Delayed diagnosis of neurofibromatosis type 1 associated phaeochromocytoma and intussuscepting sigmoid adenocarcinoma

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Case study - History

- Male, 67 year old part-retired farmer
- Initial presentation to surgeon with altered bowel habits and bleeding per rectum – colonoscopy and abdominal imaging revealed an obstructing intussuscepting sigmoid colonic adenocarcinoma (confirmed histology)
- A subsequent referral to endocrinologist due to an incidental finding of a heterogeneously enhancing 5cm right adrenal mass on imaging
- On detailed history: 5 years history of hypertension and episodic classical symptoms – light headedness, blurred vision, feeling of impending collapse with pounding chest on straining or sheering sheep (which was previously investigated and diagnosed with vasovagal episodes)
- Other past medical history: Diet controlled diabetes, benign prostatic hypertrophy
- Drug history: Felodipine MR, indapamide, simvastatin, tolterodine
- No family history of MEN

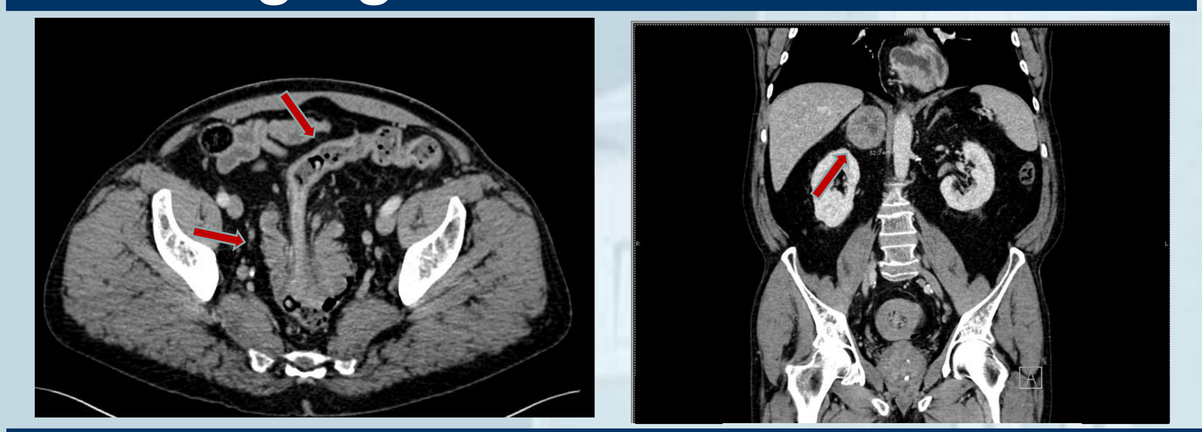
Examination Findings

- Pulse 78 and regular
- BP 140/85 mmHg
- Height 179 cm, Weight 88.4 kg, BMI 27.6
- Multiple skin nodules presumed neurofibromatoma and axillary freckling
- No café au lait spots

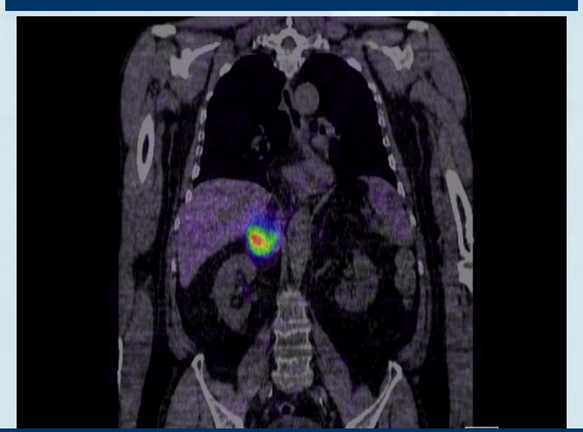
Investigations

	Results	Reference
24h Urine Normetadrenaline	4	0 – 3 umol/24h
24h Urine Metadrenaline	50	0 – 1.8 umol/24h
Plasma Normetanephrine	2,225	120 – 1180 pmol/L
Plasma Metanephrine	14,448	80 – 510 pmol/L
CT	5cm heterogeneously enhancing right adrenal mass; 4cm obstructing intussuscepting sigmoid tumour	
MIBG	Intense tracer accumulation in right adrenal mass only	

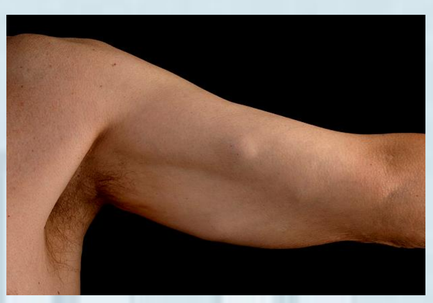
CT imaging

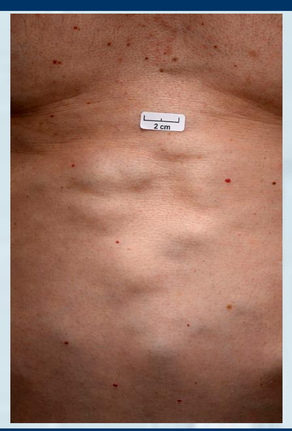


MIBG scan



Skin nodules





Case resolution

- Our patient's diagnosis of phaeochromocytoma was missed for 5 years until he had investigations for bowel symptoms despite having typical episodic symptoms, hypertension and neurofibromatoma.
- The diagnosis was confirmed biochemically and alpha blockade was immediately started with rapid dose titration, low fibre diet for pre-operative care and a plan for surgery. The care was optimised in collaboration with colorectal, adrenal surgical team and anaesthetist.
- At 3 weeks presentation, he underwent successful laparoscopic right adrenalectomy and high anterior resection, formation loop ileostomy and rectal washout followed by closure of loop ileostomy at 4 weeks.
- This case illustrates the challenges of timing safety of anaesthesia (risk of phaeochromocytoma crisis) and potential untoward complication of delaying surgery for his colonic obstruction.
- He is currently awaiting further genetic testing for probable neurofibromatosis type 1 (NF1) and a potential link to gastrointestinal cancer.

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Adrenal and steroids

Mie Tisdale

Poster presented at: