

Diagnostic component (TEAM)

Access

TEAM is available at the following address : <http://omics.it4i.cz/team/>

The address is accessible only via VPN.

Diagnostic component (TEAM)

VCF files are scanned by this diagnostic tool for known diagnostic disease-associated variants. When no diagnostic mutation is found, the file can be sent to the disease-causing gene discovery tool to see whether new disease associated variants can be found.

TEAM >(27) is an intuitive and easy-to-use web tool that fills the gap between the predicted mutations and the final diagnostic in targeted enrichment sequencing analysis. The tool searches for known diagnostic mutations, corresponding to a disease panel, among the predicted patient's variants. Diagnostic variants for the disease are taken from four databases of disease-related variants (HGMD-public, HUMSAVAR, ClinVar and COSMIC) If no primary diagnostic variant is found, then a list of secondary findings that can help to establish a diagnostic is produced. TEAM also provides with an interface for the definition of and customization of panels, by means of which, genes and mutations can be added or discarded to adjust panel definitions.

*Figure 5. **Interface of the application. Panels for defining targeted regions of interest can be set up by just drag and drop known disease genes or disease definitions from the lists. Thus, virtual panels can be interactively improved as the knowledge of the disease increases.*

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The screenshot displays the TEAM application interface. At the top, there's a 'TEAM' header and a 'Sample Data' section with search filters. The main area shows 'Diagnostic mutations' with a table of results. A black arrow points from the 'Diagnostic mutations' table to a text box that says 'If no diagnostic variants appear, then secondary findings are studied'. To the right, there's a 'Tool for defining panels' section with a diagram showing a flow from a list of genes to a panel definition. A green funnel icon is placed over the 'Tool for defining panels' section, with a text box that says 'New filter based on local population variant frequencies'. The URL <http://team.babelomics.org> is visible at the bottom left.

TEAM

Sample Data

Search

Panel: Panel Results Page 1

VCF File: C:\Users\patient_1\... (Browse...)

Butt Reset

Results

Diagnostic Secondary findings

Chromosome Position SNP ID Ref Alt Gene Conses. Type Phenotype Source SPT PolyPhen

1 1 18247534 - T C - exon_variant_18247534 - absp_1 -

2 3 18247534 - T C - RND exon_variant_18247534 - RND -

3 3 18247534 - T C - RND exon_variant_18247534 - RND -

4 3 18247534 - T C - RND exon_variant_18247534 - RND -

If no diagnostic variants appear, then secondary findings are studied

Tool for defining panels

Results

Diagnostic Secondary findings

Chromosome Position SNP ID Ref Alt Gene Conses. Type Phenotype Source SPT PolyPhen

5 2 182415239 - A G - CEBVL intron_variant_182415239 - -

6 2 182415239 - A T - CEBVL intron_variant_182415239 - -

7 2 182415239 - G A - CEBVL intron_variant_182415239 - -

8 2 182415239 - T C - CEBVL intron_variant_182415239 - -

9 4 47953515 - A T - CHGA1 intron_variant_47953515 - -

10 16 57937898 - T C - CHGB1 57937898 - -

11 16 57937898 - G C - CHGB1 57937898 - -

12 16 57937898 - G A - CHGB1 57937898 - -

13 16 57937898 - G A - CHGB1 57937898 - -

New filter based on local population variant frequencies

<http://team.babelomics.org>

Figure 1: Interface of the application. Panels for defining targeted regions of interest can be set up by just drag and drop known disease genes or disease definitions from the lists. Thus, virtual panels can be interactively improved as the knowledge of the disease increases.