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Treating the cystine stone former presents a singular clinical challenge

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Cystinuria represents a relatively rare hereditary condition leading to impairment of the renal proximal tubule's dibasic amino acid transporter. The only manifestation of this disease is calculi formation. Unlike other nephrolithiasis disease processes, cystine stone formation is due directly to supersaturation of an insoluble solute precipitating out of the urinary milieu. The genes responsible for this are SLC3A1 and SLC7A9 which encode for the protein heterodimer that is responsible for reabsorption of filtered dibasic amino acids. While the International Cystinuria Consortium recently revised its categorization of cystine patients into type A and type B where type A patients have mutations in both alleles of SLC3A1 and type B in SLC7A9, from a clinical perspective, genotyping offers little clinical benefit and does not alter patient treatment or give insight to disease penetrance. Although both of these genes and their protein products have been characterized, the stone forming tendency of cystinurics is not fully understood and further genetic mutations may exist as well. Cystinuria is most easily diagnosed with a stone analysis revealing the classic hexagonal-shaped cystine crystal, but a positive family history of cystinuria, a urine sample with elevated cystine (often >400 mg/day), or a positive sodium nitroprusside test can also make the diagnoses (1).

Treatment of these patients typically includes urinary

alkalization, typically with potassium citrate, increased hydration, and decreased sodium and animal protein intake. If these fail, then cystine binding thiol medications are initiated. Patients' tolerance of these medications is debatable and their availability can be problematic as well. Anyone that has cared for these patients can attest to the frustration that both the patient and clinician experience in trying to prevent future calculi formation. While these patients represent a relatively small proportion of stone formers, the impact of their morbidity is significant. Fortunately, new drug development continues, utilizing new techniques with computer modeling as well as atomic force microscopy. These and other novel techniques will hopefully lead to better characterization of the disease penetrance and its medical treatment.

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