

Abstract Submission No.: A-0706**Long-term clinical outcomes in pediatric patients with primary nephrogenic diabetes insipidus**

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Objectives : Nephrogenic diabetes insipidus (NDI) is a rare inherited disorder, and there is a paucity of studies assessing long-term clinical outcomes in affected individuals. This study aims to evaluate the long-term clinical outcomes in pediatric patients with NDI.

Methods : This multicenter retrospective study included forty-nine children diagnosed with NDI. The medical records of these patients were retrospectively reviewed, and a linear mixed model was employed to analyze variations in growth and kidney function with increasing age.

Results : Mutations in the AVPR2 and AQP2 genes were identified in 40 (81.6%) and 4 (8.2%) patients, retrospectively. The median ages at diagnosis and the last follow-up were 0.41 years and 9.97 years, respectively. Thiazide treatment was administered to all patients, with 43 (87.8%) and 14 (28.6%) patients also receiving amiloride and indomethacin, respectively. The estimated marginal means (EMM) for height Z score decreased from -0.114 at diagnosis to -0.899 at 19 years of age, significantly lower than at diagnosis ($P = 0.038$). The EMM for weight Z score increased from -1.580 at one year of age to 0.760 at 19 years of age, exhibiting a steady increase ($P < 0.001$). The EMM for BMI Z score demonstrated a consistent increase after age of 2 years ($P < 0.001$), which were -0.326 at 2 years of age and 1.210 at 19 years of age. The proportion of obesity increased from 3.2% at 3 years of age to 33.3% at 19 years of age. The EMM for eGFR consistently increased from 82.8 mL/min/m² to 100.8 mL/min/m². Although eGFR less than 60 mL/min/m² is not present, the eGFR less than 90 mL/min/m² is 47.5% at last follow up.

Conclusions : To improve long-term outcomes in patients with NDI, clinicians need to monitor nutritional support and monitor kidney function.