Central hypothyroidism (CeH) is a characterized by defective thyroid hormone production due to failure of stimulation by thyrotropin (TSH) with a normal thyroid gland. Disorders of the pituitary gland (secondary hypothyroidism) or the hypothalamus (tertiary hypothyroidism) cause alterations of TSH secretion. CeH can be a part of multiple pituitary deficiencies or can be isolated, its clinical presentation can be moderate or severe. As in primary hypothyroidism a delayed onset of treatment can cause profound neurological defects. Low circulating free thyroxine (FT4) concentrations in patients with low or normal serum TSH make the hormonal diagnosis of the CeH.

Introduction
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Patient report
A 9 year old boy (07 Aug. 20) was referred for proportionate short stature (-2.7 SD). He complained of fatigue and had mild peripheral edema.

### Parameter 
- **T4**: 2.9 mU/l, 2.7-4.7 mU/l
- **TSH**: 0.04 mU/l, 0.04-4.0 mU/l
- **TRH Stimulation Test**: see TSH 0.04 (mU/l) after TRH 2.70

### Reference value
- **Parameter**: Ultrasound of the Thyroid gland
- **TRH Stimulation Test**: Normal Homogenous thyroid
- **MRI of the Sella**: Enlarged “tumor”

### Genetic analysis
The most likely genes involved in central hypothyroidism were sequenced: TSHR, IGFS1 and TRHR and no gene alterations were found. **PROP1** mutations were not tested as the child did not have combined pituitary hormone deficits. Analysis of the **TBXLX and IRX4** mutations in **TBXLX** and **IRX4** are pending.

### Discussion
CeH is usually sporadic and affects patients of all ages. The estimated prevalence is from 1:16,000 to about 1:100,000.

### References