

# 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.

<	>	Scroll left and right by pressing and holding these buttons
ବ୍	Q	Zoom-in and zoom-out
¢		Toggle your mouse drag action between scroll left/right and select region
1	±	Toggle your mouse wheel action between zoom in/out and default page scroll
E.	D	Reset track and configuration
9	?	Reset focus to selected variant
2	<	Toggle full screen
(	3	Tooltips

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 lefthand 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.

Currently enabled tracks:	Available tracks:
😢 Genes	Search
😣 Morbid Genes	
😣 Missense Constraint	🕂 ClinVar
× Conservation	📀 Conservation
× Transcripts	📀 dbSNP
S DECIPHER: Sequence Variants	📀 DDD: Research Sequence Variants
8 DECIPHER: Copy-Number Variants	DECIPHER: CNV Syndromes
8 Gene disorder variants	DECIPHER: Copy-Number Variants
🗴 gnomAD variants	DECIPHER: Sequence Variants
× HGMD Public	🕀 Gene disorder variants
🗴 ClinVar	🛨 Genes
🗴 LSDB Variants	📀 gnomAD variants
	🕂 HGMD Public
	🛨 ISCA
	🕂 LSDB Variants
	🛨 Missense Constraint
	🛨 Morbid Genes
	Population: Copy-Number Variants
	Research data

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 tracks and track filters. Clicking on the "<<" allows information about the 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.

	105.00 Mb	110.00 Mb	115.00 Mb	120.00 Mb	125.00 Mb	130.00 Mb	
Genes	13)3 > GRIA4 > •	< CWF19L2 RNU6-654P > < RN	A5SP350 < NCAM1-AS1 < LINC00900 <	FXYD2 All genes	Loss Intolerance (gnon	mAD) ▼ <b>TSquish</b>	?RI\$16-∰81 P ≫
1444	NA5SP348 > < GU	ICY1A2 < KDELC2 ZC3H12C >	< ALG9 NCAM1 > < C11orf71 < BUD13		256P > < OR10S1 EI24 > KIRREL3	-AS2 > RNU6-874P > RNU6	
1777.F	CASP12	< SMARCE1P1 < TFAMP2 < I	TG4 TTC12 >< NXPE4 < APOA5	< SCN4B < DUXAP5 TBCEL > RNI	U4ATAC10P >< ROBO4 < SRPR	< ETS1 RPS27P20 >	< OPCML < IGSF
		BULLABALA AL		ART FRAME RI			
Genes Legend	pLI ranges:	0 0.1 0.2 0	3 0.4 0.5 0.8 0.7	0.8 0.9 1	No pLI score		
Genes Legend	pLI ranges: 0.0 Le	0 0.1 0.2 0 ess intolerant >	3 0.4 0.5 0.8 0.7 Intolerance to LoF mutation —	0.8 0.9 1	e intolerant		
Genes Legend Morbid Genes	pLI ranges: UI Le «Some featu	0 0.1 0.2 0 sss intolerant > ires are currently hidd	3 0.4 0.5 0.8 0.7 Intolerance to LoF mutation — en, resize to see all < APOA5	0.8 0.9 1 Mon < SCN4B < PVRL1 TECTA >	e intolerant < CLMP < SIAE HYLS1 >	FLI1> ST14>	< OFCML «
Genes Legend	pLI ranges: 0.0 Le Come featu GR(A4>	0 0.1 0.2 0 ess intolerant > irres are currently hidd ACAT1 >	3 0.4 0.5 0.8 0.7 Intolerance to LOF mutation — en; resize to see all <alg9 ttc12=""> APOG3 &gt; CQ2VAB NUMT &gt; APOG3 &gt;</alg9>	0.8 0.9 1 Mon <scn48 <pvrl1="" tecta=""> <mpzl2 sc5d=""> <scn49< th=""><th>CLMP &lt; SIAE HYLS1&gt; SOUTH STAR</th><th>FLI1&gt; ST14&gt; <kcnj1< th=""><th>&lt; OPCML</th></kcnj1<></th></scn49<></mpzl2></scn48>	CLMP < SIAE HYLS1> SOUTH STAR	FLI1> ST14> <kcnj1< th=""><th>&lt; OPCML</th></kcnj1<>	< OPCML

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.

Genes	1	 _	 	 	-	- 1	-11		Γ.		 		 - 14	-	:	 -	•••	•	« ¢
in and												-							



## 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.

Genes	TC38>     ARCN1 >     < TREH     RNU6-376P >       TMEM25 >     RNU6-1157P >     < DDX6       < IFT46     PHLDB1 >	< <tr>     &lt; SETP16     CCC65&gt;     RN75L088P × RN75L020P &gt;     VPS11&gt;     HNFP &gt;     NLRX1&gt;       &lt; SEC.P.     FOXR1 &gt; &lt; RPS25     HMS5 &gt;     ABCG4 &gt;     CBL&gt;       MR4492 &gt;     CCDC64 &gt;     &lt; H2AFX     PDZD3 &gt;</tr>	< RNU8-282P < USP2 < TK < RNF28 > USP2-AS1 > < MGAM < DU3
Genes Legend	pLI ranges: 0.0 0.1 0.2 0.3 0.4 Less intolerant > Int	05 06 0.7 0.8 0.9 1 VopLIscore 1 Verance to LoF mutation→ More intolerant	
Morbid Genes	ARON1 > < TREH < DDX8	VP511 > CBL > TRAPPC4 > HMB5 > <slc37a4 0pagt1<br="" <=""><hyqu1< th=""><th><cioines ¢<br="" «=""><merp< th=""></merp<></cioines></th></hyqu1<></slc37a4>	<cioines ¢<br="" «=""><merp< th=""></merp<></cioines>

#### **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.

