X-linked Nephrogenic Diabetes Insipidus (NDI1) – OMIM 304800; Nephrogenic Diabetes Insipidus (NDI2) – OMIM 125800

Background

Nephrogenic diabetes insipidus (NDI) is characterised by excessive thirst and excretion of large amounts of extremely dilute urine and is caused by a decrease in the ability of the kidney to concentrate the urine. This condition can be acquired as a result of a kidney condition or defect, or as a side effect to some medications; it can also be inherited. NDI type 1 is due to a mutation in the X-linked AVPR2 gene (OMIM 300538) which results in kidney being unable to reabsorb water. In more rare cases, a mutation in the *AQP2* gene (OMIM 107777) causes NDI type 2. This mutation is often inherited in an autosomal recessive manner although dominant mutations are reported from time to time.

Recommended Clinical Referral Criteria

- Patient meets the clinical criteria
- Family history

Molecular Analysis

Mutation screen: Bi-directional sequence analysis of the entire coding region of *AVPR2* and *AQP2* genes
Family follow-up: Testing for known familial mutations in *AVPR2* and *AQP2* genes

Test (Price available on request)	TAT
Testing for known familial mutations (any of the genes listed above)	42cd
Mutation screening (*AVPR2*)	42cd
Mutation screening (*AQP2*)	42cd

Contact Details
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Accredited to ISO 15189:2012
(8988)

Sample Requirements
Blood – 5ml in EDTA (1ml neonates/infants);
please contact lab prior to sending a
prenatal sample.
Please label samples with three identifiers and date of collection

All samples must be accompanied by request form

Links

Orphanet
http://www.orpha.net/
EDDNAL
http://www.eddnal.com/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/