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Paraaortical paragangliomas as incidental findings in a female adolescent

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Introduction

Paragangliomas are neuroendocrine tumors rarely present in children and adolescents usually in a syndromic form. They origin from neural crest chromaffin cell lineage and they extraadrenally located mainly across the spine. Paragangliomas produce catecholamines, like dopamine, norepinephrine, and epinephrine. Head and neck located paragangliomas are more often of non – secreting, of parasympathetic origin. Abdominal paragangliomas in contrast tend to be sympathetic with catecholamine secretion potential. Paragangliomas are commonly associated with MEN IIa and IIb, von Hippel-Lindau Syndrome, neurofibromatosis type I, Carney's triad and other hereditary pheochromocytomas/ paragangliomas syndromes.
Approximately 10–20% of all paragangliomas are diagnosed during childhood with mean age at diagnosis about 11 years-old approximately. The case of a Greek girl with bilateral abdominal paragangliomas and familial multiple exostosis syndrome is presented.

Case Presentation

A 13.5-year-old girl was presented for investigation of two large, nodal, oval shaped para-aortic lesions incidentally found during abdominal U/S for menstrual cycle disturbances.
Perinatal medical history revealed:

- 2nd child, gestational age: 39 weeks
- caesarean section due to failure to progress in labor
- birthweight, length and head circumference in 50th percentile for gender

Medical history of the girl included:

- non-autoimmune hypothyroidism since 10 year-old and treated with levothyroxine
- multiple exostosis disease
- Dyslipidaemia
- menarche at 12.5 years old- normal menses except the last two months

Family history revealed:

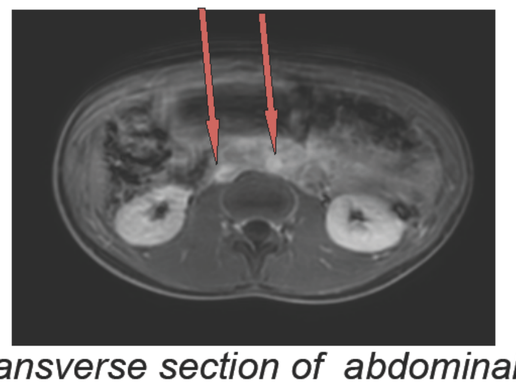
- multiple exostosis disease in mother, maternal grandfather and brother
- dyslipidaemia in father and maternal grandmother
- hypertension and diabetes mellitus type 2 in maternal grandmother.

Physical examination was normal, with symmetrical anthropometrics (25th percentile), pubertal Tanner stage IV and normal vital signs.
The girl presented multiple exostotic lesions in upper and lower extremities and thorax, along with a café-au-lait spot in left iliac fossa.
Full blood count and serum biochemistry were within normal.

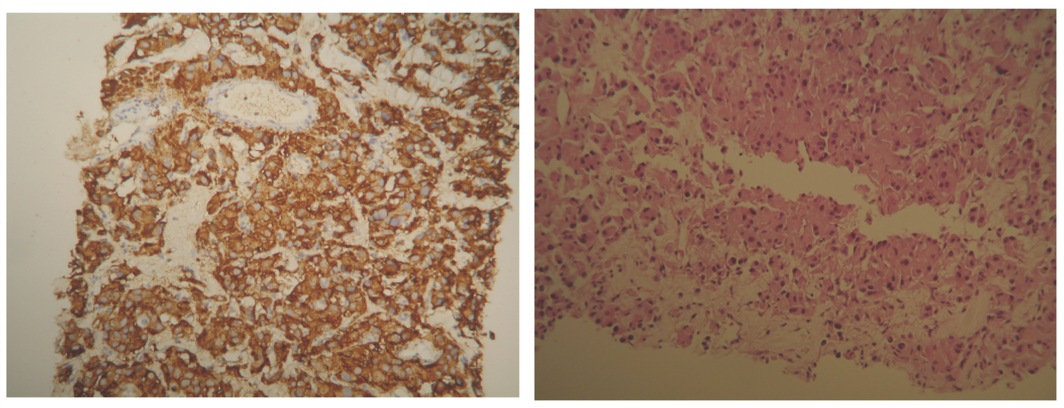
Hormonal evaluation revealed euthyroid status (under substitution therapy with levothyroxine) with negative thyroid autoantibodies. Levels of PTH, PRL, DHEA-S, ACTH, FSH, LH, E2, renin, calcitonin and thyreoglobulin were within normal range for her age. Negative values for AFP, β-hCG, CEA were found.

24-hour urine vanillylmandelic acid (VMA) levels were found marginally elevated (11.09 mg/24h) in one time-point evaluation but normal in re-screening (5.78 mg/24h).

Abdominal MRI revealed two bilateral para-aortic lesions at the level of renal arteries and CT-guided biopsy was performed (Picture 1). Histology established the diagnosis of benign retroperitoneal paraganglioma (Picture 2).

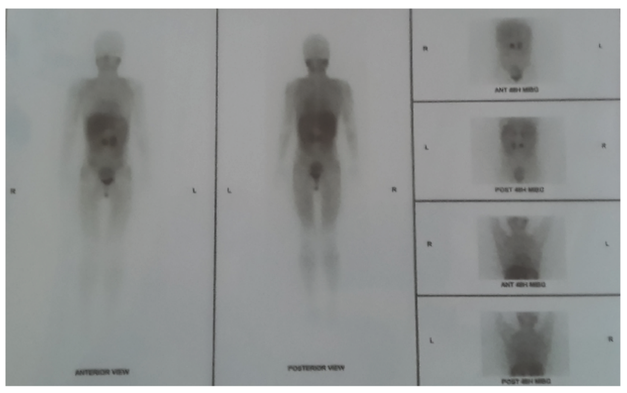


Picture 1: Transverse section of abdominal MRI



A *B*

Picture 2: Biopsy pathology
A: Immunohistochemical positive stain for synaptophysin x 200
B: HE x 200



Picture 3: 123I-MIBG images

Functional imaging with 123I-MIBG single-positron emission computed tomography confirmed the presence of two paragangliomas without secondary lesions found (Picture 3). Additional somatostatin receptor scintigraphy with Tc-99m-Tektrotyd didn't reveal any further MIBG-negative lesions. Scheduled genetic testing is expected to clarify possible genetic substrates.

Conclusions

Paragangliomas although are rare childhood tumors have a significant potential for malignancy. Accurate diagnosis and removal is important in this aspect. The combination of multiple paragangliomas and multiple exostosis disease, to our best knowledge has not been previously reported

References

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