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MPHD / IGHD
Autosomal recessive
Autosomal dominant

Molecular Genetics Service Profile **POU1F1 (PIT1; GHF-1)**

Introduction

- ✓ The name POU1F1 derives from POU domain, class 1, transcription factor 1. The term POU is an acronym of PIT1, OCT1 and UNC96, which are all related transcription factors from different organisms, with similar DNA binding domains. It is a homeobox gene located on chromosome 3p11 and consists of six exons covering 17 kb in total. It encodes a protein of 290 amino acids which is called PIT1.
- ✓ The PIT1 protein is first expressed during the early embryonic development. Its expression persists throughout life. PIT1 activates expression from the GH, TSH and PRL promoters and is also essential for the embryological development of the anterior pituitary.
- ✓ Mutation at the POU1F1 locus leads to autosomal recessive or dominant MPHD (Multiple Pituitary Hormonal Deficiency) with a consecutive loss of **GH, TSH, PRL**.
- ✓ MRI may show a hypoplastic pituitary gland.

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected MPHD.

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 1-6 of POU1F1 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

- ✓ MPHD Full mutation screen (PROP1, POU1F1, HESX1, LHX3, LHX4) - € 1560. Screening the POU1F1 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

- ✓ Understanding the genetics of growth hormone deficiency. R.Pfäffle and W.F.Blum. (2000). TMG Healthcare Communications Ltd: 49-53.
- ✓ Dattani, Mt and Ic Robinson. "The molecular basis for developmental disorders of the pituitary gland in man." Clinical Genetics 57.5 (2000): 337-46.

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