

## Research Areas

The research focus of our clinically and genetically oriented laboratory is the molecular genetics of primary immunodeficiencies. Among these, we are mostly interested in studying adult patients with antibody deficiencies, who are clinically summarized under the diagnostic term "Common variable immunodeficiency – CVID". The methods applied to identify novel genetic defects are genetic linkage analysis and the investigation of selected candidate genes. Both approaches have been quite successful during the past years. Genetic linkage analysis revealed several chromosomal loci, which have been associated with familial cases of CVID (Chromosome 5p, 4q and 16q). The systematic functional and genetic screening of several candidate genes revealed five gene defects which present with CVID-like phenotypes: ICOS, TACI, CD19, BAFF-R and CD21. Currently we aim at understanding how these genetic defects cause the disease and influence its immunological and clinical presentation. In this respect, the further analysis of genetic defects in TACI and their functional evaluation are of great importance.

### Projects:

- [Taci deficiency](#)
- [New Antibody Defects">Characterization of novel genetic defects in patients with antibody deficiency](#)
- [Analysis of microRNAs in normal human peripheral B-cell development and patients with primary antibody deficiencies](#)