

Backman S, et al.

Supplementary materials – patient descriptions

1 Male

The patient did not have a known family history of endocrine tumors. At age 62 he was diagnosed with acromegaly. Subsequently, at age 65 he was diagnosed with hyperparathyroidism (no symptoms) which was treated surgically. The pathology report showed hyperplasia. The eventual cause of death at age 84 was heart failure.

MEN1 criteria: Pituitary adenoma and HPT.

2 Female

The patient did not have a known family history of endocrine tumors. At age 67 a prolactinoma (causing vision loss) was diagnosed. At age 68, hyperparathyroidism (causing fatigue) was diagnosed and treated surgically. The pathology report showed hyperplasia and normal parathyroid tissue. The eventual cause of death at age 83 was kidney failure.

MEN1 criteria: Pituitary adenoma and HPT.

3 Male

The patient did not have a family history of endocrine tumors. At age 62 an insulinoma, causing hypoglycemia was diagnosed and removed. Also at age 62, asymptomatic hyperparathyroidism due to parathyroid hyperplasia was diagnosed.

MEN1 criteria: Insulinoma and HPT.

4 Male

The patient did not have a family history of endocrine tumors. At age 69 a prolactinoma causing a vision field defect, as well as asymptomatic hyperparathyroidism due to an adenoma were diagnosed. Additionally, the patient has been diagnosed with malignant melanoma, and an enlargement of the right adrenal gland has been noted.

MEN1 criteria: Pituitary adenoma and HPT.

5 Female

The patient did not have a family history of endocrine tumors. At age 39, hyperparathyroidism causing fatigue, depression and nephrolithiasis was diagnosed and treated. The pathology report showed a parathyroid adenoma. At age 55, a non-functioning pancreatic neuroendocrine tumor was diagnosed and treated. The pathology report additionally showed islet cell hyperplasia. The patient has also been diagnosed with a hypernephroma, and eventually passed at age 80 due to non-MEN1 related causes.

MEN1 criteria: HPT and PNET

6 Female

The patient's father and daughter have been diagnosed with hyperparathyroidism. At age 45 the patient was diagnosed with hyperparathyroidism with symptoms of depression. The pathology report showed a parathyroid adenoma. At age 58 a non-functioning neuroendocrine liver metastasis was found. The lesion stained positive for pancreatic

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polypeptide and chromogranin A. Pancreatic origin was presumed but the primary lesion was never detected. The eventual cause of death at age 73 was ductal adenocarcinoma of the breast.

MEN1 criteria: HPT and suspected pancreatic NET

7 Male

The patient did not have a family history of endocrine tumors, although the mother passed at a young age due to breast cancer. At age 35 the patient was diagnosed with hyperparathyroidism causing depression and nephrolithiasis. The patient was treated surgically and the pathology report showed hyperplasia. At age 52 the patient was diagnosed with multiple gastrinomas causing gastritis and diarrhea. The adrenal glands were found to be bilaterally enlarged. The eventual cause of death at age 63 was pancreatic NET.

MEN1 criteria: HPT and multiple gastrinomas.

8 Female

The patient did not have a family history of endocrine tumors. At age 52 the patient was diagnosed with hyperparathyroidism causing fatigue and a non-functioning pituitary adenoma without symptoms. The pathology report showed parathyroid hyperplasia. At age 53 two non-functioning pancreatic neuroendocrine tumors were found. The eventual cause of death at age 72 was uncertain.

MEN1 criteria: Pituitary adenoma, Pancreatic NET and HPT.

9 Female

The patient did not have any certain history of endocrine tumors. At age 34 she was diagnosed with a prolactinoma causing amenorrhea and galactorrhea. At age 42 she was diagnosed with and treated for asymptomatic hyperparathyroidism. The pathology report showed parathyroid hyperplasia.

MEN1 criteria: Pituitary adenoma and HPT.

10 Male

The patient did not have a family history of endocrine tumors. At age 54 he was diagnosed with hyperparathyroidism causing nephrolithiasis. He was treated surgically and the pathology report showed hyperplasia and adenoma. Also at age 54, he was diagnosed with an asymptomatic prolactinoma. An adenoma has been noted on the left adrenal gland.

MEN1 criteria: Pituitary adenoma and HPT.

11 Female

The patient did not have a certain family history of endocrine tumors. At age 43 the patient was diagnosed with hyperparathyroidism causing nephrolithiasis. The pathology report showed an adenoma. At age 44 the patient was diagnosed with an insulinoma causing hypoglycemia. The pathology report showed islet cell hyperplasia.

MEN1 criteria: Insulinoma and HPT.

12 Female

The patient had a sister with metastatic pancreatic neuroendocrine tumors. At age 41 the patient was diagnosed with hyperparathyroidism causing depression. The pathology report was inconclusive but showed macroscopically enlarged parathyroid glands. At age 50 the patient was diagnosed with a GH-producing prolactinoma presenting with headaches and galactorrhea.

MEN1 criteria: Pituitary adenoma and HPT.

13 Male

The patient did not have a family history of endocrine tumors. At age 25 he was diagnosed with hyperparathyroidism causing nephrolithiasis and insulinoma causing hypoglycemia. The pathology report showed hyperplasia and adenoma. The patient has developed multiple pancreatic NETs (insulinoma and gastrinoma) and an adrenal adenoma has been noted.

MEN1 criteria: Multiple pancreatic NETs and HPT.

14 Male

The patient's brother has been diagnosed with parathyroid cancer. At age 24 the patient was diagnosed with hyperparathyroidism causing nephrolithiasis. The pathology report showed parathyroid hyperplasia.

MEN1 criteria: Not met. The initial suspicion of MEN1 arose due to early onset hyperparathyroidism. The diagnosis of parathyroid cancer in a relative makes the HPT-JT syndrome (eventually confirmed) a more likely diagnosis.