P056 The Importance of evaluating for potential underlying causes of kidney stones: A survey of physician experiences in diagnosing Primary Hyperoxaluria type 1 (PH1)

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Introduction & Objectives: PH1 is a rare genetic disorder characterized by persistent hepatic overproduction of oxalate. Oxalate crystalizes with calcium and leads to recurrent kidney stones, nephrocalcinosis, progressive renal failure, and multiorgan damage from systemic oxalosis. Among patients with PH1, clinical phenotype is heterogeneous with a variable age of presentation and an unpredictable progression rate, leading to delayed diagnosis and increased disease burden. A series of case-based physician interviews was conducted to identify triggers that raised suspicion of PH1 and factors that contributed to diagnostic delays throughout the patient journey.

Materials & Methods: Select participation criteria included: physicians in practice 2+ years; active role in diagnosing, treating, or managing 1+ PH1 patient(s) within last 5 years; spend ≥50% of time in direct patient care; able to review PH1 patient records. Patient history served as basis for further probing in 60-minute interviews involving open-ended questions from a semi-structured interview guide.

Results: 37 Physicians participated and reported on 54 PH1 patients. Diagnosis age ranged from 1 month – 48 years (median 7.5 years). At diagnosis 54% of patients had progressed to advanced kidney disease. Diagnosing Specialties were nephrologists (83%), geneticists (13%), and urologists (4%). 63% of patients had initially seen a urologist and remained under urology care for treating stones for several years with no metabolic evaluation; these patients spent an average of 5 years in this stone treatment cycle before being referred and diagnosed. The following were identified as triggers raising initial suspicion leading to diagnosis: recurrent kidney stone events (39%); nephrocalcinosis (17%, all in pediatric cases); progressive kidney disease (13%); acute renal failure (13%); single kidney stone event (11%, all in pediatric cases). Patients were primarily diagnosed via genetic testing (76%). Mean time to diagnosis was 1.8 months in infants, 1.3 years in pediatrics and 9.2 years in adults. When asked what, in retrospect, should have triggered suspicion in these cases, kidney stone(s), a single event in pediatrics and recurrent events in adults were most commonly identified.

Conclusions: Although all patients with kidney stones should be evaluated according to existing stone management guidelines, many patients in this study remained in the stone treatment cycle without evaluation for extended periods of time. A single stone event in children and recurrent events in adults should immediately trigger suspicion and evaluation for an underlying genetic cause, such as PH1, to provide an opportunity to modify the disease course.