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Adrenal and Steroids

Epidemiology of PPGLs – A population based approach

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Phaeochromocytoma/paragangliomas (PPGLs) are relatively rare tumours and the health burden of such tumours is not very well known.

Aim: This population based study aims to characterise all the phaeochromocytomas, paragangliomas and adrenal medullary hyperplasia diagnosed between 2007 and 2016 in Malta; looking into presentation, hormonal analysis, imaging characteristics and

21–62 years (mean 50±14). The standardised incidence rate is 4.3/1,000,000/year. From the whole cohort 11 (69%) had phaeochromocytomas confirmed histologically, 3 (19%) had paraganglioma, and another 2 patients (12%) had adrenal medullary hyperplasia (adrenal medullary cell mass hyperplasia <1 cm, thought to be a precursor of phaeochromocytoma). 9 patients (56%) presented with hypertension, whereas 6 patients (38%) were found following investigation of an adrenal incidentaloma. All patients except 1 had either plasma free metanephrines or urinary fractionated metanephrines checked prior to surgery. In the phaeochromocytoma and adrenal medullary hyperplasia patients, CT was documented to be suggestive of phaeochromocytoma or an adrenal lesion not in keeping with an adenoma in 11 out of 13 patients (85%). Longest radiological tumour size ranged from 20–127 mm (mean 52±28.9) All patients except 2 underwent surgical resection of the tumours. The latter 2 patients presented late with metastasis and died soon after diagnosis. Genetic testing was done in 6 patients (38%) and a VHL mutation was identified in one patient with phaeochromocytoma. 6 patients (38%) were found to have a malignant phaeochromocytoma on follow up.

Conclusion: This review highlights the extensive workup needed for patients with PPGL. Presentation can range from asymptomatic to life threatening clinical conditions. The high risk of malignancy found in our cohort emphasizes the need for long term follow up.

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