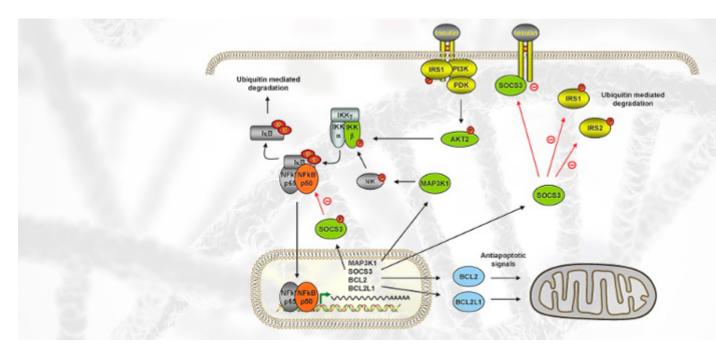


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## **Genetics of Complex Diseases**



We develop and apply computational tools to answer biomedical questions to better understand complex diseases by using all types of genetic and genomic data. We work on generating efficient computational strategies to better understand the association between human genetic variability and complex diseases.

## Summary

We have developed and applied several strategies and computational methods to enhance the analysis of genomic information for genome-wide association studies (GWAS) in many ways. Among others, we have developed filtering schemes that minimize false positive associations from genotype imputation results and have applied them successfully to different complex diseases, such as Asthma and Type 2 Diabetes. We are also developing alternative strategies to enhance the association signals of large GWAS studies using gene and pathway information.

A major current limitation of GWAS is the downstream interpretation of the functional role of the variants associated to the pathology. Our group, beyond identifying the genetic and molecular basis of disease, is also devoted to provide functionality to these variants, which are often located in apparently non-functional genomic regions. Using a combination of genotype imputation and Systems and Network biology approaches we aim to identify the potential causal variants, often not detected using a standard analytical approach, and to infer its potential functional impact. These approaches are already allowing us to identify candidate disease genes by using a combination of functional relationships together with gene-based tests.

## Objectives

• Developing and applying computational tools to answer biomedical questions to better understand complex diseases by using all types of genetic and genomic data.

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