

# Case report of familial adenomatous polyposis – Gardner syndrome, thyroid papillary carcinoma T1b, total thyroidectomy, post-operative, compensated hypothyroidism.

Ieva Strauhmane-Brence<sup>1</sup>, Gunda Zvigule-Neidere<sup>1</sup>,  
Ieva Kravale<sup>1</sup>, Iveta Dzivite-Krisane<sup>2</sup>, Inita Kaze<sup>2</sup>

1Rīga Stradiņš University, Faculty of Continuing Education, Latvia  
2Children Clinical University Hospital, Paediatric Clinic, Latvia

## **Introduction**

Familial adenomatous polyposis (FAP) is an autosomal dominant hereditary disease characterized by the formation of more than 100 adenomas in the large intestine. The prevalence of FAP worldwide is 2-3 cases per 100,000 people. It is important that the adenomas in the large intestine are formed in the early days, often in childhood and adolescence. Depending on the phenotypic nature of this disease, variants such as typical or classic FAP, Gardner syndrome, the mildest manifestation type, and Crail syndrome (formerly known as Turco Syndrome) can be categorized. The development of various disease variants is closely related to various APC gene mutations. APC gene mutations are found in 80-90% of hereditary FAP, but up to one third of new cases cause a new mutation or mosaic without a positive family history. MUTYH gene changes are also associated with a similar clinical picture, especially the easiest manifestation type. The APC gene has raised the inhibitory gene, thus causing pathological changes in its structure to promote tumors.

## **Case report description**

On the visit of endocrinologist in Children's Clinical University Hospital came 16-year-old girl to evaluate and treat post-operative hypothyroidism. The patient has complaints about periodic pain in the neck of the scar area. From an anamnesis it is known that in December 2016, a patient was diagnosed thyroid papillary carcinoma at stage T1b. 07.12.2016. extirpation of total right thyroid lobe on is complete, after receiving a histological answer on March 27, 2017 - total extirpation of the left thyroid lobe. The girl is taking 25 µg levothyroxine once a day in the morning. When evaluating laboratory performance, hypothyroidism is uncompensated. It is recommended to increase the dose of levothyroxine to 75 µg once daily and repeat the US thyroid gland one month later.

