Phaeochromocytoma

Rare catecholamine secreting tumour derived from chromaffin cells usually in adrenal medulla.

They may secrete constantly or intermittently. The familial type tend to produce mostly noradrenaline but the sporadic type produce mostly adrenaline. Dopamine may also be produced.

Epidemiology

- Old rule of 10s: 10% familial, 10% bilateral adrenal, ~10% paraganglionic, 10% malignant.
- Newer data suggests: 20% familial, 18% paraganglionic, and ~15% are malignant
- ~10% occur in children (50% solitary adrenal tumours, 25% bilat and 25% paraganglionic)
- M=F, No particular racial predisposition.

Inherited Forms

- Multiple endocrine neoplasia syndrome (MEN II) – 50% affected with 70% bilateral
- Neurofibromatosis type 1 - 1% affected
- von Hippel-Lindau syndrome - 15% affected(VHL)
- Familial paragangliomas

History

Symptoms are intermittent.
Classically: Headache, profuse sweating, palpitations, tremor
Also: Nausea, weakness, anxiety, sense of doom, epigastric pain, flank pain, constipation, wt loss.

Examination

Classically: ↑BP (often paroxysmal), postural hypotension, tremor.
Also: Hypertensive retinopathy, pallor, fever, reflex bradycardia (from NA secretion→↑BP).
Neurofibromas may be felt and café au lait patches may be seen.

Investigations

24hr urine - total catecholamines, vanillylmandelic acid (VMA) and metanephrines.
Bloods: Plasma free metanephrines, TFTs, BSL
Imaging: CT/MRI or PET. Also metaiodobenzylguanidine (MIBG) scan labelled with ^{131}I or ^{123}I

Differential Diagnosis

Include: Anxiety disorder, carcinoid tumour, EtOH withdrawal, labile hypertension, drug abuse
Differentiate familial causes:

- Bilateral tumours suggest MEN.
- There may be features of neurofibromatosis including café au lait spots.
- The von Hippel-Lindau (VHL) syndrome is associated with phaeochromocytoma, cerebellar haemangioblastoma, and renal cell carcinoma.

Management

Hypertensive crisis:

- **Phentolamine** 2-5mg IV or **nitroprusside** infusion may be required
- Alpha blockade with **phenoxybenzamine** PO once controlled.
- Once this block achieved, then can consider beta blockade (otherwise unopposed alpha stimulation can precipitate a hypertensive crisis) to control reflex tachycardia.
- **CCB** also useful.

Hypotension:

- Catecholamines
- Intra-aortic balloon pump.

Definitive surgery – curative in 90%

Palliative radiotherapy and chemotherapy in the rare malignant cases

Genetic counselling if familial cause.

Complications

Any of the following may precipitate a hypertensive crisis:

- Induction of anaesthesia
- Opiates
- Dopamine antagonists
- Decongestants such as pseudoephedrine
- Drugs that inhibit the reuptake of catecholamines, including TCAs and cocaine
- X-ray contrast media
- Childbirth

Prognosis

- 5-year survival rate for non-malignant phaeochromocytoma >95%
- 5-year survival rate for malignant phaeochromocytoma <50%.
- Risk of malignancy higher in children.

Phaeochromocytoma and Pregnancy

Pre-eclampsia is fairly common whilst phaeochromocytoma is very rare and only a few hundred cases in pregnancy have been reported in the literature.

- If diagnosed in pregnancy → high maternal & fetal mortality.
- If diagnosis precedes pregnancy, the outcome is vastly better.
- If surgery performed in the 1st or 2nd trimester TOP not needed but miscarriage is high.
- In 3rd trimester as soon as fetal lung maturity is confirmed, perform LSCS followed by surgical removal of the tumour.
- Conservative management during pregnancy has been described.