

Noninfectious but Genetic Bilateral Neck Swelling



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Background:

Head and neck paragangliomas (HNPGs) are rare tumors mostly arising from the parasympathetic paraganglia such as the carotid body. They are usually detected because of local mass effects or during the work-up of a genetic syndrome. We describe a case of a 32 year old woman with bilateral carotid body tumors.

Case Report

A 32-year old previously healthy mother of 3 children was referred because of bilateral neck swelling, presumably lymphadenopathy, associated with recurrent upper respiratory tract infections during the past year. Cervical ultrasound raised the suspicion for bilateral carotid body tumors which were confirmed by MR-imaging and a subsequent DOPA-PET-CT, which showed no evidence for multifocality or metastases. Plasma metanephrines including methoxy-tyramine were within the normal range and although a detailed family history revealed no evidence for a syndromic presentation, the genetic work-up revealed a SDHD mutation and the diagnosis of paraganglioma syndrome typ 1. Because of impending mass effects and the prospect of a curative approach according to the result of an MR-angiography, resection of the tumors has been performed.

Discussion:

HNPGs are rare tumors (incidence 1:30'000 – 100'000) derived from extraadrenal chromaffin cells and 95% arise from the parasympathetic nervous system, typically the carotid body (glomus caroticum), jugular bulb (glomus jugulare) and different branches of the vagal nerve (glomus vagale, glomus typanicum). Although plasma methoxytyramine may be elevated in up to one third, they are clinically nonsecretory in most cases and present because of their mass effects. Around 35% are associated with a genetic defect, with SDHD-mutations accounting for > 50%, SDHB for 20-35% and SDHC for 15%. Mutations in other PHEO/PGL susceptibility genes such as SDHAF2 are exceptional. The inheritance pattern is autosomal dominant for SDHx mutations, but in SDHD and SDHAF2 the disease occurs only, if the mutation is inherited from the father, which is in line with maternal imprinting (=imprinted and not expressed in the offspring). Malignancy is rare (3.5%) although metastases occur up to 20 years after initial diagnosis. For both diagnosis and detection of multifocality and/or malignancy functional imaging with 18F-DOPA- or 68Ga-DOTATATE-PET-CT is considered superior whereas anatomical imaging with CT and/or MRI gives a better locoregional resolution. Surgical resection should be attempted in referral centers but may be associated with significant morbidity including cranial nerve injuries. The requirement for perioperative alpha-adrenergic blockade is assessed by biochemical testing. Radiotherapy/surgery can be applied to non-resectable symptomatic tumors and chemotherapy is reserved for advanced/progressive disease. Genetic counselling and testing is mandatory due to the high rate of germline mutations and familial disease.

