

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** AVPR2 Sequencing
- **Test Code:** 4016

Related Diseases & Genes:

- “Diabetes insipidus, nephrogenic” (OMIM #: 34800) || Gene: AVPR2 (OMIM #: 300538)
- Nephrogenic syndrome of inappropriate antidiuresis (OMIM #: 300539)

Synonyms:

- ARGININE VASOPRESSIN RECEPTOR 2
- ANTIDIURETIC HORMONE RECEPTOR; ADHR
- VASOPRESSIN V2 RECEPTOR; V2R
- “DIABETES INSIPIDUS, NEPHROGENIC, TYPE I”
- NDI
- “DIABETES INSIPIDUS, NEPHROGENIC, X-LINKED”

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dysmorphology
- Neurogenetics
- Inborn Errors of Metabolism

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