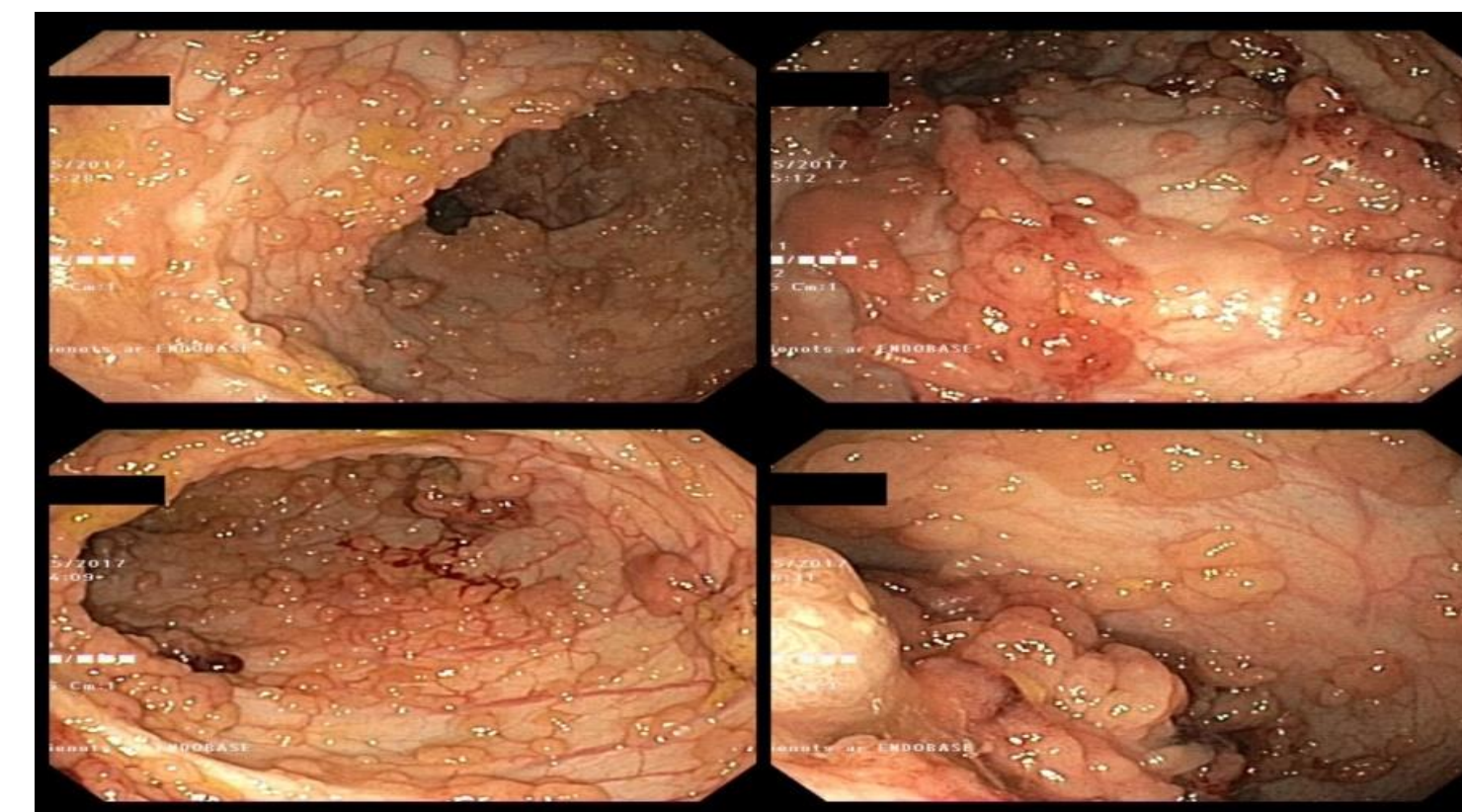
## **Anamnesis of disease**

After birth – the girl was found a congenital abnormal formation in the left arm of the right muscle. Radical surgery was not possible. Pathologistological response - aggressive fibromatosis, extra-abdominal desmoid tumor, infantile variant. Further treatment was discussed at the congress of hematologists and oncologists, which decided to start chemotherapy following the COS protocol. At seven months of age, the girl was subjected to a partial desmotomy tumor excision.

At the age of ten, a girl was found formation in the left gluteal region. Diagnosis - Liposus regio glutei sinistra. The patient has a colon prolapse of 15 years of age. Surgical treatment was performed. Colonoscopy conclusion - tubular adenoma structures of the large intestine. (Picture nr.1) After few month the patient was hospitalized in a regional hospital with complaints of headache, dizziness, chills, subfebrile temperature, decreased appetite. Thyroid ultrasonography is a solitary cystic formulation in the right lobe, suspected of malignancy. Laboratory tests performed, hemoglobin 8.9 g/dl found, thyroid function of the euthyroid. After a couple of months, the patient undergoes total thyroid gland extinction due to papillary carcinoma in the histological material (T1b). There is a suspicion of family adenomatous polyposis - Gardner syndrome. Blood sample for APC gene examination sent for confirmation of the diagnosis.

In control analysis was detected an iron deficiency anemia (HGB 8.2g/dl, ferritin 3.91 ng/ml), serum calprotectin 161.8 µm/g, secondary hyperparatiroidism (PTH – 8.6 pmol/l, total vitamin D 9.57ng/ml, hypothyroidism, wich diminishes in a dynamic manner after increasins levothyroxine dose.

Picture nr.1



## **Discussion**

This clinical case is manifested by the atypical manifestation of first symptoms and the lack of clinical classical symptoms of familial adenomatous polyposis, such as the appearance of blood in the faeces, fluid abdominal pain, abdominal pain. The first manifestation of the patient's family of adenomatous polyposis was the desmoid tumor at infancy. Desmoid tumor is rare in the general population - about 5 to 6 cases per million people per year. Identifying family members of adenomatous polyposis and performing a targeted mutation assay for APC gene family members has significantly improved the survival of these patients.

## **Contact information**

Ieva Štrauhamen-Brence, MD, 1st year endocrinology trainee

E-mail:ieva.strauhmane@inbox.lv

