

Chair of Genetics Working Party – Job Description

The Genetics Working Party supports the exchange of genetic diagnostic tools to improve the diagnosis and the genetic counselling of patients with PIDs, which is important for patients' support and care. Moreover, in the context of the technological advances in genomics, the Genetics WP is an incubator aiming to develop these new applications for the diagnosis and research of new PIDs.

Main tasks:

- **Identify the different laboratories in Europe and abroad that perform genetic and prenatal genetic diagnostic assays for PIDs and provide this information on the ESID Website to ESID members**
- **Develop consensus statements for genetic diagnosis of PIDs**
- **Establish links from the ESID website to various immunodeficiency websites and genetic laboratories in regard to already identified mutations in PID genes.**
- **Collaborate with the ESID Registry WP**
- **Chair the "PID next-generation sequencing consortium" that has identified the following goals:**
 - **Efficiently implement and exchange experiences of genetic laboratories using next-generation sequencing (NGS)**
 - **Exchange mutation data, genotypes and phenotypes (for the interpretation of 'new' variants)**
 - **Develop Europe-wide consensus guidelines for PID NGS data handling and patient information/informed consent forms**
 - **Support research collaboration for study of genetic basis of PIDs**
- **Collaborate with the Molecular Testing Working Group of ERN RITA**
- **Attend the ESID Board meetings to update the Board**
- **Contribute to the ESID Newsletter**