Annachiara Pastore

Identification of single-cell RNA sequencing in a mouse model of Fanconi Bickel Syndrome

Duration	Six months
Short Bio	I'm a PhD student at University of Campania "Luigi Vanvitelli (Naples) where I am working on Fanconi Bickel syndrome rare kidney condition to find out how we can improve diagnosis and theraphy by looking at new incentives. After one initial period in experimental in vitro design, I moved in the fascinating word of NGS. Thanks to the ERKnet grant, I joined one of state-of-the-art laboratory devoted to multi-omic single-cell sequencing in Germany, where I had the possibility to acquire single-cell sequencing and spatial transcriptomic expertise performing my own experiments. Last, but not least, I am learning to analyze single-cell expression data using bionformatic programming language.
Home Institution	University of Campania "Luigi Vanvitelli"; Department of Cardiothoracic and Respiratory Sciences.
Host Institution	Internal Medicine and Nephrology (Division of Nephrology and Clinical Immunology) University Hospital RWTH Aachen.
Project Description	Fanconi Bickel Syndrome (FBS), is a rare autosomal recessive disorder that affects the kidney's proximal renal tubule. The pathology of FBS is due to mutation in the SLC2A2 gene encoding the GLUT2 transporter. Currently, there is no treatment available apart from dietary electrolyte replacement. Single cell sequencing conbined with spatial transcriptomic is constructive in understanding the mechanism behind the disease, identifying the novel diagnostic and promoting target discovery.
Personal Statement	Thanks to EJPRD fellowship I had the possibility to work in a laboratory on the front line in NGS, working with a brilliant team that supported me in each step of single cell analysis. This experience gave me also the possibility to grow personally since it was my first period abroad and gave me the chance to develop a

	network of collaboration that would be useful for my present and future as researcher. This project fullfill the main goals of ERKNet due to its high impact in development of new diagnosis guideline and future advances for development of new treatment that will help Fanconi Bickel Syndrome's patient life.