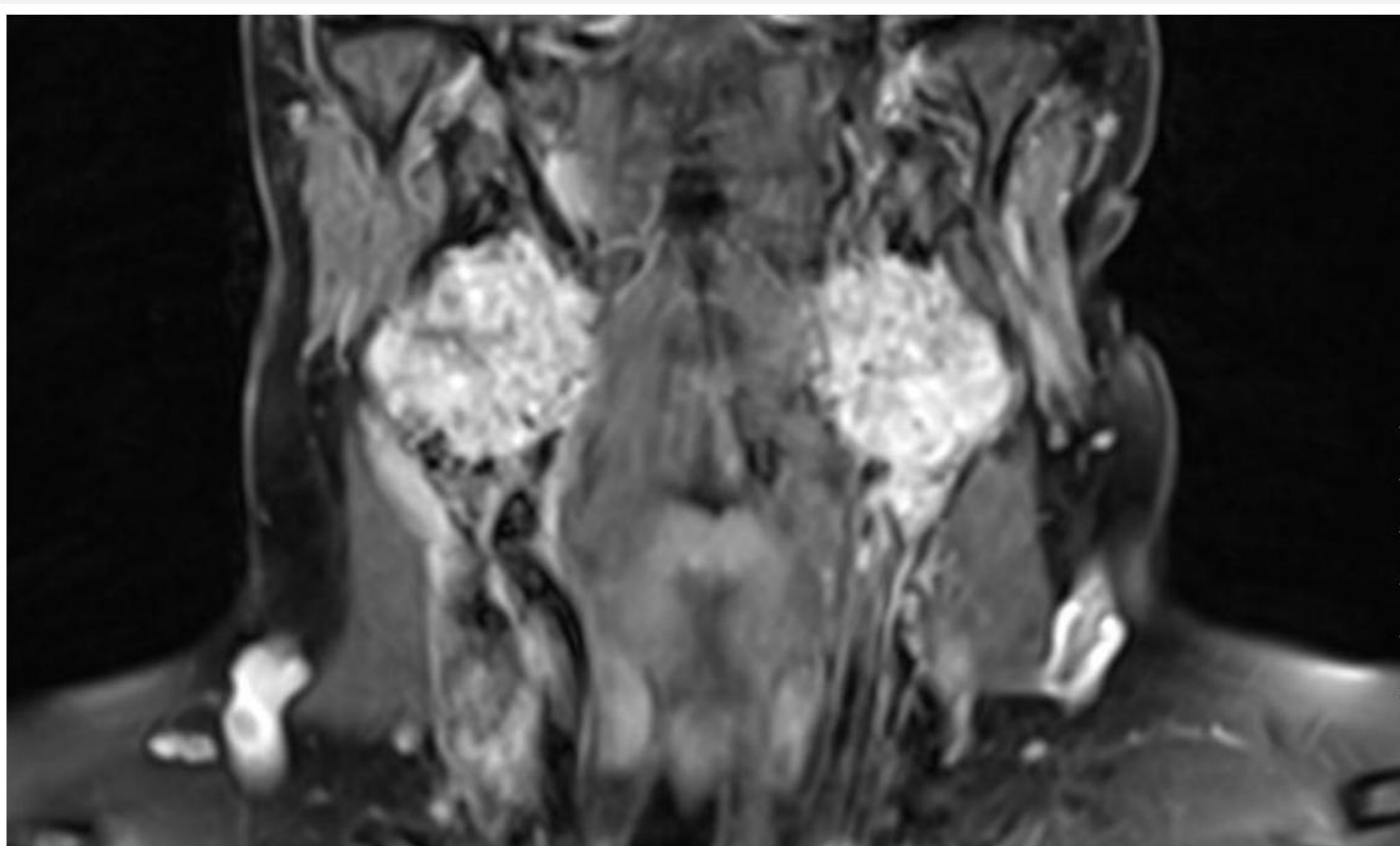


Fig 1: Coronal T2-weighted MR-scan of the neck showing bilateral contrast enhancing mass lesions

