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TSH deficient hypothyroidism
Autosomal recessive

Molecular Genetics Service Profile

TSH-BETA

Introduction

- ✓ The TSH β gene is encoding the Thyroid-stimulating hormone beta subunit. The TSH β gene has been mapped to chromosome 1p13, spans approximately 4.53 kb, and consists of 3 exons, which encode a predicted protein of 138 amino acids.
- ✓ Mutations within the TSH β gene lead to **congenital isolated TSH deficiency**, this disease is inherited autosomal recessively.
- ✓ Features: **Clinical signs of hypothyroidism, normal/low TSH, low T3/T4, antithyroid antibodies. Imaging studies may reveal a hypoplastic thyroid and/or a hyperplastic pituitary gland.**

Please photocopy and distribute this sheet as required

Reasons for referral

- ✓ Mutation screening in patients with clinically confirmed or suspected TSH deficient hypothyroidism.

Samples

- ✓ Minimum of 2 ml blood sample in EDTA (or minimum of 50 μ g 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 TSH β 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

- ✓ TSH deficiency mutation screen (TSH β) - € 240. In special cases we will provide this service as part of our research program. Please contact us directly.

References

- ✓ Borck G et al. . "Four new cases of congenital secondary hypothyroidism due to a splice site mutation in the thyrotropin-beta gene: phenotypic variability and founder effect." J Clin Endocrinol Metab. 2004 Aug;89(8):4136-41.
- ✓ Doeker BM, Pfaffle RW, Pohlenz J, Andler W. "Congenital central hypothyroidism due to a homozygous mutation in the thyrotropin beta-subunit gene follows an autosomal recessive inheritance." J Clin Endocrinol Metab. 1998 May;83(5):1762-5.

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