Audit of Patients with Multiple Endocrine Neoplasia Type 1 (MEN1): Screening of Pancreatic Neuroendocrine Tumours (pNETs), Parathyroid Tumours and Pituitary Adenomas

MP Kyithar¹, L Cullen², CS Lee², N Swan², J Geoghegan², S Skehan², R Crowley¹,², D O'Shea¹,², D O'Toole²

¹Endocrinology Department, St. Vincent’s University Hospital, Dublin, Ireland
²Neuroendocrinology Multidisciplinary Team, St. Vincent’s University Hospital, Dublin, Ireland

INTRODUCTION

Patients with MEN1 have increased morbidity and mortality compared to those patients with sporadic NETs. No genotype-phenotype correlation is described and age-related clinical penetrance surpasses 50% and 90% by 20 and 40 years, respectively. The aim of the audit was to compare the screening programme for MEN1 patients with MEN1 clinical guidelines.

METHODS

Case notes of MEN1 patients attending a tertiary NET-multidisciplinary team (MDT) in Ireland were reviewed. All patients attending the NET-MDT have gastrointestinal hormone, parathyroid, pituitary profiles, endoscopic and imaging studies according to guidelines.

RESULTS

Patients with MEN1

- 13 patients (11 kindreds), 100% Caucasians, mean age 43 years (range 28-67): 69% male, 31% female
- 85% (n=11) had confirmed MEN1 mutations and two had clinical or familial MEN1

Reasons for Patients referred to NET-MDT

- Screening for known MEN1 mutations
- Family screening
- Management of pNETs
- Screening due to PHPT & acromegaly

Prior to NET-MDT assessment,

- 46% did not have endoscopic/radiology studies to screen for pNETs
- None of the patients had CT/MRI thorax to screen for thymic/bronchial NETs

Age at screening

- Age referred to NET-MDT: 41.5±12.2 years.
- Age at endoscopic/radiological screening for NETs (prior to/at NET-MDT): 37.1±14.3 years
- Age at diagnosis of NETs: 34.9±14.3 years

On screening of NET-MDT, the following new diagnoses were made:

- pNET: 8
- Duodenal/gastric NET: 7
- PHPT: 2
- Pituitary adenomas: 3
- Adrenal adenomas: 4

CONCLUSIONS

Endoscopic/radiological screening of NETs occurred at later age than recommended by current guidelines. Surveillance methods were also largely at variance with guidelines. Referral to a dedicated MDT has identified a significant number of previously unrecognised neuroendocrine pathologies.

REFERENCES

Guidelines for diagnosis and therapy of MEN type 1 and type 2. Broado et al, J Clin Endocrinol Metab. 2001;86(12):5658-71
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