Viva

Intercollegiate Exam – Viva – Carotid Body Tumour

Abstract

Carotid body tumours are one of the more uncommon causes of a lateral neck lump. It is important to have a logical approach to the diagnosis and management of all neck masses. Clinicians must also be able to pick up on more subtle signs that allow differentiation between overt malignancy and other, rarer tumours. This will ensure appropriate investigation and management of what is a relatively rare clinical problem.

Keywords

Carotid body tumour, paraganglioma, malignant, familial, management.

Introduction

Questions regarding the investigation and management of neck lumps are extremely common in the intercollegiate viva examination. It is important to have a logical and stepwise progression from initial history and examination findings to eventual diagnosis and treatment. Although the diagnosis of cancer is always a priority not all neck lumps in the examination will turn out to be malignant and it is important to consider each case on its own merit.

Describe what you see? (Figure 1)

This is a clinical photograph of a right lateral neck mass at level II. There do not appear to be any overlying skin changes associated with the mass.

What symptoms would you ask about?

You should divide your questions into two groups. The first set of questions should concentrate on the mass itself, the second should enquire about any other head and neck, or systemic symptoms that may aid your diagnosis.

Questions about the mass include:

Duration of swelling, rate of growth or resolution, pain/tenderness and associated skin changes in the area.

Other symptoms that should be elucidated include: Dysphonia, dysphagia, referred otalgia, persistent sore throat, difficulty breathing, hemoptysis, fever, sweats, nasal obstruction, nasal discharge, weight loss and diminished appetite. A systemic enquiry

Figure 1: 45-year-old gentleman with right lateral neck swelling.
for other associated symptoms such as flushing, palpitations, loose stools and tremor is also appropriate. Enquiry about relevant past medical history, medication, social history and family history should also be made.

This particular patient is 45 years old and reports a 12-month history of a slowly enlarging neck mass, which is painless. He has no associated symptoms from his head and neck but mentions occasional episodes of flushing and palpitations over the last few months.

What examination would you perform?

As well as examining the neck, a thorough examination of the oral cavity, oropharynx, anterior nose and flexible nasolaryngoscopy should be performed. With respect to the mass itself, size, mobility, associated skin changes; lymphadenopathy and bruits should be elucidated. You should also ascertain whether it will transilluminate, whether it is fluctuant and whether it is pulsatile. A full cranial nerve examination will also be necessary.

In this particular case, the salient positive findings of the examination are a 3 cm pulsatile, expansile mass in right level II. There is lateral mobility of the mass, but it doesn’t transilluminate and there is no associated bruit or lymphadenopathy. The rest of the head and neck examination is completely normal.

What is your differential diagnosis?

- Paraganglioma – Carotid body tumour
  - Glomus vagale (unlikely to be palpable unless huge)
- Glomus jugulare
- Other vascular lesion – High flow vascular malformation
- Head and neck malignancy
- Lymphoma
- Branchial cyst
  (Non-vascular lesions may still feel pulsatile if transmitting carotid pulsations)

What investigations would you perform and why?

- MRI scan – This is superior to CT in giving information regarding the soft tissue characteristics of the mass. Paragangliomas typically have a ‘salt and pepper’ appearance on MRI scanning. This is due to flow-voids within in the lesion. MRI gives better resolution of soft tissue planes and may give increased information about the relationship of the mass to the great vessels. It can also identify associated lymphadenopathy and may be helpful delineating primary tumours especially within deeper structures. Its disadvantage is that some patients are unable to tolerate it and access to MRI scanning can still be limited in some areas.

- Contrast enhanced CT – This will often provide exact localization of the mass and give information about its immediate anatomical relations. The contrast will allow assessment of the vascularity of the mass and its relationship to the great vessels. It also gives information about any associated bony erosion and lymphadenopathy. It may identify a primary head and neck malignancy or systemic lymphadenopathy. CT scanning is usually well tolerated and readily available. It does not give the superior soft tissue resolution of MRI scanning.

Investigations show an avidly enhancing mass at the right carotid bifurcation causing splaying of the internal and external carotid arteries. (Figures 2 and 3)

What is your provisional diagnosis?

Right carotid body tumour

What other investigations can be considered for diagnosing carotid body tumours?

