Molecular Genetics Service Profile

GHRHR (Growth Hormone Releasing Hormone Receptor Gene)

Introduction

- The GHRHR gene is mapped on chromosome 7p15 and consists of thirteen exons of about 15 kb in total, encoding a protein of 423 amino acids. This protein belongs to the family of G-protein coupled receptors with seven transmembrane domains - the GHRH receptor.
- Expression of GHRHR is localised to the pituitary gland, hypothalamus, renal medulla, placenta and other tissues and is up-regulated by the PIT1 protein. GHRHR is involved in anterior pituitary cell development and differentiation, and may play a role in proliferation of the somatotroph cell lineage.
- Mutation at the GHRHR locus leads to autosomal recessive IGHD (Isolated Growth Hormone Deficiency). Affected heterozygote individuals have not been observed.
- MRI may show a hypoplastic anterior pituitary.

Reasons for referral

- Mutation screening in patients with clinically confirmed or suspected IGHD.

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 1-13 of GH1 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

- IGHD Full mutation screen (GH1 and GHRHR) - € 1080. Screening the GHRHR gene is part of the GeNeSIS study sponsored by Eli Lilly and Company. Please contact your local representative. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- Salvatori, Roberto et al. "Three New Mutations in the Gene for the Growth Hormone (GH)-Releasing Hormone Receptor in Familial Isolated GH Deficiency Type IB." J Clin Endocrinol Metab. 86.1 (2001): 273-79.

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