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Background

The pathology of thyroid gland in Ukraine is quite common and in 2018y included 277,708 patients with diffuse goiter (36.47 per 1000 population), 2,311 patients (0.30:1000) with nodular goiter, 341 patients with diffuse toxic goiter (0.04:1000), 7,157 (0.94:1000) with autoimmune thyroid disease and 129 patients with thyroid cancer (1.7:100000).

However the syndrome of resistance to thyroid hormone (RTH) or Familial Dysalbuminemic Hyperthyroxinemia (FDH) are relatively uncommon disorders and have not been previously diagnosed.

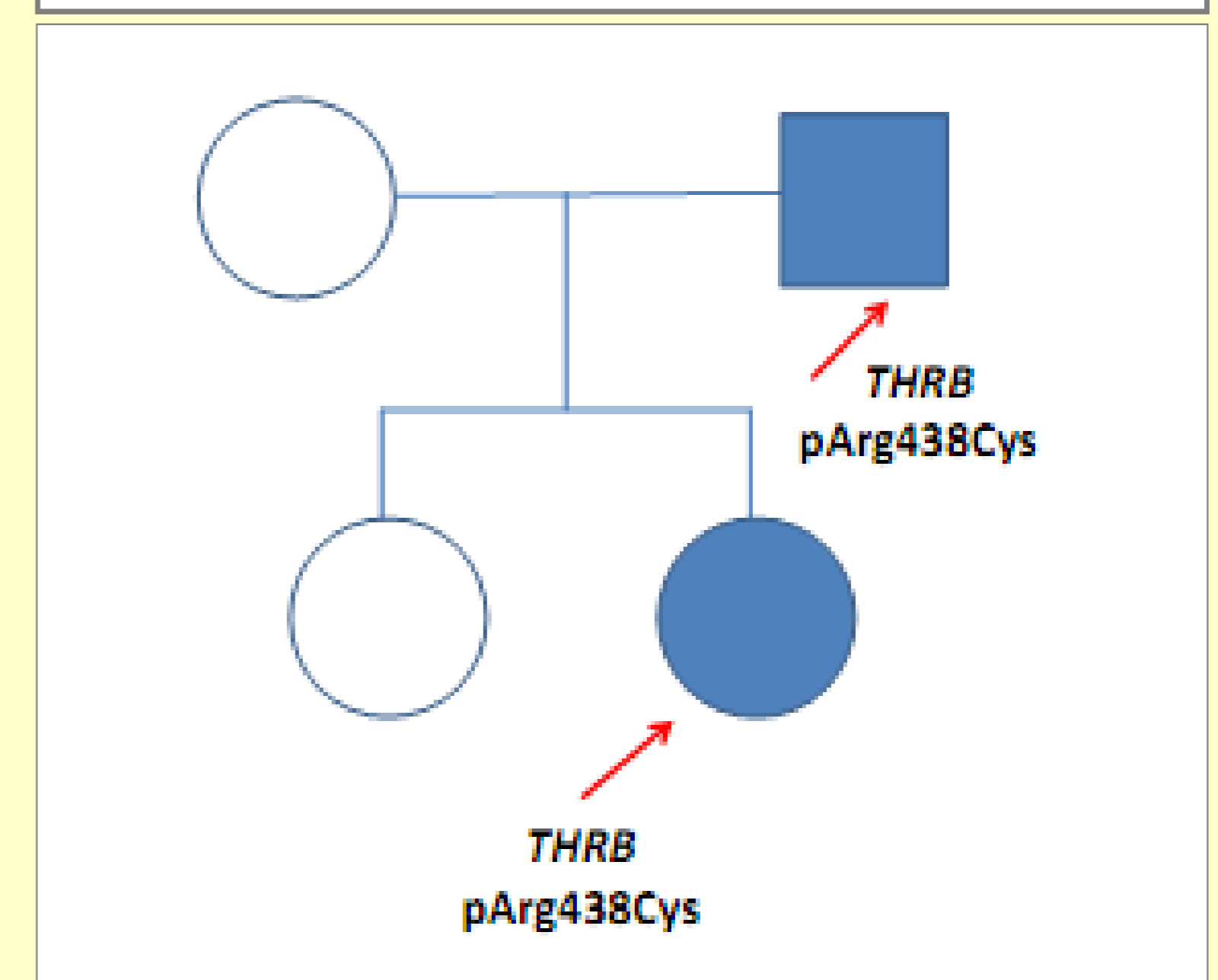
Case presentation 1

A 5.5 y.o. girl presented to endocrinologist with complaints on emotional lability symptoms and hyperactivity disorder. Diffuse euthyroid goiter was diagnosed, and she received iodine supplements within 6 months without improvement of her condition. Hereafter fT4 was measured, and raised level (5.7 ng/dl [normal range, 0.6-1.1]) was revealed on a background of normal TSH. Iodine supplements were cancelled and after 2 months repeated measurements of fT4 and fT3 showed their permanent increased levels (ranged 1.4-2.4 ng/dl and 5.1-6.2 pg/ml [normal range, 2.5-3.9], accordingly) on a background of normal TSH and negative TSH-receptor antibodies (TRAb), Ab-TPO and Ab-TTG levels.

