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**Diabetes insipidus
X-linked recessive**

Molecular Genetics Service Profile V2R

Introduction

- ✓ The V2R gene has been mapped to chromosome Xq28, spans 4.63 kb, and consists of 4 exons, which encode a predicted protein of 371 amino acids. This protein belongs to the family of G-protein coupled receptors with seven transmembrane domains - the V2 receptor.
- ✓ In the kidney, water reabsorption is mainly regulated by the binding of arginine vasopressin to the vasopressin type 2 receptor (V2R). This receptor is expressed selectively in principal cells of the collecting ducts.
- ✓ Mutations within the V2R gene lead to X-linked recessive inherited **Nephrogenic Diabetes Insipidus (NDI)**. While only males are symptomatic among patients with X-linked inheritance, females with heterozygous mutations (carriers) sometimes present with mild NDI symptoms due to variable inactivation of the mutated gene (Lyon mechanism).

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected Nephrogenic Diabetes Insipidus.

Samples

- ✓ Minimum of 2 ml blood sample in EDTA (or minimum of 50 µg DNA from peripheral lymphocytes) can be sent to our laboratory by express mail. In special cases a investigation of DNA from prenatal samples can be made, however you should contact our laboratory for further details.

Technical

- ✓ Mutation scanning of exons 1-4 of V2R by direct analysis with Dideoxy sequencing (ABI 310).

Target turn-round time

- ✓ 3-4 weeks from the receipt of all required samples and clinical information.

Cost

- ✓ NDI Full mutation screen (V2R and AQP2) - € 700. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- ✓ Birnbaumer M "Vasopressin receptor mutations and nephrogenic diabetes insipidus. Review." Arch Med Res. 1999 Nov-Dec;30(6):465-74.
- ✓ Knoers NV, Deen PM "Molecular and cellular defects in nephrogenic diabetes insipidus. Review." Pediatr Nephrol. 2001 Dec;16(12):1146-52.

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