

Taroni F.¹, Gnech M.², Capone V.¹, Berrettini A.², Pavesi M.³, Manzoni G.², Montini G.¹

¹Fondazione IRCCS Ca' Granda - Ospedale Maggiore Policlinico, Dept. of Pediatric Nephrology, Dialysis and Transplant Unit, Milan, Italy,

²Fondazione IRCCS Ca' Granda - Ospedale Maggiore Policlinico, Dept. of Pediatric Urology, Milan, Italy, ³Fondazione IRCCS Ca' Granda - Ospedale Maggiore Policlinico, Dept. of Pediatric Radiology, Milan, Italy

Introduction & Objectives: Urolithiasis is a rare disease in children. For many years children with kidney stones have been managed like "small adults", but there are significant differences between the pediatric and the adult age in clinical presentation, etiology and treatment. Management of this condition in children has some peculiarities, as it is often the sign of an underlying metabolic abnormality (1). Some of these metabolic alterations can lead to serious consequences, such as chronic renal failure, if not adequately diagnosed and treated. Moreover, stones in children with a metabolic abnormality can recur throughout their life, with the need for repeated surgical procedures over the years. So a systematic work up is useful to diagnose metabolic defects and establish a personalized therapy (2). The aim of the study was to identify the etiology of urolithiasis and the risk factors related to a metabolic rare disease. In pediatric patients diagnosed at the Pediatric Nephrology Department of the IRCCS Policlinico, Milan, Italy, from 2014 to 2019.

Materials & Methods: We reviewed the medical records of all children diagnosed at the Pediatric Nephrology Department of the IRCCS Policlinico, Milan, Italy, from 2014 to 2019. Inclusion criteria were: Age < 18 years, absence of comorbidities. Data were collected about age, sex, family history, clinical features, surgical management and metabolic abnormalities. All children had undergone a complete metabolic work up on blood and urine.

Results: 136 patients were enrolled in the study. A metabolic rare disease was diagnosed in the 25% of children (70% cystinuria, 13% defect in acid uric metabolism, 4% hyperoxaluria type 1, 10% hypersensitivity, Vitamin D, 3% Dent Disease). Metabolic defects without a rare metabolic disease was identified in the 75% of patients (28% hypercalciuria, 25% hypercalciuria and hypocitraturia, 8% hypocitraturia, 6% hypomagnesiuria, 4% secondary hyperoxaluria, 2% hypercuria, idiopathic 27%). Age at diagnosis, US characteristic and indication to surgery were significantly different patients with a rare metabolic disease and not having a rare metabolic disease ($p < 0,0001$).

Conclusions: 25% of pediatric patients affected by urolithiasis is at risk of having a rare metabolic disease. Patients at higher risk are children with early onset (< 5 years), multiple stones at US and indication to surgery at diagnosis. A metabolic work up is useful in every pediatric patients with urolithiasis in order to identify patients affected by a rare metabolic disease.