- Angiography – This is useful in very vascular lesions. It allows accurate assessment of the feeding vessels and will give information on the ease of embolization should the operating surgeon feel it necessary. It nicely illustrates the splaying of the internal and external arteries classical for carotid body tumours, this is known as the ‘Lyre sign’ (Figure 5). Angiography can be combined with either CT or MRI imaging. Identification of the feeding vessels may be possible and this can assist in planning a management strategy.

- Angiography can be combined with balloon occlusion testing if resection of the carotid artery is thought to be a possibility. This will identify adequacy of the collateral circulation and may guide assessment of the risk of cerebrovascular complications.

- Urinary catecholamines – A 24 hour urine collection for urinary catecholamines will identify if a patient has a functioning paraganglioma.

- MIBG (meta-iodo-benzyl-guanidine) scanning – This may be useful if urinary catecholamines are positive i.e. there is a functioning tumour. MIBG is a radionuclide with similar properties to noradrenaline and can trace catecholamine uptake and storage. It has a tendency to localise to adrenergic tissues. Simultaneous potassium iodide administration is
What is a carotid body tumour?
A carotid body tumour is a rare vascular neoplasm arising from the paraganglia (neural crest cells) of the autonomic nervous system. It is the commonest form of paraganglioma encountered in the head and neck. The incidence of paragangliomas overall is only 1 in 30,000 to 1 in 100,000 and carotid body tumours make up 65% of these. It is named for its site of origin – the carotid body, a small aggregation of tissue situated at the carotid bifurcation responsible for the regulation of oxygenation. A normal carotid body is 3-5mm long and weighs approximately 15mg. It is particularly sensitive to a reduction in the partial pressure of oxygen. Hyperplasia of the gland can occur in medical conditions that give rise to chronic hypoxia (chronic pulmonary disease) or in those populations who live at high altitude.

Are they benign or malignant?
The majority of paragangliomas are benign however a small subset, approximately 10%, are malignant. Malignancy can be difficult to diagnose pre-operatively and is based on the presence of local metastasis to lymph nodes or disseminated disease. It is usually only diagnosed on the basis of a pathology specimen taken from somewhere distant to the primary site.

There are some features from the primary tumour that may make one more suspicious of malignancy in the absence of local or distant metastases. Lesions that are painful or show rapid enlargement and occur in younger patients should all be offered expedient treatment. Lymphovascular invasion, perineural invasion, poor circumscription and invasion through the fibrous capsule, high cellularity and widespread profound nuclear pleomorphism, increased or atypical mitotic figures and necrosis may all lead to a poorer prognosis and indicate a higher risk of metastatic disease.

Do they have any systemic effects?
Approximately 5% of tumours are functioning. This means that they secrete catecholamines and produce clinical effects such as flushing, tachycardia and sweating episodes.

What are the histopathological appearances?
Carotid body tumours, as all paragangliomas, are characterised by ‘zelballen’ which are densely packed nests of type I (chief) catecholamine producing cells. These have a rich vascular supply and tend to grow slowly in an expansile fashion. They have the capacity to infiltrate and erode bone. The type I (chief) cells are supported by network of type II (sustentacular) cells.

Are there any relevant classification systems?
A system was proposed in 1971 by Shamblin et al. This is a surgical classification system based on the tumour-vessel relationship, intraoperative findings and post-operative specimen. There are three groups: Group I are localised tumours that do not involve the surrounding major vessels. Group II are adherent or partially surround the vessels and group III are large and completely encase the vessels. Group I and II can usually be excised without vascular complications whereas group III often require resection and vascular reconstruction.

The more advanced (group III) tumours may also carry a risk of a higher complication rate particularly with respect to cranial nerve damage.
Are there any risk factors for developing carotid body tumours?

Approximately 10% of carotid body tumours are familial. If a patient is found to have bilateral tumours there is an 80% risk that there is an underlying genetic susceptibility.

Paragangliomas can occur as part of tumour syndromes such as MEN II or due to hereditary paraganglioma genes. These genes code for subunits B, C and D of succinate dehydrogenase (SDHD), a mitochondrial enzyme important in the respiratory cycle. Mutations in SDHD subunit D and B predispose to familial pheochromocytoma paraganglioma syndrome, however subunit D carriers are more likely to develop head and neck paragangliomas and subunit B carriers are more likely to develop pheochromocytomas. Both confer an added risk of malignancy. In contrast subunit C mutations are usually isolated and is seldom associated with malignancy. These patients tend to develop isolated head and neck paragangliomas.

