Patient journey in cystinosis: focus in non-adherence and disease management

Lay summary

Cystinosis is a rare hereditary disease that can be fatal if not treated. Its clinical manifestations are due to the accumulation of cystine, a component of proteins, within cells of virtually all organs of human body. It initiates in the kidneys and can eventually lead to renal failure. Furthermore, it progressively affects other organs including eyes, thyroid, pancreas, reproductive organs, muscles and central nervous system. It requires lifelong treatment.

A patient journey mapping (PJM) is a way to know the trajectory and experience with the disease. Cystinosis PJM was developed from interviews and workshops with parents/caregivers and patients. An important result was that patients were not fully aware of risks of not following schedules and dosages as prescribed by their doctors. Impaired sleep, chronic fatigue and gastrointestinal symptoms were the main reasons for not taking medication correctly. Furthermore, these factors had a negative impact on patients' quality of life. Finally, a series of lines of action were proposed and suggestions were made to improve PJM.