



MPHD / IGHD
Autosomal recessive
Autosomal dominant

www.endogenet.org

Molecular Genetics Service Profile **HESX1 (Rpx)**

Introduction

- ✓ The term HESX1 derives from “homeobox gene expressed in embryonic stem (ES) cells”. It is a homeobox gene on chromosome 3p21.2 and consists of four exons covering about 1.7 kb in total, encoding a protein of 185 amino acids.
- ✓ During the early embryonic development HESX1, as the first of four homeobox transcription factors, is expressed in the prospective forebrain tissue (this later becomes the Rathke’s pouch). It is essential for expression of other factors involved in pituitary organogenesis.
- ✓ Mutations of the HESX1 gene lead to either dominantly or recessively inherited MPHD (Multiple Pituitary Hormonal Deficiency) with a variable combination of anterior pituitary hormone deficits. They can be associated with major central nervous system defects.
- ✓ MRI may show a hypoplastic pituitary gland or signs of septo-optic-dysplasia (de Morsier syndrome).

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 1-4 of HESX1 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 HESX1 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.

Contact:

Prof. Dr. Roland Pfäffle, University Hospital for Children and Adolescents, Oststr. 21-25, 04317 Leipzig. Tel.: +49-341-9726123. Fax: +49-341-9726349. Email: rpfaeffle@medizin.uni-leipzig.de