Sante Princiero Berlingerio

Treatment of cystinosis podocytes with non-cystine lowering drugs

Short Bio	I am PhD student in the laboratory of Pediatric Nephrology in KU/University Hospitals Leuven in Belgium, with a great interest in a rare metabolic disease called cystinosis. During my academic education, I have always been intrigued by renal pathophysiology and my PhD allowed me to further deepen my knowledge of the molecular mechanisms necessary to maintain a functional glomerular filtration barrier. These insights may be applied not only to cystinosis but also to other chronic kidney diseases.
Home Institution	University of Leuven, Leuven, Belgium
Host Institution	University of Torino, Torino, Italy
Project description	My project is focused on cystinosis, a rare and incurable disease characterized by cystine accumulation in all tissues due to mutations in the CTNS gene, where kidneys are the first organ affected. In parallel with renal Fanconi syndrome, patients also display glomerular dysfunction leading to progressive deterioration of their kidney function. The current treatment with cysteamine depletes intracellular cystine but only postpones the development of end-stage kidney disease. Our group has studied podocytes isolated from urine of cystinotic patients, and has shown that they present pathologic features such as altered cytoskeleton, impaired cell adhesion and increased motility, which cannot be restored by cysteamine (Ivanova et al. KI 2016). Therefore, other molecular mechanisms need to be tackled in order to restore glomerular function in cystinosis. In my PhD, I found that cystinosis podocytes show altered metabolic profile, known to affect cell adhesion and motility. With this exchange, I aim at restoring metabolic dysfunction in cystinosis podocytes for the long-term preservation of glomerular function.
Personal statement	This exchange will benefit the knowledge in the rare disease domain since cystinosis is an incurable disease and its study has been hampered by the lack of functional model systems that properly mimic the disease state, especially in the glomerulus. Moreover, the outcome of the research will be useful to various renal diseases that present dysfunctional glomerular filtration barrier, such as diabetic nephropathy.