



**ISS, Laron Syndrome**  
**Autosomal recessive**  
**Autosomal dominant**

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## Molecular Genetics Service Profile GHR (Growth Hormone Receptor Gene)

### 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).
- ✓ Biologically active growth hormone binds its transmembrane receptor (GHR), the following signal cascade leads to synthesis and secretion of insulin-like growth factor I.
- ✓ Mutations within the GHR gene may lead to autosomal recessive **Laron Syndrome (Growth Hormone Insensitivity)**. Patients may present with high levels of GH and low levels of IGF1 in the presence of “**Idiopathic short stature (ISS)**”.

**Please photocopy and distribute this sheet as required**

### Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected Growth Hormone Insensitivity.

### 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 2-10 of GHR 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

- ✓ Mutation screen (GHR) - € 720. In special cases we will provide this service as part of our research program. Please contact us directly.

### References

- ✓ Woods KA et al. “Phenotype: genotype relationships in growth hormone insensitivity syndrome.” J Clin Endocrinol Metab. 1997 Nov;82(11):3529-35.
- ✓ Laron Z " Laron syndrome (primary growth hormone resistance or insensitivity): the personal experience 1958-2003.” J Clin Endocrinol Metab. 2004 Mar;89(3):1031-44.

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