

# Interpreting Prioritization Results

1. After prioritization the results are received in tab separated format with **RANK, RANK-RATIO, GENE, SCORE, EXPLORE** as shown in Table in left side (below)
2. On the right, is the heat map plot of functional annotation matrix X which was used to predict the corresponding phenotypic concordance scores.

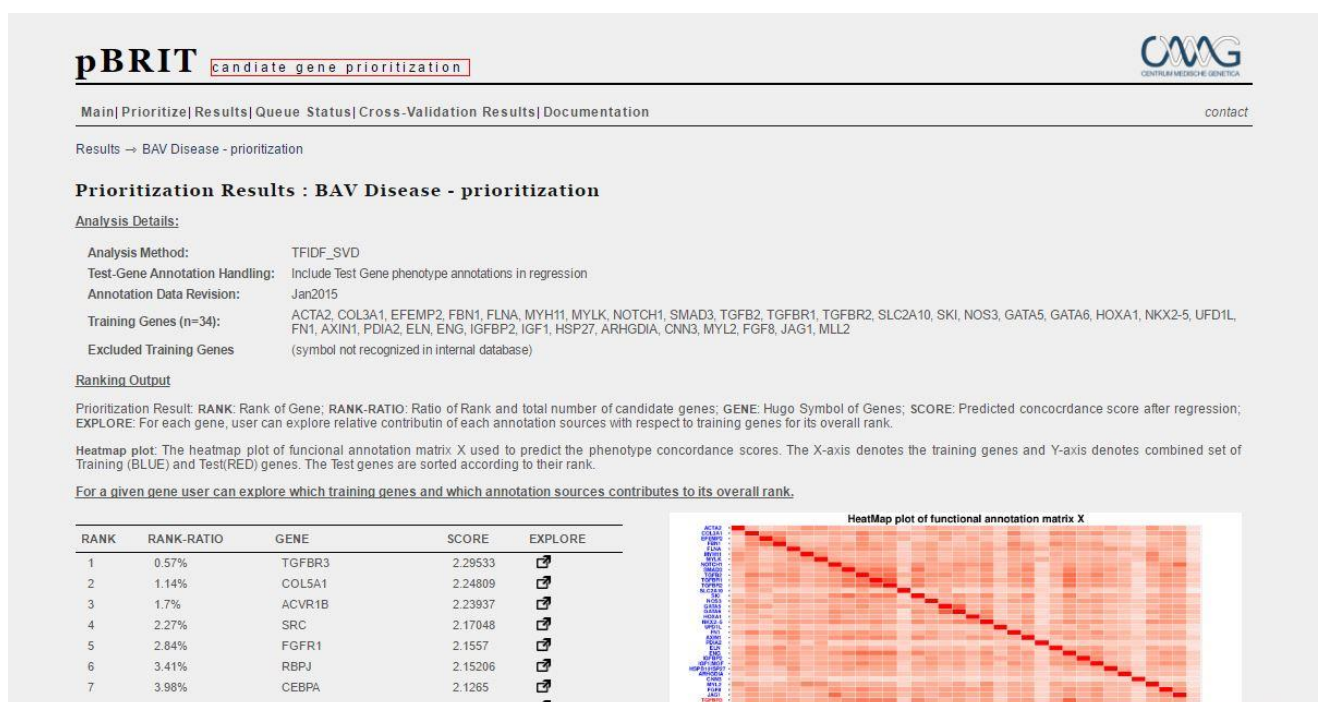


Figure 1: Example prioritization of BAV disease genes

## Heatmap plots of functional annotation Matrix.

1. The heat map plot of functional annotation matrix (combination of Training gene and Test Genes) is sorted according to the rank of candidate genes. Training genes are denoted in blue and Test genes are marked red.

2. These heatmap plots show why certain genes are ranked higher corresponding to the given training genes.
3. Additionally, for each of the genes user can explore which annotation source and training gene contributed the most for its prioritization. This can be done by using the link for each gene under “**EXPLORE**” heading.
4. Exploration of annotation sources contribution can be found in Figure 3.