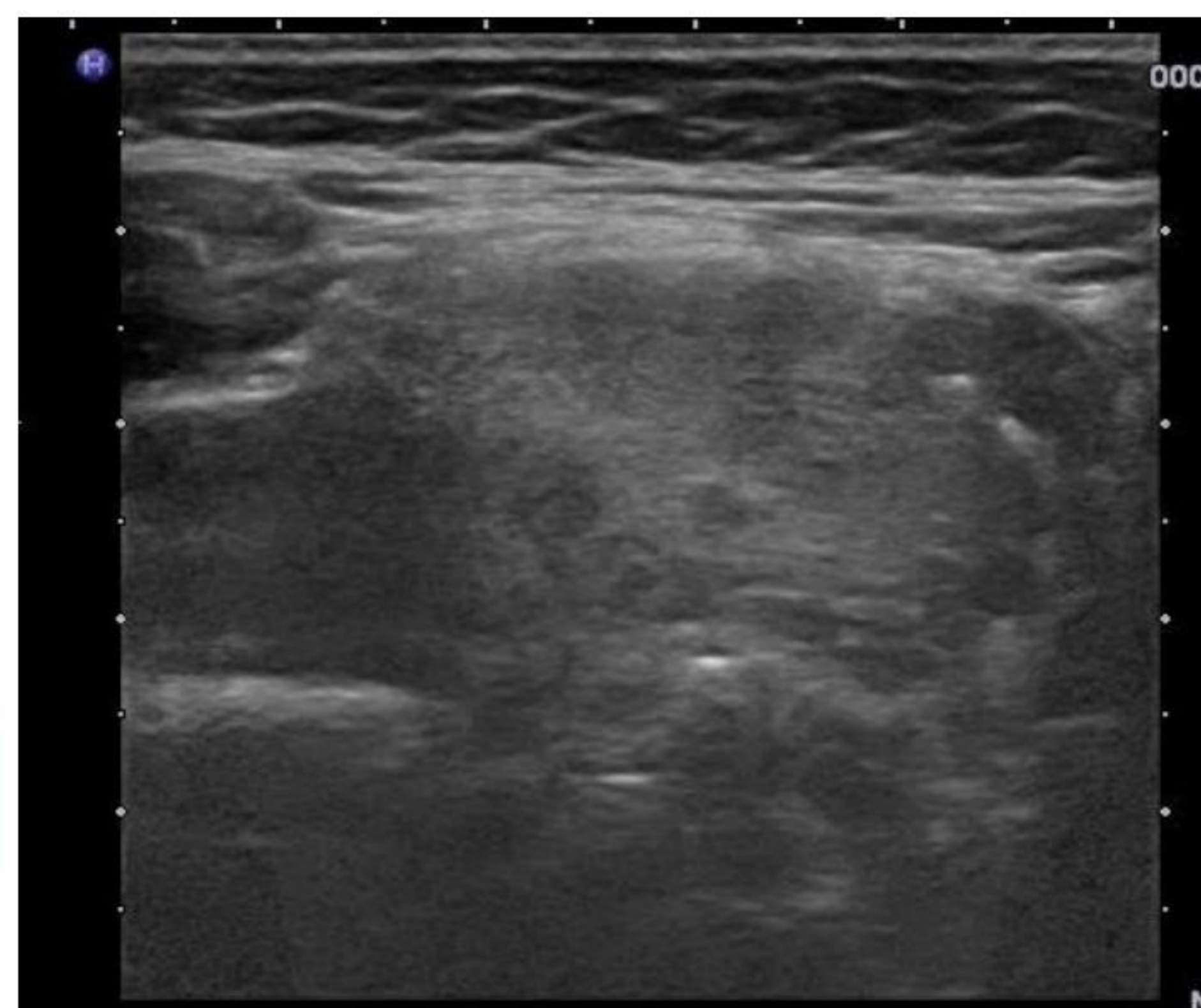


Fig 3: Transverse ultrasound scan of the right neck showing mildly hypoechoic, inhomogenous, irregular shaped mass

