diagnosed with HUS between 2000 and 2016, in France. All cases were registered by the Institut National de Veille Sanitaire. Neurological, renal and digestive outcomes were evaluated 1 month and 1 year after the diagnosis.

Results: 40 patients were included. The median age was 3 months (IQR, 1.25–5 months). Diarrhea preceding HUS was reported in 80% of patients, which was bloody in 62.5% of patients. The source of infection was identified in 47.5% of cases and was most commonly inter-individual (30.0%). During the acute phase, renal impairment was mild. Only 25.0% of patients required dialysis. 35.0% of patients presented with digestive complications, 27.5% with neurological symptoms and 10.0% with cardiac abnormalities. More than 60% of patients had renal consequences at 1 month and 1 year after follow-up (64.8% and 63.5%, respectively) which mainly manifested as proteinuria. 12.1% of patients presented neurological symptoms at 1 year follow-up. Two patients had unexplained biliary cirrhosis during the follow-up and needed hepatic transplantation.

Conclusions: The acute clinical presentation of typical HUS in children of this age group (0-6 months) has not been studied in isolation. This work highlights the unique implications for this age group, in particular patients displayed poor renal and neurological outcomes. Long term follow-up is necessary with research of proteinuria.

O-26 LONG-TERM OUTCOME AND TREATMENT PRACTICES IN DISTAL RENAL TUBULAR ACIDOSIS


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Introduction: Inherited distal renal tubular acidosis (dRTA) is a rare disorder and consequently little data about treatment and long-term outcome.

Material and methods: We contacted paediatric and adult nephrologists through European professional organizations. Responding clinicians entered demographic, biochemical, genetic and clinical data in an online form.

Results: 333 responses with complete data were received (29 countries, 69% male, 31% female, median age 11 (range 0-70 years). Mutation testing had been performed in 206 (62%), with pathogenic mutations identified in 168 (82%), divided into: ATP6V1B1 37%, ATP6V0A4 34% and SLC4A1 29%. The median (range) age at presentation was 0.5 years (0-54) and at last follow up 11.0 years (0-70.0).

The median (interquartile range) prescribed dose of alkali treatment in mg/eq/kg/day was 1.9 (1.1-3.1), normal plasma bicarbonate levels were achieved in the majority of patients (78.4%). Adult height was only slightly below average with a mean height SDS (standard deviation score) of -0.51 (±1.04) at a median (range) age of 28 (18-70) (N=68). Estimated glomerular filtration rate (eGFR, by Cockcroft-Gault) at last follow up in adults established the presence of chronic kidney disease stages 2-4 in 52%. Nephrocalcinosis was common (87.1%). A history of nephrolithiasis was more common with SLC4A1 mutations (45.8%) compared to the autosomal recessive forms (19.1%), p=0.002. Hearing loss was reported (35.4%) with the highest prevalence in patients with ATP6V1B1 mutation (83.1%).

Conclusions: Long term follow-up from this large cohort of patients with dRTA shows an overall favourable outcome with regards to growth but half of adult patients have decreased eGFR. Treatment adequacy as judged by plasma bicarbonate and normocalciuria is achieved in most patients.


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