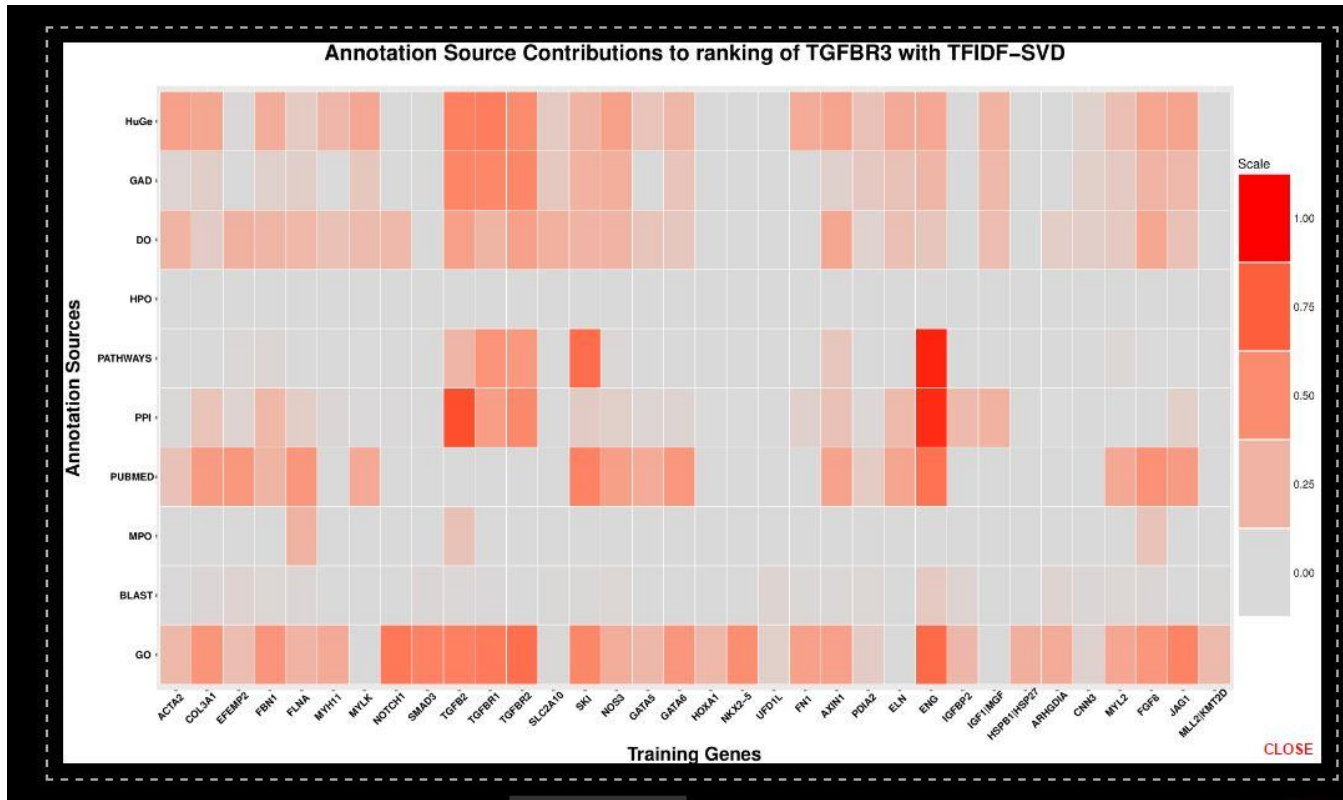


**Figure 2:** Heatmap plot of functional annotation matrix. The test genes are sorted according to their rank in the final output.

## Contribution of Annotation sources and training genes in prioritization

1. Using the link in the “EXPLORE” for *TGFBR3* gene one can explore the contribution of annotation sources ( Y –axis) with respect to given set of training genes (X-axis). (**Figure 3**)
2. In this example, *TGFBR3* gene is ranked higher, because it has high similarity to training genes: *TGFB2*, *TGFBR1*, *TGFBR2* and *ENG* and major contribution comes from Pathways, Gene Ontology, PPI dataset. Additionally, the information from

phenotypical annotation: HuGe, GAD and Disease Ontology also adds to the overall ranking (since in the prioritization phenotypic information from test genes were included)



**Figure 3:** Heatmap plot for TGFBR3 gene explaining contribution of annotation sources and training genes.