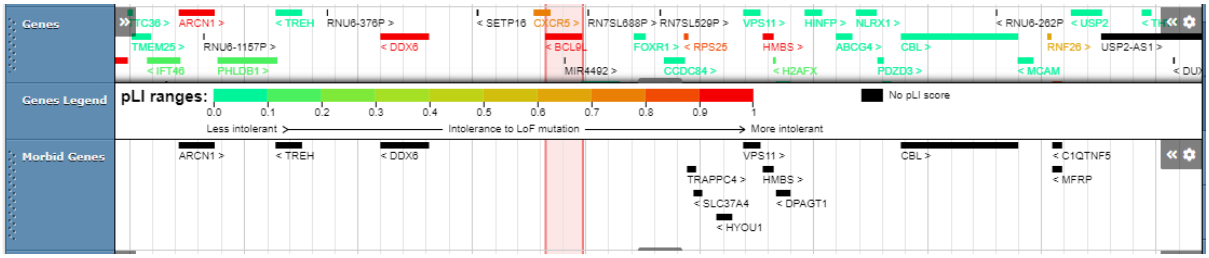
There is also a “Squish” option, which condenses the display of features on the track vertically (which is helpful when there are many stacked features), as shown below. Once the data is squished, there is an “Unsquish” option, which returns the track to the original view.

Focusing on and Highlighting features

Sometimes a feature on the genome browser (such as a gene or variant) is either too small at the current zoom level to see in relation to other features, or so large that it cannot all be seen. To adjust the view to centre on the feature and zoom in or out accordingly, click on the feature and select “Focus here” from the popup menu.



It is often necessary to read down multiple tracks, for example to see which variants overlap a gene. To help with this, there is a “Highlight this feature” option (as shown above), which will show the position of the relevant feature in all of the tracks. In the example below, the gene BCL9L has been highlighted. This is especially helpful when many tracks are enabled.



Karyotype browser

A static karyotype browser is shown when information is shown over multiple chromosomes, such as for some search results. This shows the distribution of variants across the genome. It is possible to access more information about individual variants by clicking on the histogram or bar charts.