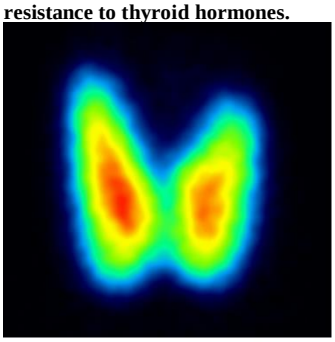
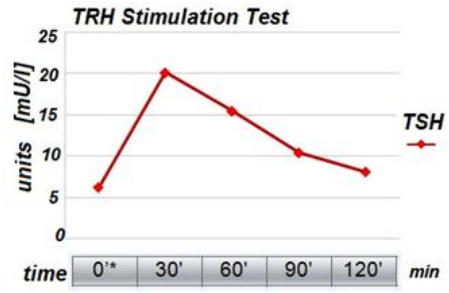
# TREATMENT OF THE RESISTANCE TO THYROID HORMONS IN A GIRL

**Elzbieta Budzynska[1]; Iwona Ben-Skowronek[1]; Elzbieta Korobowicz[2]; Maria Klatka[1]**

1. Department Paediatric Endocrinology and Diabetology Medical University in Lublin, Poland
2. Department Patomorphology Medical University in Lublin, Poland

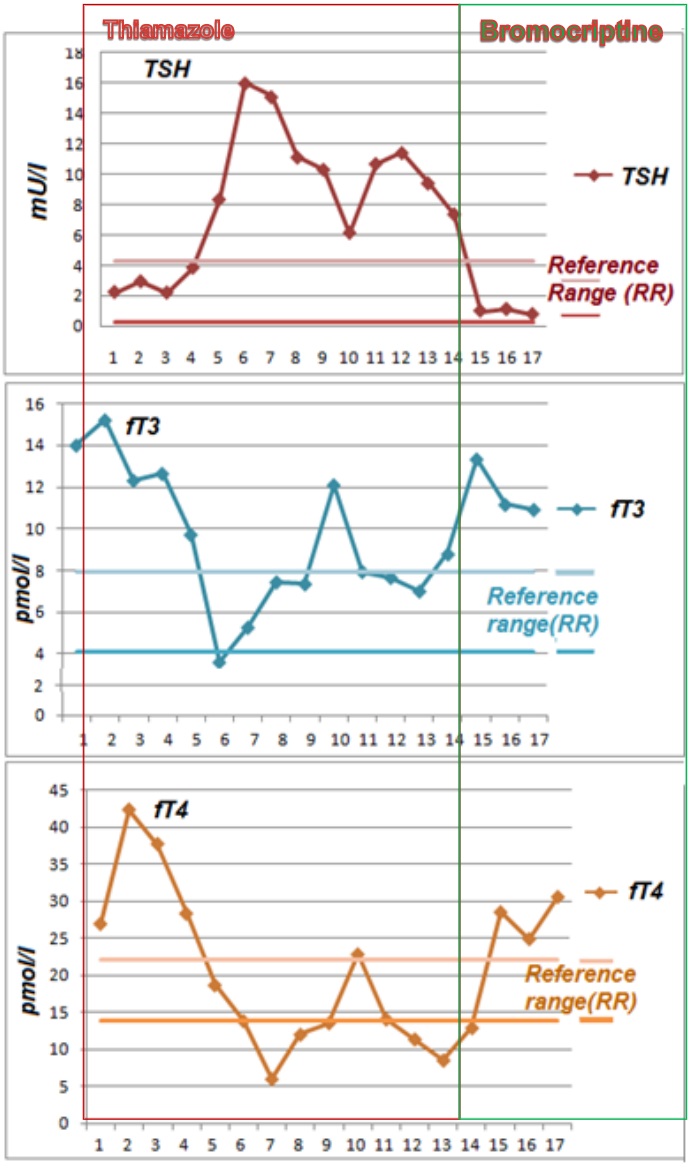
**Background:** The cardinal feature of the resistance to thyroid hormone (RTH) is reduced responsiveness of target tissues to thyroid hormone action caused by thyroid hormone receptor β gene *(THRB)* mutations impairing hormone binding in the majority (90%) of cases. It results in elevated serum levels of free thyroxine (FT4) and triiodothyronine (FT3) associated with unsuppressed thyroid stimulating hormone (TSH).
**The aim of the study is presentation of rare syndrome resistance to thyroid hormones.**






**Case report:** A 11-year old female with Marfan –like phenotype was referred with clinical suspicion of hyperthyroidism She appeared hyperkinetic, complained of palpitation persisted on beta adrenergic blocking agent, which was administered by her cardiologist. The current examination revealed a goiter, exophthalmos and tachycardia. The elevated serum levels of free thyroxine (FT4) and triiodothyronine (FT3) coexisted with unsuppressed thyroid stimulating hormone(TSH). Well responsive TRH test excluded TSH secreting adenoma. A magnetic resonance imaging (MRI) study of the pituitary did not reveal any pathologic mass. In the presence of thyrotoxic signs her treatment with thiamazole was initiated before the final diagnosis of RTH. During the treatment the patient's clinical state significantly improved, while the goiter was increasing in size. The final estimated gland volume based on ultrasonography was 40 ml. Ultrasonography revealed the hypoechoic foci up to 8 mm in diameter. The thyroid scintigraphy with Tc-99m showed excessive uptake with the total suppression of the background activity. The thyroid foci was biopsy-verified (FNAB), and classified in 2nd category by the Bethesda System.
**Genetic testing:**
**Mutation: p.Gly344Glu/-NM_000461.4:c.[1031G>A];[=]TRβ gene was sequenced. The typical mutation for RTH (p.Gly344Glu alternative name p.G344E) was identified in one allele of TRβ gene. Exons 7,9 and 10 of TRβ gene were analysed, with sequencing of codons: 243,310-347 and 417-459.**

Now patient was ordered bromocriptine with cardioselective beta-blocker. We observed involution of the goiter, normalisation of TSH, normal heart rate and well-being.




**The presented patient during thiamazole treatment developed nodular goiter, which is the background to the thyroid cancer under the condition chronic overstimulation by TSH. The elevated cardiovascular risk in RTH, connected to the high levels of FT3 and its influence on isoform THR alpha predominated in the heart. The better effect was noticed during bromocriptine treatment.**