

Treatment and long-term outcome in primary nephrogenic diabetes insipidus

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Abstract

Background: Primary nephrogenic diabetes insipidus (NDI) is a rare disorder and little is known about treatment practices and long-term outcome.

Methods: Paediatric and adult nephrologists contacted through European professional organizations entered data in an online form.

Results: Data were collected on 315 patients (22 countries, male 84%, adults 35%). Mutation testing had been performed in 270 (86%); pathogenic variants were identified in 258 (96%). The median (range) age at diagnosis was 0.6 (0.0–60) years and at last follow-up 14.0 (0.1–70) years. In adults, height was normal with a mean (standard deviation) score of -0.39 (± 1.0), yet there was increased prevalence of obesity (body mass index >30 kg/m²; 41% versus 16% European average; $P < 0.001$). There was also increased prevalence of chronic kidney disease (CKD) Stage ≥ 2 in children (32%) and adults (48%). Evidence of flow uropathy was present in 38%. A higher proportion of children than adults (85% versus 54%; $P < 0.001$) received medications to reduce urine output. Patients ≥ 25 years were less likely to have a university degree than the European average (21% versus 35%; $P = 0.003$) but full-time employment was similar. Mental health problems, predominantly attention-deficit hyperactivity disorder (16%), were reported in 36% of patients.

Conclusion: This large NDI cohort shows an overall favourable outcome with normal adult height and only mild to moderate CKD in most. Yet, while full-time employment was similar to the European average, educational achievement was lower, and more than half had urological and/or mental health problems.

Keywords: AQP2; AVPR2; chronic kidney disease; flow uropathy; nephrogenic diabetes insipidus.