Integrated genomics approaches for the identification of causative variants and genes in GWAS-identified risk loci: application to Inflammatory Bowel Disease

Summary
Hundreds of risk loci for tens of common complex diseases have been identified by Genome Wide Association Studies. This represents a tremendous progress and is already providing new leads for the pharmaceutical industry. Yet, for the vast majority of risk loci, causative variants and genes remain unknown. Overcoming this limitation is essentially to reap the full benefits of GWAS.

We have developed integrated genomic approaches to achieve these goals. The identification of causative variants is based on at the application of advanced statistical approaches combined with genome imputation from the 1000 Genomes project. Identifying causative genes exploits transcriptome and eQTL analyses in a panel of disease relevant cell types, followed by resequencing the identified candidates in large case-control cohorts. The latter has been accomplished in collaboration with Riken, Yokohama. Latest results will be presented.