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APS, Type I (APECED)
Autosomal recessive
Autosomal dominant

Molecular Genetics Service Profile
AIRE

Introduction

- ✓ The term AIRE derives from an acronym of Autoimmune Regulator. The AIRE gene has been mapped to chromosome 21q22.3, spans approximately 13 kb, and consists of 14 exons, which encode a predicted protein of 545 amino acids.
- ✓ AIRE is expressed in medullary epithelial cells, monocyte-derived cells of the thymus, cells of the lymphnodes, spleen, and in the fetal liver and therefore may play a role in the induction and maintenance of immune tolerance.
- ✓ Mutations within the AIRE gene lead to autosomal recessive or dominant Autoimmune Polyglandular Syndrome (APS), Type I with an immune-mediated destruction of endocrine tissues, chronic candidiasis, and additional ectodermal disorders.
- ✓ Clinical features: **Addison disease, hypoparathyroidism, chronic mucocutaneous candidiasis, diarrhea, malabsorption, autoimmune hepatitis** and others (see OMIM #240300).

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected Autoimmune Polyendocrinopathy, Type I.

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 1-14 of AIRE by dHPLC (WAVE), denaturing high pressure liquid chromatography. Fragments with abnormal elution patterns are directly analyzed with Dideoxy sequencing (ABI 310).

Target turn-round time

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

Cost

- ✓ APECED mutation screen (AIRE) - € 960. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- ✓ Vogel A, Strassburg CP, Obermayer-Straub P, Brabant G, Manns MP. “The genetic background of autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy and its autoimmune disease components.” J Mol Med. 2002 Apr;80(4):201-11.

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