



**MPHD**  
**Autosomal recessive**

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## Molecular Genetics Service Profile **PROP1**

### Introduction

- ✓ The term PROP1 derives from an acronym of Prophet of PIT1. It is a "paired"-like homeobox transcription factor on chromosome 5q and consists of three exons spread over 3 kb in total, encoding a protein of 226 amino acids.
- ✓ During the embryonic development PROP1 is necessary for the proper expression of PIT1 from the POU1F1 gene.
- ✓ Mutations within the PROP1 gene lead to autosomal recessive MPHD (Multiple Pituitary Hormonal Deficiency) with a consecutive loss of **GH, TSH, PRL, LH, FSH and often ACTH**. Affected heterozygote individuals have not been observed.
- ✓ MRI may show either hypocellular pituitary or intra- and extrasellar cell masses that must not be confused with pituitary adenomas.

**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 1-3 of PROP1 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 PROP1 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.
- ✓ Bottner A et al. "PROP1 Mutations Cause Progressive Deterioration of Anterior Pituitary Function including Adrenal Insufficiency: A Longitudinal Analysis." J Clin Endocrinol Metab. 2004 Oct;89(10):5256-65

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