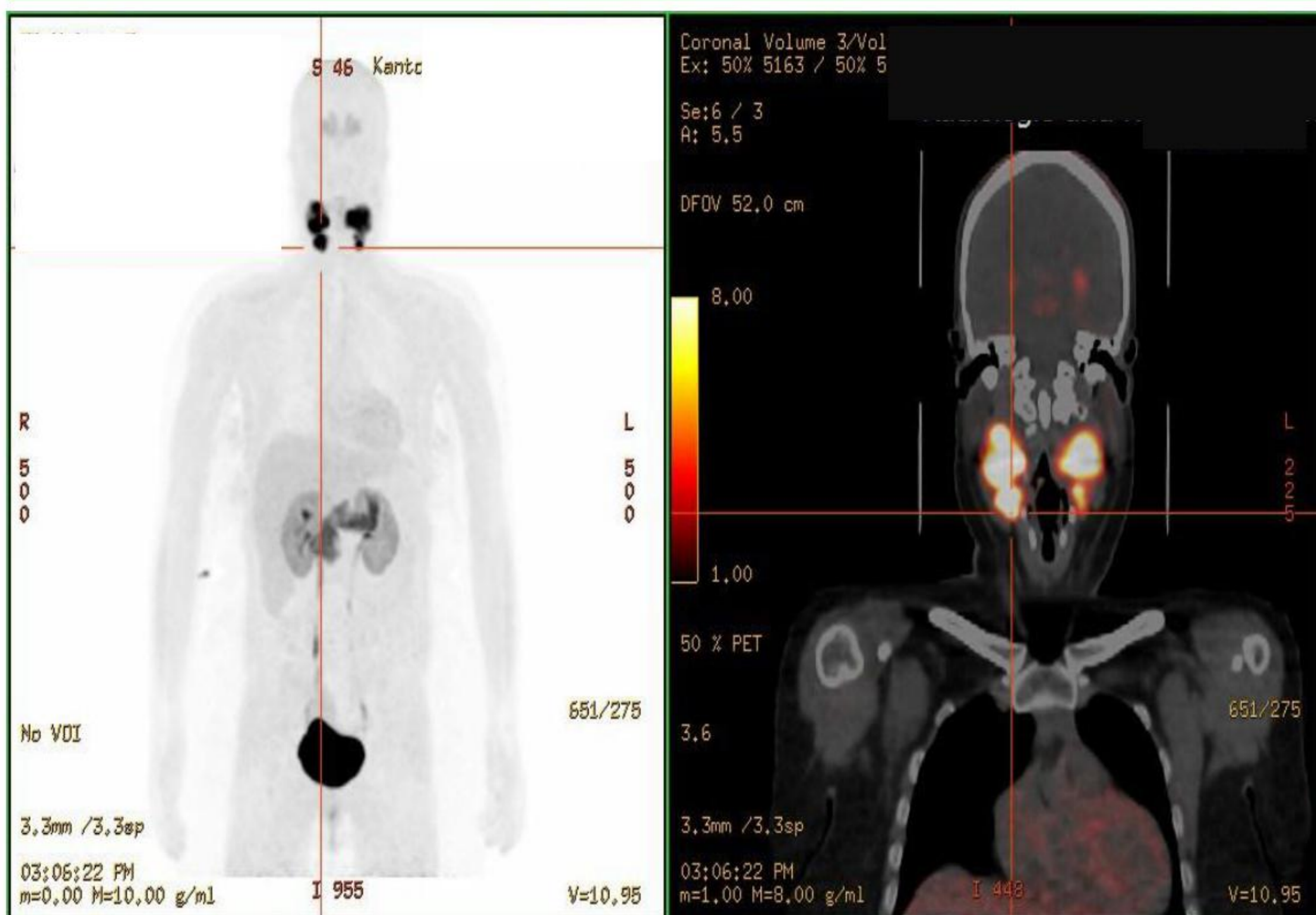


Fig 2: 18F-Fluoro-DOPA PET CT showing intense tracer uptake in the bilateral neck masses but no other foci indicative of Paragangliomas

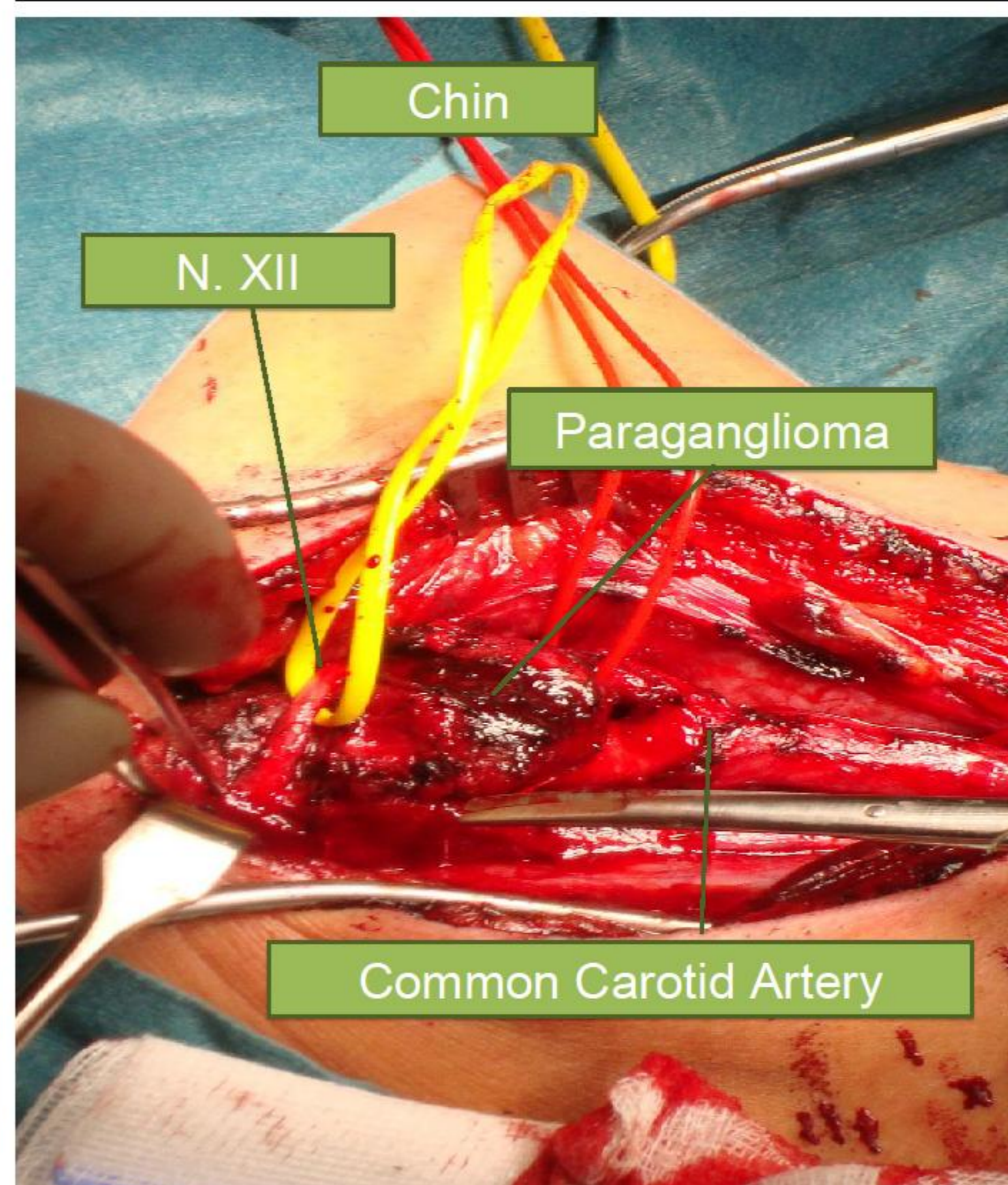


Fig 4: Intraoperative view during resection of the right-sided paraganglioma

References

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