

First Experience of Prophylactic Thyroidectomies in Children

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Introduction. Prophylactic surgical treatment can be offered for carriers of inherited mutations in order to decrease the cancer risk. Such prophylaxis gains an even higher importance if the related malignancy is known to be aggressive and/or difficult to diagnose in early stages. However, the approach necessitates the removal of seemingly healthy tissues; therefore, discussions can arise, especially if the prophylactic treatment involves children. In addition, the genetic testing in children has been the matter of discussions.

Multiple endocrine neoplasia (MEN) 2A is a rare familial cancer syndrome caused by mutations in the *RET* proto-oncogene. The penetrance is 70% by the age of 70 years, and the manifestations include medullary thyroid carcinoma, pheochromocytoma, and parathyroid hyperplasia. Total thyroidectomy has been recommended for patients as young as three years of age if the presence of inherited mutation is proved [Richards, et al., 2012].

Aim. The aim of the present study is to report the first two cases of prophylactic thyroidectomies in children due to molecularly approved multiple endocrine neoplasia type 2A syndrome in Latvia.

Material and methods. In order to accomplish the set aim, the medical history was re-evaluated, analysing the pedigree, results of genetic tests, surgical treatment as well as laboratory and histological findings.

Results. Initially, MEN2A was suspected in a young lady who underwent laparoscopic transperitoneal left-sided adrenalectomy for multifocal pheochromocytoma featuring an elevated risk of malignant behaviour. In addition, total thyroidectomy was performed for multifocal medullary cancer. Genetic investigation revealed an inherited *RET* gene mutation. Other family members underwent genetic testing as well, and the mutation was identified in 3 of 5 sisters. The mutation carriers were detected in 6 children. After informed consent was received from the parents, 2 children were found to be mutation carriers, but three were free of genetic defect. A newborn baby was not tested yet. Examination of *RET* gene revealed mutation in codon 791 exon 13. In 2013, total thyroidectomies were performed for two cousins aged 5 and 6 years. Basal calcitonin level before the operations was 11 and 18 pg/mL, respectively. Seven and five months after thyroidectomies, the patients were clinically and biochemically well. After the surgical treatment, the basal levels of calcitonin in blood were less than 2 pg/mL. Calcium levels were within normal range, and voice cords were working properly. At present, patients receive thyroid hormone substitution therapy and regular follow-up.

Conclusions. Prognosis of syndrome is related to the stage-dependent prognosis of hereditary disease indicating the necessity of early thyroidectomy for screened at risk subjects.