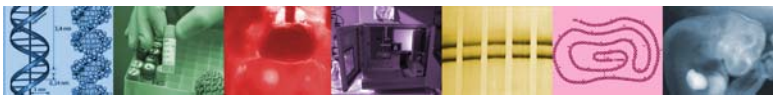

www.endogenet.org

Diabetes insipidus
Autosomal dominant

Molecular Genetics Service Profile
AVP (ARVP, ADH)

Introduction

- ✓ The term AVP is an abbreviation of Arginine Vasopressin, a hypothalamic hormone which is identical to the Antidiuretic Hormone (ADH). The AVP gene has been mapped to chromosome 20p13, spans 2.86 kb, and consists of 3 exons, which encode the preprohormone of 164 amino acids.
- ✓ Mutations within the AVP gene lead to autosomal dominantly inherited **Neurohypophyseal Diabetes Insipidus**.
- ✓ The mutant vasopressin prohormones may accumulate in the endoplasmatic reticulum with consecutive degeneration of the affected hypothalamic neurons causing the autosomal dominant nature of the observed mutations.
- ✓ MRI may show absence or diminution of the hyperintense signal normally emitted by the posterior pituitary.

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected Central 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 1-3 of AVP 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

- ✓ Mutation screen (AVP) - € 350. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- ✓ Christensen JH et al. “Six novel mutations in the arginine vasopressin gene in 15 kindreds with autosomal dominant familial neurohypophyseal diabetes insipidus give further insight into the pathogenesis.” Eur J Hum Genet. 2004 Jan;12(1):44-51.

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