

## POSITION

### 1. Project Title/ Job Position title:

Towards Precision Medicine in Primary Immunodeficiencies: Unraveling new Genetic Etiologies by 'Omics' Approaches.

### 2. Area of Knowledge:

Life Sciences

### 3. Group of disciplines:

Human Biology, Human Immunology, Molecular Biology, Genetics, Cellular Biology, Genomics and Proteomics, Biochemistry

### 4. Research project/ Research Group description

Primary immunodeficiencies (PIDs) were initially defined as rare genetic disorders affecting the development or function of the immune system. However, it has become apparent in recent years that PIDs are much more common than initially thought. It is now even suspected that a high percentage of patients with severe infections since childhood actually have inborn errors of immunity. The **main challenge** in the study of PIDs is that each patient is unique and the causal gene must be explored individually, to determine the most appropriate treatment. Whole-exome sequencing (WES), the targeted sequencing of the protein-coding portion of the human genome, has been shown to be a powerful and efficient method for identifying the disease variants underlying genetic disorders. However, even in the best-case scenario, this process takes time, and time is at a premium as many patients die before their genetic defect is characterized. So, despite these improvements, there is an urgent need to develop ways of providing these patients with the necessary precision medicine treatments more rapidly.

### 5. Job position description

**Role:** The candidate will be in charge of performing the experimental activities of the project, in collaboration with other members of the group, in order to unravel new genetic etiologies in PIDs. He/she will be trained accordingly and mentored through the completion of his/her PhD thesis.

#### Responsibilities

Set up and perform experiments, maintain experimental resources (as cell lines, reagents, etc.) according to protocols, analyze & interpret results and contribute to the development of experimental strategies with accuracy and honesty.

Keep updated the laboratory notebook and properly store and manage the data produced during the project.

Collaborate with colleagues and participate in team activities (such as meetings, seminars, workshops, etc.) across the research group and wider scientific community while keeping up to date with current knowledge and recent advances.

Participate in knowledge exchange with both society and industry, to promote the value of research in public health and to contribute to the dissemination of his/her research results in the principles of EU's Open Science policy.

Undertake any other duties of equivalent standing as assigned to him/her.

### Skills

Degree in Life Sciences (Biology, Biochemistry, Biotechnology or similar).

Experience in cell culture is desirable.

Motivation, critical thinking and problem-solving oriented skills.

Good interpersonal skills, including team working.

Good communication skills, willingness to engage in public presentations and ability to transmit complex concepts in a clear way.

Good time and workload management skills, including both initiative and flexibility.

### **GROUP LEADER**

1. **Title:** Dr.
2. **Full name:** Rebeca Pérez de Diego
3. **Email:** [rebeca.perez@idipaz.es](mailto:rebeca.perez@idipaz.es)
4. **Research project/Research group website:** [www.lighd.org](http://www.lighd.org)