

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

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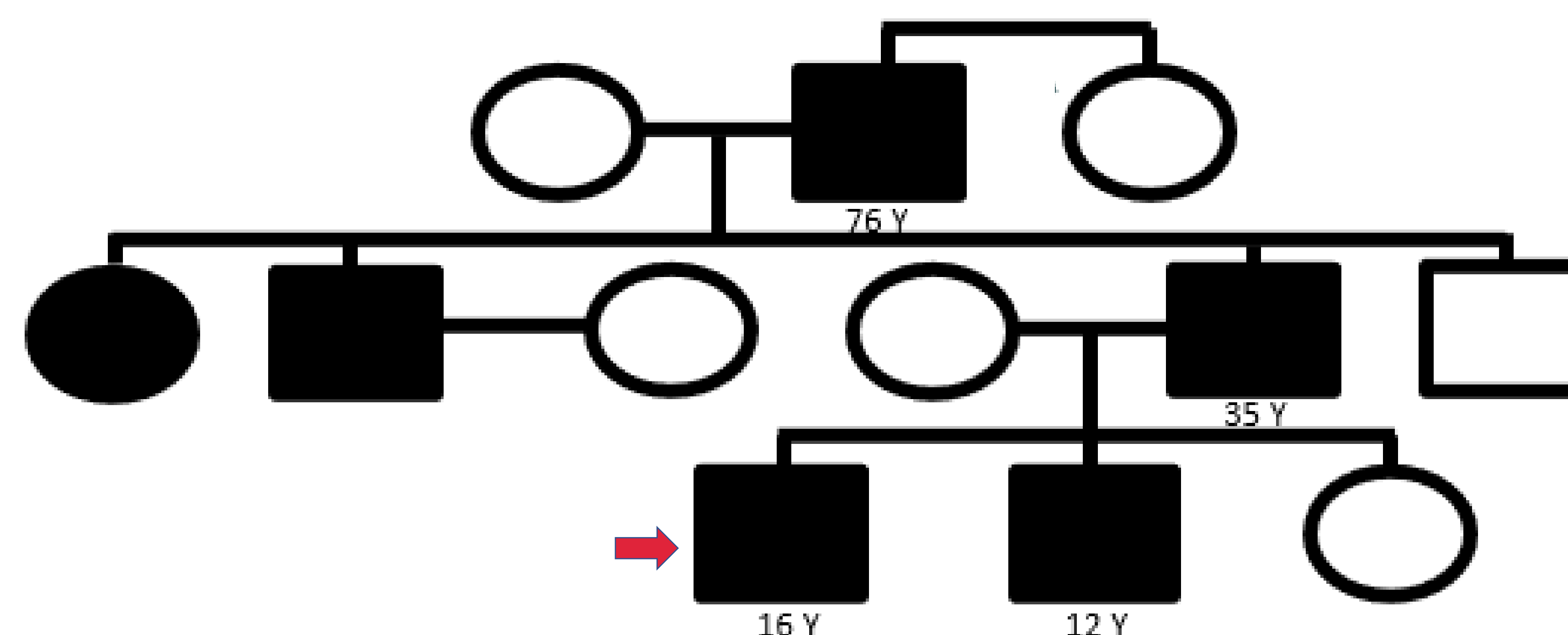
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INTRODUCTION

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



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

Index case

♂ 75Y - **Neuroendocrine pancreatic tumor + Parathyroid hyperplasia**
- **2 Ps** – clinical suspicion → genetic testing

Germinal mutation in exon 8 of MEN1 gene
(c1087G>T (p.Glu363STOP))

2nd Generation

♂ 35Y - Asymptomatic - same mutation confirmed
- Screening → **Neuroendocrine pancreatic tumor + Parathyroid hyperplasia**

3rd Generation

♂ 16Y - Asymptomatic - same mutation confirmed
- Screening → **Neuroendocrine pancreatic tumor + Parathyroid hyperplasia**



♂ 12Y - Asymptomatic - same mutation confirmed
- Screening → **Negative**

REFERENCES

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