Translational Approaches for Cystinosis, New Targets, Same Hope

Sergio D. Catz, PhD
Scripps Research

In the last decade we performed research leading to the discoveries that two fundamental biological processes are affected in cystinosis: white blood cell-mediated inflammation and autophagy, a cellular process important to preserve resources and cell function. Dysregulation of either of these processes leads to cell malfunction including proximal tubule cell dysfunction. Our research has two fundamental objectives. First, it is directed at discovering new mechanisms that are affected in cystinosis and are not corrected by current therapies. Second, once those mechanisms are identified, we design strategies to discover new translational approaches, i.e. drugs to improve defective mechanisms and pathways to improve cell function in cystinosis. In my presentation, I will discuss the discovery of new strategies to reduce inflammation and improve autophagy in cystinosis and will present new data supporting the potential effectiveness of these approaches.