

ESID Junior Country Representative Elections Candidacy

Candidate: Laura Batlle Masó

Country: Spain

I'm a human biologist specializing in genetics, bioinformatics, and immunology. My major research interest is to provide molecular diagnoses for patients with inborn errors of immunity (IEI). For that, I use bioinformatic tools and cutting-edge sequencing technologies such as WES, WGS, optical gene mapping, long-read sequencing, deep amplicon sequencing, etc. Despite the increasing number of genetic defects associated with IEI over the past decades, many patients remain undiagnosed, leading to uncertain prognoses and treatment options. Over the past years, I have been working to enhance the understanding of IEI to better diagnose and treat these diseases. A deeper understanding of the genetic origins of IEI will lead to more accurate, rapid, and personalized diagnoses and genetic counseling for patients and their families.

I am a strong advocate for cooperation and collaboration. To this end, I have been actively involved in international collaborations, including research stays at Uppsala University and Radboud University, participation in international consortia (Solve-RD, ESID Registry, ClinGen/NIH Variant Curation Expert Panel), and involvement in international meetings and conferences (ESHG, ESID, ERN).