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Analysis of a Novel *AVPR2* Mutation in a Turkish Family with Nephrogenic Diabetes Insipidus

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ABSTRACT Congenital nephrogenic diabetes insipidus (NDI) is a rare X-linked recessive disorder associated with germline mutations of the arginine vasopressin (AVP) receptor type 2 (*AVPR2*) gene. The researchers describe a novel mutation in the *AVPR2* gene in a three-generational Turkish family with NDI. In the present report, a 22-year-old man is reported with polyuria and bilateral non-obstructive hydronephrosis. He was diagnosed with partial NDI based on the clinical phenotype, the water deprivation test and the inadequate response to 1-desamino-8-Darginine vasopressin (DDAVP) administration. All family members who were suspected to have diabetes insipidus and/or related symptoms were studied. Sequencing analysis of the AVPR2 gene revealed the novel missense mutation c.392 T>C; p. Leu 131 Pro:L131P (*AVPR2* gene (coding seq # NM_000054.4;prot seq # NP_000045.1). In conclusion, the proband carries a novel *AVPR2* missense mutation inherited from his carrier mother.