Introduction to "Extra-Renal Complications of Cystinosis"

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Cystinosis is one of the earliest described inborn errors of metabolism, first documented in 1903.[1] It is inherited as an autosomal recessive condition and presents as a broader phenotype than merely the initial renal Fanconi syndrome, one that affects different organ systems as the patient ages. Understanding of this pan-tissue and pan-organ disease has evolved over the past century, and is the subject of several recent reviews.[2,3] A pediatric nephrologist or biochemical geneticist usually makes the diagnosis after referral from a primary care physician who has noted polyuria, glucosuria, and often, failure to thrive. In children diagnosed later in childhood, either photophobia because of corneal cystine accumulation or rickets associated with hypophosphatemia from the renal proximal tubulopathy may be the presenting chief complaint. Whereas the complex pathophysiology is still being elucidated, the single hallmark that characterizes the disease is lysosomal cystine storage because of failure of the lysosomal cystine transporter, cystinosin. The diagnosis is established primarily by determination of leucocyte cystine, and now that molecular testing is available, determination of the genotype as well.[1]

This supplement is the result of a colloquium held in Miami in January 2016, attended by a group of 6 cystinosis experts with the intention of summarizing the current knowledge on many extra-renal complications of the nephropathic form of cystinosis to assist in early recognition, treatment, and prevention of such complications. ■

Author Disclosures

The author declares no conflicts of interest, real or perceived.

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From the University of Michigan School of Medicine, Ann Arbor, MI
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0022-3476/$ - see front matter.
http://dx.doi.org10.1016/j.jpeds.2016.12.051