THREE GENERATIONS OF MEN-1: THE IMPORTANCE OF FAMILY SCREENING

M. DUARTE, A. LEMOS, J. GALHARDO, L. LOPES
1. Unit of Paediatric Endocrinology, Hospital de Dona Estefânia – Central Lisbon University and Hospital Centre, Lisbon, Portugal

INTRODUCTION

Multiple endocrine neoplasia type 1 (MEN-1) is a rare and underdiagnosed syndrome caused by inactivating mutations of the tumor suppressor gene MEN-1, predisposing to multiple (endocrine and nonendocrine) tumors, classically situated in anterior Pituitary, Parathyroid and Pancreas. Being that the mutation is transmitted in an autosomal dominant way, the screening of all first-degree relatives is mandatory when an index case of MEN-1 is identified.

CASE REPORT

- Neuroendocrine pancreatic tumor + Parathyroid hyperplasia
  - 75Y - 2 Ps – clinical suspicion → genetic testing
- Asymptomatic - same mutation confirmed
  - 35Y - Screening → Neuroendocrine pancreatic tumor + Parathyroid hyperplasia
- Asymptomatic - same mutation confirmed
  - 16Y - Screening → Neuroendocrine pancreatic tumor + Parathyroid hyperplasia
- Asymptomatic - same mutation confirmed
  - 12Y - Screening → Negative

GERMINAL MUTATION IN EXON 8 OF MEN1 GENE
(c1087G>T(p.Glu363STOP))

DISCUSSION

Identifying MEN-1 gene mutation in the grandfather allowed its detection in 3 young and asymptomatic relatives, making possible periodic screening and timely therapeutic intervention, reducing the morbidity and mortality associated to this syndrome.

MANAGEMENT:

Genetic test
ALL MEN1 patient first grade relatives
Clinical, analytical and radiological screening
EVERYTIME MEN1 mutation is identified

CONTACT INFORMATION

Júlia Galhardo
Unit of Paediatric Endocrinology - Hospital Dona Estefânia – CHULC, Lisbon, Portugal
juliagalhardo@chlc.min-saude.pt

REFERENCES