18) A novel mutation in the AVPR2 gene in a Palestinian family with nephrogenic diabetes insipidus

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Abstract

Background: Nephrogenic diabetes insipidus (NDI) is a clinical disorder characterized by a urinary concentrating defect resulting from resistance of the collecting duct to the antidiuretic action of vasopressin (AVP).

NDI is classified into hereditary and acquired causes.

X-linked recessive NDI is caused by mutations in the gene encoding the V2 vasopressin receptor (V2R) and is the most frequent genetic cause of the inherited NDI.

Here we describe a novel mutation in the AVPR2 gene in a Palestinian family with NDI.

Clinical Data: A male infant, born to a non consanguineous Palestinian family, presented in the neonatal period with failure to thrive, vomiting, irritability & fever. Blood sodium was high up to 170mmol/L, blood osmolality raised over 330mOsm/kg while urine osmolality remained low between 45-135mOsm/kg, urine output was 7cc/kg/hr & positive family history of a brother diagnosed previously to have NDI suggesting X-linked inheritance of the disease.

Molecular Data: Sequencing the AVPR2 gene revealed a novel mutation (C82Y) in affected patients in exon 2 of the gene, predicting Cysteine to Tyrosine substitution at the 82 amino acid residue of the AVPR2 gene, while the mother being carrier for the mutation and healthy brother and father does not have the mutation.

Conclusion: We describe a novel mutation in the AVPR2 gene in a Palestinian family with NDI, allowing early diagnosis to prevent severe dehydration and complications in addition to genetic counseling.

19) Molecular diagnoses of Tyrosinemia Type II following Identification and Characterization of Tyrosine Aminotransferase (TAT) Gene Mutations Among Suspected Patients

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Tyrosinemia Type 11, also called Richener- Hanhart syndrome (RHS) is a rare autosomal recessive disorder. It is caused by deficiency in tyrosine aminotransferase (TAT), resulting in elevated tyrosine levels in plasma and urine, and leading to painful palmoplantar hyperkeratosis, pseudodendritic keratitis and variable mental retardation.

Sizteen different mutations have been reported in the TAT gene worldwide. Although the clinical complications of tyrosinemia type 11 were described in patients from the Middle East, the molecular basis has been identified only in a limited number of Tunisian and Palestinian patients. Molecular genetic analysis represents the most reliable and accurate approach to identity heterozygote (carrier) and homozygous (patients) genotypes among suspected individuals since TAT is not expressed in chorionic villi or animosities. This investigation involves the molecular characterization of TAT gene mutations in