



Responsiveness to GH treatment

www.endogenet.org

Molecular Genetics Service Profile d3-GHR (Growth Hormone Receptor Gene Polymorphism)

Introduction

- ✓ The GHR gene is encoding the Growth Hormone Receptor, which is expressed in all human tissues. The GHR gene has been mapped to chromosome 5p13-p12, spans approximately 300 kb, and consists of 10 exons, which encode a predicted protein of 638 amino acids.
- ✓ Due to alternative splicing there are 2 isoforms of GHR in humans: a full-length isoform and an isoform that lacks exon 3 (d3-GHR).
- ✓ Patients with a hetero- or homozygous allele encoding the d3-GHR isoform have **increased responsiveness to exogenous Growth Hormone (GH)**. Therefore screening the d3-polymorphism may be useful in SGA (small for gestational age) or ISS (idiopathic short stature) children before treatment with GH to predict the patients individual response.

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ d3-polymorphism screening in patients before treatment with GH to predict individual responsiveness.

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.

Technical

- ✓ DNA amplification using multiplex PCR and agarose gel electrophoresis.

Target turn-round time

- ✓ 1 week from the receipt of all required samples and clinical information.

Cost

- ✓ Allelic variant detection (d3-GHR) - € 60. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- ✓ Dos Santos C et al. "A common polymorphism of the growth hormone receptor is associated with increased responsiveness to growth hormone." Nat Genet. 2004 Jul;36(7):720-4. Epub 2004 Jun 20.
- ✓ Pantel J et al., "Species-specific alternative splice mimicry at the growth hormone receptor locus revealed by the lineage of retroelements during primate evolution.", J Biol Chem. 2000 Jun 23; 275(25):18664-9.

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