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MEN 2A SYNDROME – MULTIPLE ENDOCRINE NEOPLASIA WITH AUTOSOMAL DOMINANT TRANSMISSON

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Introduction: Multiple endocrine neoplasias (MEN) are rare and complex autosomal dominant inherited syndromes caused by germline RET mutation and characterized by the association of tumors of two or more endocrine glands in the same patient. It occurs in 1:30000 individuals, and is reported in approximately 500–1000 families worldwide. Sipple syndrome (MEN2A) is characterized by the association of medullary thyroid carcinoma (MTC) (in 80-100% of cases), uni- or bilateral pheochromocytoma (in over 50% of cases) and primary hyperparathyroidism (15% to 30% of cases) in the same patient. The diagnosis can be confirmed by imaging methods and early lab tests (calcitonin, methanephrines). Genetic confirmation is mandatory when the Sipple syndrome is suspected. Laparoscopy is the choice of approach in surgical treatment.

Keywords: Multiple endocrine neoplasia, Sipple syndrome, medullary thyroid carcinoma

Case report: The patient, a 20-years old with MEN 2A syndrome, which has been manifested by bilateral pheochromocytoma and medullary thyroid carcinoma. It was a familial form, having first degree relatives (mother) with pheochromocytoma. Genetic testing was not performed due to the death of the first degree relatives. After a full check-up (imaging methods, lab tests) adrenal and thyroid gland tumors were detected. The patient underwent laparoscopic adrenalectomy on her left in 2015, on the right in 2019 and total thyroidectomy in 2020. Histopathological examination of the adrenal glands revealed multicentric alveolar pheochromocytoma, the thyroid tissue - non-encapsulated medullary carcinoma. The postoperative evolution is favorable. She is undergoing hormone replacement therapy. Recent tests (25.05.20): PTH - 19,3 pg/ml,TSH- 0,2ul/ml, ionic calcium – 1,09 mmol/l, calcitonin decreased from 121 to 18.7 pg/ml.

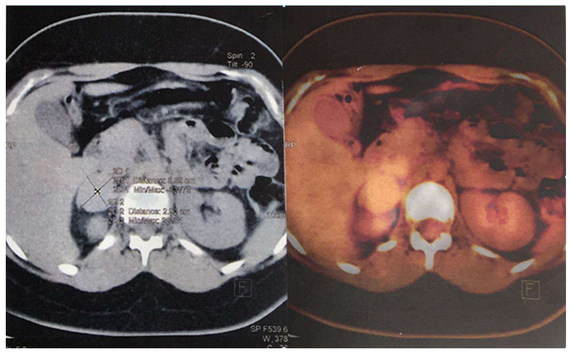

Fig.1 PET-CT: Increased metabolic activity of FDG at the level of a tumor of the right adrenal gland (36x 29mm)

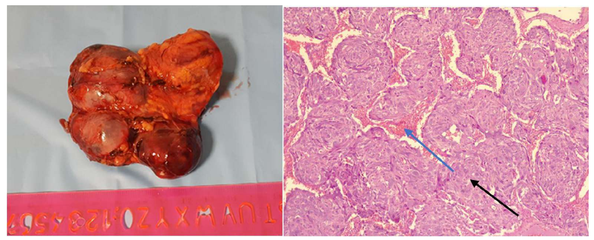

Fig.2 (A, B) A - macroscopic view of multinodular solid adrenal tumor, the biggest was 41x32 mm. B - Photomicrograph (x10; hematoxylin-eosin stain). Multifocal pheochromocytoma of adrenal medulla with alveolar pattern of growth (Zellballen architecture) (black arrow) and rich vascular network (blue arrow).

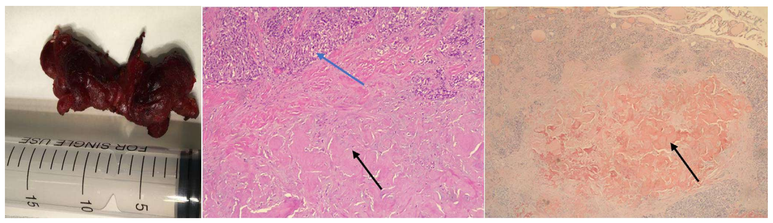

Fig.4 A.Macroscopic view of the thyroid gland, 8 x 3 cm
B- x10; hematoxylin-eosin stain. Medullary thyroid carcinoma with trabecular pattern of growth. 1.Amyloidosis of the stroma (black arrow). 2. Plasmacytoid tumoral cells with round nuclei (blue arrow).C- (Congo red stain). Positive reaction for amyloid in tumoral stroma (black arrow).

Conclusions: The radical approach to MEN 2A syndrome is very important from both a therapeutic and surgical point of view. Imaging check-up in combination with annual monitoring of calcitonin, chromogranin A, and metanephrines in a patient with MEN 2A syndrome is a practical way to supervise the case and make timely decisions for surgical intervention and to prevent complications. If a pheochromocytoma is detected, adrenalectomy should be performed before thyroidectomy or other surgery to avoid intraoperative catecholamine release. Laparoscopy is the choice of approach in surgical treatment. Limitations only arise because of technical difficulties or tumor size.

Table 1. Laboratory tests 2014-2020 years

Laboratory tests	29.12.14	30.06.18	08.10.18	13.11.18	15.01.20
Cortisol	449,9(171-536)	318,2(171-536)			
ACTH	55,33 (7,2-63,3)				165,3
Aldosterone	21,2 (1,76-23,2) Supine (2,52-39,2) standing				
Vanilmandelic acid	26,45 (1-11)children (0,1-0,18) adults				
Phosphor	1,24 (1,09-2)				
PTH	30,8 (15-65)				42
Calcium ionic	1,06 (1,05-1,3)				
Calcium	2,39 (2,3-2,75)				
Sodium		136,8(135-148)			
Potassium		4,81 (3,5-5,3)			
Anti-TPO	15 (up to 34)				
T3	2,4 (1,4-3,34)				5,7
T4	115,4 (76,1-170)				
TSH	2,52 (0,51-4,3)	2,14 (0,51-4,3)			3,06
FT4		13 (12,6-21)			
creatinine		59 (up to 88,4)			
urea		3,4 (up to la 8,3)			
metanephrines			571 (up to 375)		
normetanephrines			1197 (up to 550)		
3-metoxi-tiramina			75,7 (up to 460)		
Chromogranin A				263 (27-94)	
Calcitonin				121(norm <10 pg/mL)	62,8

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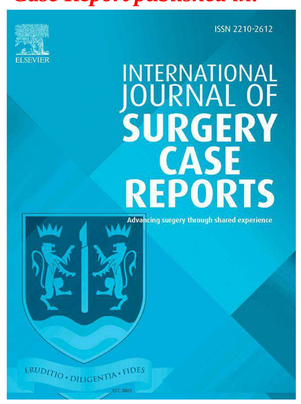

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