



## **General task description of the ESID Working Party (WP) Chair**

- To act as an ESID Ambassador
- To oversee the WP Steering Group
- To design WP activities workplan, strategy, task development and execution and present these at board meetings and general assembly
- To work closely with the other ESID's WPs' Chairs and the President, Secretary and Treasurer
- To contribute to the mission of ESID through activities, ideas and innovation
- To attend the monthly Board meetings
- To contribute to ESID's quarterly newsletter content
- To liaise and collaborate with the ESID Executive Director
- To respond in a timely manner to Board/Society emails
- To ensure an efficient and complete transition for incoming WP Chairs

## **Job Description of the ESID Genetics Working Party (WP) Chair**

The ESID Genetics WP supports the exchange of genetic diagnostic tools to improve the diagnosis and the genetic counselling of patients with PIDs, 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 of and research into new PIDs.

### **Main tasks and commitments:**

- Identify the different laboratories in Europe and abroad that perform genetic and prenatal genetic diagnostic assays for PIDs and provide this information via the ESID Website to ESID members.
- Develop consensus statements for the 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 the study of the genetic basis of PIDs.
- Collaborate with the Molecular Testing Working Group of ERN RITA.