Audit of Patients with Multiple Endocrine Neoplasia Type 1 in a Tertiary Referral Centre

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INTRODUCTION

Multiple endocrine neoplasia type 1 (MEN1) is characterised by parathyroid, pituitary and pancreatic tumours in association with neoplasia of intra-thoracic endocrine tissue, adrenal glands and cutaneous manifestations. We reviewed patients in our centre attending a dedicated MEN1 clinic, where detailed radiological and biochemical surveillance is undertaken (Thakker et al. 2012).

METHODS

Case notes and electronic records of patients attending a tertiary centre clinic for care of MEN1 were reviewed.

DEMOGRAPHICS

48 patients reviewed
• 100% Caucasian
• Mean age 49 years (range 14 – 89 years)
• 54% female, 46% male
• 3 deceased
  • 1 metastatic gastrinoma
  • 1 chest NET
  • 1 breast cancer
• Mean age of death 56 years (range 45 – 75)
90% had confirmed MEN1 mutations
• 77% from families with more than one affected patient
• 38% detected through screening of family members
• 1 patient had novel MEN1 mutation Q547X

Prevalence in this catchment area is 1/2000
In this cohort there were
• 42 primary hyperparathyroidism
• 32 pancreaticoduodenal NETs
• 13 pituitary tumours
• 10 adrenal masses
  • 3 bilateral
  • 1 functioning (resected)
  • 2 thymic masses
  • 1 resected
  • 1 awaiting resection
  • 1 multiple gastric carcinoid on SSA/surveillance
• 2 chest NETs surveillance
• 1 DIPNECH surveillance

PANCREATICODUodenAL NETs

Mean size of tumour was 1.5cm
Insulinomas (n=4)
• Mean age of diagnosis 25 years (16 – 34 years)
• Glucose mean level 2.4mmol/l (range 1.7 – 3.5)
• Insulin mean level 57pmol/l (range 32 – 101)
• Proinsulin mean level 18pmol/l (range 15 – 20)
• 100% underwent surgery, with 100% currently in remission
Gastrinomas (n=16)
• Mean age of diagnosis 46 years (19 – 72 years)
• Gastrin mean level 339pmol/l (range 9 – 2483)
• 5 patients underwent surgery due to enlargement or worsening symptoms (2 Whipple’s procedures, 2 total pancreatectomies, 1 distal pancreatectomy)
• 3 patients who underwent surgery were found to have positive lymph node metastases on histology
• PPs were given to 73% of patients with gastrinomas
Glucagonoma and non-functioning PNETs have been managed with surveillance only
Somatostatin analogues given to 4 patients

PITUITARY

27% developed pituitary adenomas
• Mean age at diagnosis 48 years (23 – 60 years)

The characteristics of MEN1 patients presenting to our service is consistent with previous reports (Pieterman et al. 2011).

Five patients have undergone pancreatic surgery, one of whom died 10 years after surgery. Two patients had previously unidentified lymph nodes discovered at surgery – it remains uncertain whether these patients will remain disease free. Optimal management of such patients remains unclear. NETs in MEN1 may often behave in an indolent manner and conservative management is often appropriate. Systemic therapies with conventional chemotherapy and newer biological agents are available but efficacy in this context is yet to be seen.

The wide-ranging manifestations of MEN1 emphasise the need for specialist review with a multidisciplinary team approach to achieve optimum outcomes.

REFERENCES