Table 1. Hormone levels in other family members

	fT4 normal range: 0.6-1.1 ng/dl	fT3 normal range: 2.5-3.9 pg/ml	TSH	TRAb	Ab-TPO/ Ab-TTG
Proband	↑ 1.4-2.4	↑ 5.1-6.2	normal	negative	negative
Father	↑ 2.9	↑ 7.4	normal	negative	negative
Mother	normal	normal	normal	negative	negative
Sister	normal	normal	normal	negative	negative

Fig.1. Results. Genetic testing



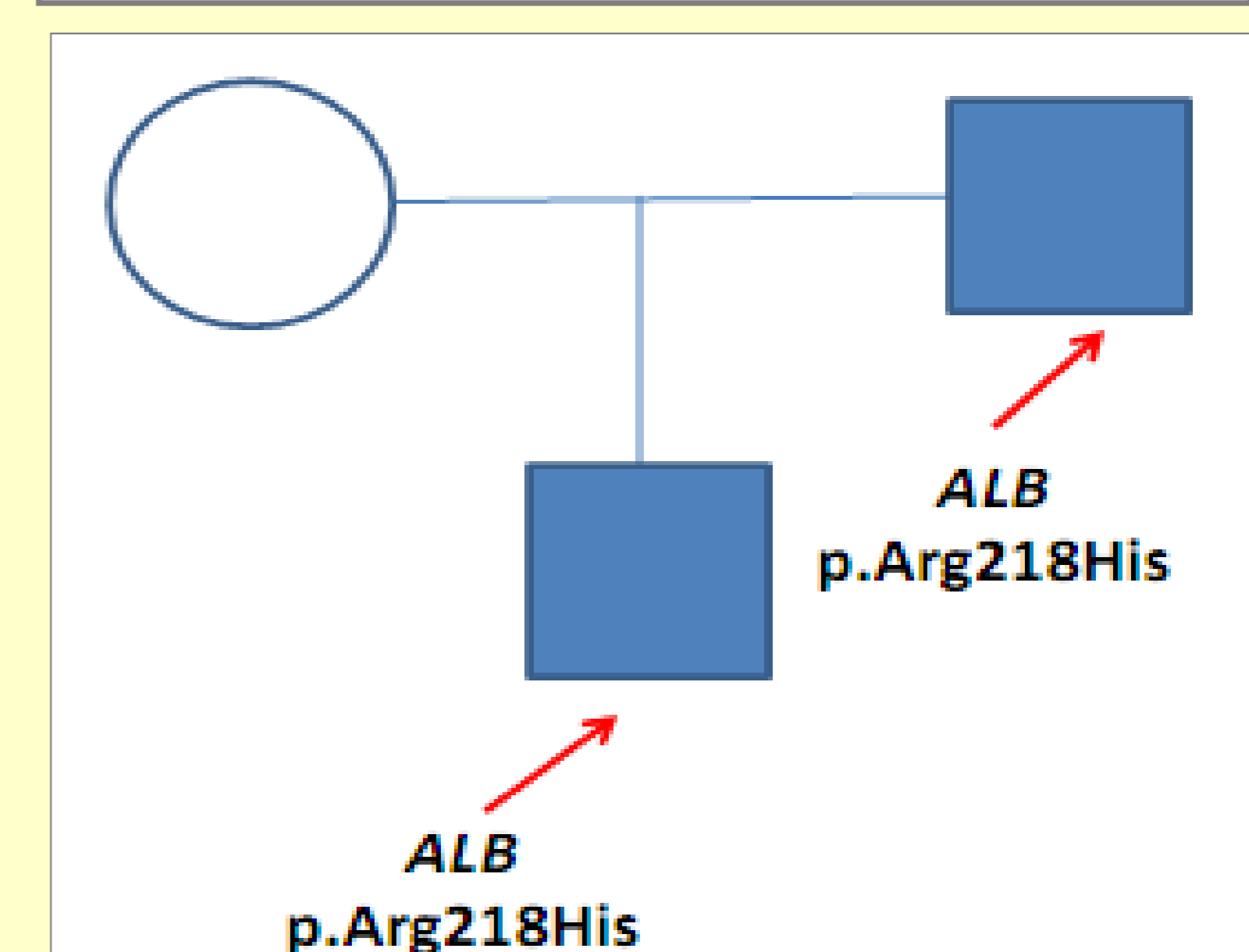
Case presentation 2

A 13.4 y.o. Caucasian boy presented to endocrinologist with complaints of growth retardation since 2 years of age. According to the results of examination his height was 140 cm (-2.4 SD), repeated hormonal test revealed a constantly elevated level of fT4 1.65-2.14 ng/dl [normal range, 0.7-1.46], T4 14.9 mcg/dl [normal range, 6.4-13.4], fT3 4.15-5.16 pg/ml [normal range, 2.4-3.9] on a background of normal TSH, Ab-TPO, Ab-TTG and TRAb. Clonidine test and measurement of IGF-1 level didn't confirm GH-deficiency in a child.

Table 2. Hormone levels in other family members

	fT4 normal range: 0.7-1.46 ng/dl	T4 normal range: 6.4-13.4 mcg/dl	fT3 normal range: 2.5-3.9 pg/ml	TSH	TRAb	Ab-TPO/ Ab-TTG
Proband	↑ 1.65-2.14	↑ 14.9	↑ 4.15-5.16	normal	negative	negative
Father	↑ 1.73	normal	normal	normal	negative	negative
Mother	normal	normal	normal	normal	negative	negative

Fig. 2. Results. Genetic testing



Conclusions

Patients with euthyroid hyperthyroxinemia should undergo genetic testing due to the similar clinical/hormonal presentation of RTH and FDH syndromes.

