Enhanced Human NGS Variant and Gene Regulation Analysis

BIOBASE is the leading provider of expert-curated biological databases, software and services for the life sciences. Our products and services identify relations critical to drug and biomarker discovery as well as improve biomedical research by transforming data into scientific concepts.

In the first session (A), you will learn more about the TRANSFAC® database of eukaryotic transcription factors and in the second session (B) more about the NGS analysis tool Genome Trax™.

(A) Applying TRANSFAC ® for Transcription Factor Binding Site Prediction

TRANSFAC® is a knowledgebase containing information on eukaryotic transcription factors and miRNAs, their experimentally-proven binding sites and regulated genes. Based on TRANSFAC®'s extensive compilation of binding sites, positional weight matrices are derived which can be used with the included Match™ tool to predict potential transcription factor binding sites in DNA sequences.

You will learn how to

- search for individual transcription factors and miRNAs, their experimentally-characterized binding sites and regulated genes, and ChIP experiments
- predict transcription factor binding sites and composite models within a promoter or DNA sequence

(B) Applying Genome Trax™ for Human NGS Variant Analysis in Personalized Medicine

Genome Trax™ allows you to quickly and confidently identify pathogenic variants in human whole genome or exome sequences. The database includes the only comprehensive collection of disease causing mutations from **HGMD® Professional** and pharmacogenomics variants from **PGMD™**, as well as regulatory sites from **TRANSFAC®**, and disease genes, drug targets and pathways from **PROTEOME™**. It further integrates the best public data-sets on somatic mutations, allele frequencies and clinical variants, in their most up-to-date version, for a total of more than 180 million annotations.
Genome Trax is fully compatible with many software packages, such as ANNOVAR™, CLC Genomics Workbench, and Alamut®.

You will learn how to

- find relevant variants within minutes
- identify known pathogenic variants, remove harmless common variants, and obtain deleterious predictions for novel variants.
- conduct trio comparisons to identify variants that are de novo or compound heterozygous only in the offspring