



University
of Basel

Department
Biozentrum



Swiss Institute of
Bioinformatics

BIOZENTRUM

The Center for
Molecular Life Sciences

Basel Computational Biology Seminar

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“Functional annotation of GWAS regulatory variants”

The Ensembl annotations are widely used for the analysis and interpretation of genome data using tools such as the Ensembl Variant Effect Predictor (VEP), which can quickly annotate the known variants of an individual and report on the potential effects of each. However, the analysis of individual variants is limited when analyzing the results of genome wide association studies (GWAS). In a majority of cases, the associated variants are not likely causing coding variants, rather potential regulatory variants with weak phenotypic associations.

I will present how weak associations with causal genes can be detected through a genome-wide and multi-layered integrative analysis. Ensembl's Regulatory Build synthesizes epigenomic datasets produced by large-scale projects such as ENCODE, Roadmap Epigenomics or BLUEPRINT. To gain further insight, we are collecting cis-regulatory interaction data such as GTEx to attach them to their target genes. Having brought all this data together, it is possible to develop advanced functional analysis methods without being constrained by the scale of the data, as we demonstrate with our post-GWAS analysis platform. This algorithm compares human GWAS results, as stored in public archives or in a private individual study, to a collection of genomic annotations, many of which are stored in Ensembl, producing a list of putative causal genes along with linked evidence.

Date: **Monday, May 27th, 2019**
Time: **16:00 h**
Room: **Lounge (level 13), Klingelbergstrasse 61**
(vis-à-vis Pharmazentrum)
Contact: **Maria Katsantoni (maria.katsantoni@unibas.ch)**