Ensembl Walk-Through
9 May 2009

VARIATIONS WORKED EXAMPLE

In this worked example we will explore information with regard to the PTPN22 (Tyrosine-protein phosphatase non-receptor type 22) gene with a focus on variation. Note that this worked example only covers a small amount of all the information available in Ensembl!

Note: This worked example is based on Ensembl version 54 (March 2009). After in future a new version has gone live, version 54 will still be available through http://May2009.archive.ensembl.

- Go to http://www.ensembl.org.

(1) Searching for the human PTPN22 gene:

- Enter 'human PTPN22 gene' in the text box under 'Search Ensembl'.
- Click [Go].
- Click on 'Ensembl protein_coding Gene: ENSG00000134242 (HGNC (curated): PTPN22)' on the page with search results.

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(2) Genomic sequence of the PTPN22 gene:

- Click on 'Sequence' in the side menu.

This page, as well as many others, can be customised using the 'Configure this page' link in the side menu.

To add variations to the display:

- Click on 'Configure this page' in the side menu.
- Select 'Show variations: Yes and show links'.
- Click [SAVE and close].

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(3) Spliced sequence of the PTPN22-001 transcript:

- Click on 'ENST00000359785' in the list of transcripts.
- Click on 'Sequence - cDNA' in the side menu.

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(4) Unspliced sequence of the PTPN22-001 transcript:

- Click on 'Sequence - Exons' in the side menu.

To show the full intronic sequence and 1000 basepairs of flanking sequence:

- Click on 'Configure this page' in the side menu.
- Enter '1000' in the text box behind 'Flanking sequence at either end of transcript:'.
- Check the box behind 'Show full intronic sequence'.
- Click [SAVE and close].

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(5) Cross references to other databases for the PTPN22-001 transcript:

- Click on 'External References - General identifiers' in the side menu.

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(6) Genomic region around the PTPN22 gene:

- Click on the 'Location: 1:114,157,963-114,215,857' tab.
- Zoom out one step in the third panel using the zoom tool in the upper right hand corner of the panel.

To add variations and DECIPHER and DGV tracks to the display:

- Click on 'Configure this page' in the side menu.
- Enter 'variation' in the text box behind 'Search display' in the pop-up screen.
- Select 'All variations – Normal'.
- Enter 'decipher' in the text box behind 'Search display'.
- Select 'DECIPHER - Normal'.
- Enter 'dgv' in the text box behind 'Search display'.
- Select 'DGV loci - Normal'.
- Click [SAVE and close].

DECIPHER and DGV are DAS (Distributed Annotation System) sources; these data are not stored in the Ensembl database, but elsewhere. Ensembl is in this case only used as a means to display these data.

Clicking on a feature on this page will give a pop-up menu with information about the feature. Often the pop-up menu will also contain one or more links to pages with more detailed information.

To get more information and go to the original DECIPHER entry for a DECIPHER feature:

- Click on the DECIPHER feature.
- Click on the link in the pop-up menu that links to the DECIPHER website.
- Go back to Ensembl by using the back button of the internet browser.

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(7) Variations in the PTPN22 gene:

- Click on the 'Gene: PTPN22' tab.
- Click on 'Genetic Variation - Variation Table' in the side menu.

By default all variations that are located in the exons are shown as well as those that are located in the introns and flanking sequence and are within 100 bp from an exon.

To show all variations in exons, introns and within 5000 bp up- and downstream of the 5' and 3' end of the transcripts:

- Click on 'Configure this page' in the side menu.
- Select 'Context: Full Introns'.
- Click [SAVE and close].

To show only non-synonymous variations:

- Click on 'Configure this page' in the side menu.
- Deselect all options under 'Select Variation Type' except 'Non-synonymous'.
- Click [SAVE and close].

- Click on 'Genetic Variation - Variation image' in the side menu

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(8) Variations in the PTPN22-001 transcript in different individuals:

- Click on the 'Transcript: PTPN22-001' tab.
- Click on 'Genetic Variation – Population comparison' in the side menu.

By default only data for Watson and Venter are shown.

To add other individuals:

- Click on 'Configure this page' in the side menu.
- Select all options under 'Select Individuals'.

- Click [SAVE and close].

- Click on 'Genetic Variation - Comparison image' in the side menu.

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(9) Variations in the PTPN22-001 protein:

- Click on 'Protein Information – Variations' in the side menu.

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(10) Detailed information on variation rs2476601:

- Click on 'SNP ID rs2476601' in the list of variations.
- Click on 'Gene/Transcript' in the side menu.
- Click on 'Population genetics' in the side menu.
- Click on 'Individual genotypes' in the side menu.
- Click on 'Phenotype data' in the side menu.