All SDHD genes are inherited in an autosomal dominant manner and a carrier of the gene can expect to develop a paraganglioma by the time they are 30 years old 50% of the time, this increases to 80% at the age of 50 years. One interesting point about the inheritance is that the patient will only develop paragangliomas if they inherited the gene from their fathers – ‘parent of origin’ inheritance. This leads to silent carriers within families and an often difficult to trace pattern of inheritance.

Molecular screening for mutations in genes coding for subunits B, C and D is recommended both in cases of familial and sporadic head and neck paraganglioma to assist in management decisions. Testing may be extended to the immediate family if a mutation is found. Up to 79% of carotid body tumours may be positive for a mutation in subunit D, this necessitates thorough investigation at the point of diagnosis for potential synchronous paragangliomas.

Patients who have familial paraganglioma syndrome should be screened with annual urinary catecholamines and abdominal ultrasound. They should also receive three yearly MRI scans of the head, neck, thorax and abdomen.

How are carotid body tumours managed?

Surgery has been the mainstay of treatment for carotid body tumours for many years. More recently radiotherapy has become an important alternative treatment option either on its own or as an adjunct to surgery.

Surgical resection is advocated for definitive diagnosis and to avoid progression of symptoms. As stated previously, it is almost impossible to ascertain whether the tumour is malignant preoperatively making it difficult to advocate a “wait and see” approach unless the patient is unable to undergo an anaesthetic. Pre-operative morbidity can be predicted to some extent using the Shamblin classification – the larger the tumour the more likely the chance of post-operative cranial nerve palsies or cerebrovascular complications. This is another reason for early surgical intervention.

Embolisation prior to surgical intervention is often used routinely in the immediate pre-operative management of carotid body tumours. Its proponents argue that it reduces the vascularity of the tumour making blood loss less of a problem during the procedure which can consequently lead to fewer technical difficulties. Others feel that it makes little difference to blood loss, operating time and that it can increase the neurological complication rate. Carotid body tumours also tend to have multiple feeders from the carotid bulb not amenable to embolisation.

Most tumours can be operated on through a cervical approach, occasionally a cervico-parotid approach. Other approaches, for example via mandibulotomy, are unusual in these tumours. Meticulous haemostasis is required. Risks to cranial nerves (especially X and XII) increases with size of tumour as does the risk of resection of the affected carotid artery. Internal carotid artery sacrifice through necessity or through a tear is always a possibility and this surgery should be done with a vascular surgeon at least on stand-by. If sacrificed or injured, a bypass catheter (or shunt) should be used immediately, allowing for the completion of the resection. Following this the artery can be grafted using either vein or a prosthetic material.

What about radiotherapy?

Post-operative radiotherapy has been used for the treatment of malignant paragangliomas or residual disease after surgery in routine practice for many years. As a primary treatment in head and neck paragangliomas in general, it has a high rate (around 90%) of tumour ‘control’, i.e. no progression and some tumour shrinkage. Comparatively small doses are used and the morbidity is low, especially if intensity-modulated radiotherapy (IMRT) is used. Those who advocate its use suggest that the absence of progression is equivalent to surgical cure.

Tumour control with both surgical and non-surgical treatment modalities appear to be equal and tumour size does not appear to a significant factor in response to treatment at 45Gy. Complications do not increase with size as they do in the surgical group. A disadvantage of primary radiotherapy is that no tissue diagnosis is confirmed leaving a question about whether a tumour is benign or malignant unanswered.

In general, radiotherapy is a worthwhile option for very big tumours in which surgery may convey an unacceptable high morbidity; for unfit patients and for contralateral tumours in patients who have already had one side operated on. However, it is used much less commonly for carotid body tumours than for glomus vagale tumours.

Radiotherapy is also appropriate for recurrent tumours.

Summary

Carotid body tumours are an uncommon cause of a lateral neck mass. They typically present with a painless neck swelling which may or may not be associated with systemic upset. MRI scanning will show typical features indicating the diagnosis. Investigations to ascertain whether this is an isolated paraganglioma or part of a syndrome must be carried out prior to surgery. Appropriate imaging will guide surgical resection and allow assessment of potential operative complications. Radiotherapy can be appropriate for both residual and recurrent disease, as well as for local control in patients in whom surgery may not be a viable, or the best, option.

Declaration of competing interests: Nothing to declare.
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References


