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Multiple angiofibromas and collagenomas in a 45-year-old man with recurrent nephrolithiasis, fatigue, and vision loss

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CASE SUMMARY

History

A 45-year-old man was referred to the Dermatology Branch Consultation Service at the National Institutes of Health for evaluation of a 20-year history of multiple asymptomatic papules on the face and papulonodules on the trunk. The truncal lesions had been increasing in number and size over the past 5 years. Two lipomas located on the left anterior thigh and the right posterior thigh had been excised in his late teens.

Eighteen months before presentation, the patient was evaluated for recurrent nephrolithiasis, constipation, and fatigue. He was found to be hypercalcemic secondary to primary hyperparathyroidism and underwent right-sided parathyroidectomy without complete normalization of his serum calcium values. Six months later, he developed bilateral visual field deficits. A 5-cm pituitary mass was identified by magnetic resonance imaging, and his serum prolactin level was elevated to 5380 $\mu\text{g/L}$ (reference range, 3–29 $\mu\text{g/L}$). A diagnosis of macroprolactinoma was made and medical therapy with cabergoline was

Abbreviations used:

MEN1: multiple endocrine neoplasia
MTC: medullary thyroid carcinoma
TSC: tuberous sclerosis complex

instituted, with complete recovery of his visual fields. Computed tomography of the abdomen demonstrated bilateral adrenal masses, and the patient was referred to the National Institutes of Health (NIH) for further evaluation and treatment. Upon presentation, he complained of chronic headaches associated with nausea and vomiting without visual changes, myalgias, or bone pain. The patient also had a 20-year history of severe gastroesophageal reflux. He denied diarrhea and galactorrhea. There was no family history of endocrine disorder.

Physical examination

On examination at the NIH, the patient weighed 144.2 kg and was 151 cm tall (body mass index, 63.2). Deep longitudinal furrows consistent with cutis verticis gyratum were noted on the parietal and occipital scalp. There was a prominent posterior neck fat pad. On the neck and axillae were approximately 50 soft, pedunculated papules, 3 to 10 mm, suggestive of acrochordons (Fig 1, A). Larger lesions had mammillated surfaces. The skin in the neck and axillae was hyperpigmented with velvety hyperkeratosis, consistent with acanthosis nigricans (Fig 1, A). Approximately 10 flat, skin-colored to pink papules, 1 to 2 mm in size, several with visible telangiectasia, were noted on the tip of the nose, the nares, and close to the vermilion border of the lips (Fig 1, B). Several small (<2 mm) erythematous papules were present on the attached gingiva over the maxillary incisors. The anterior abdominal wall was studded with more than 100 discrete, oval and round, soft to

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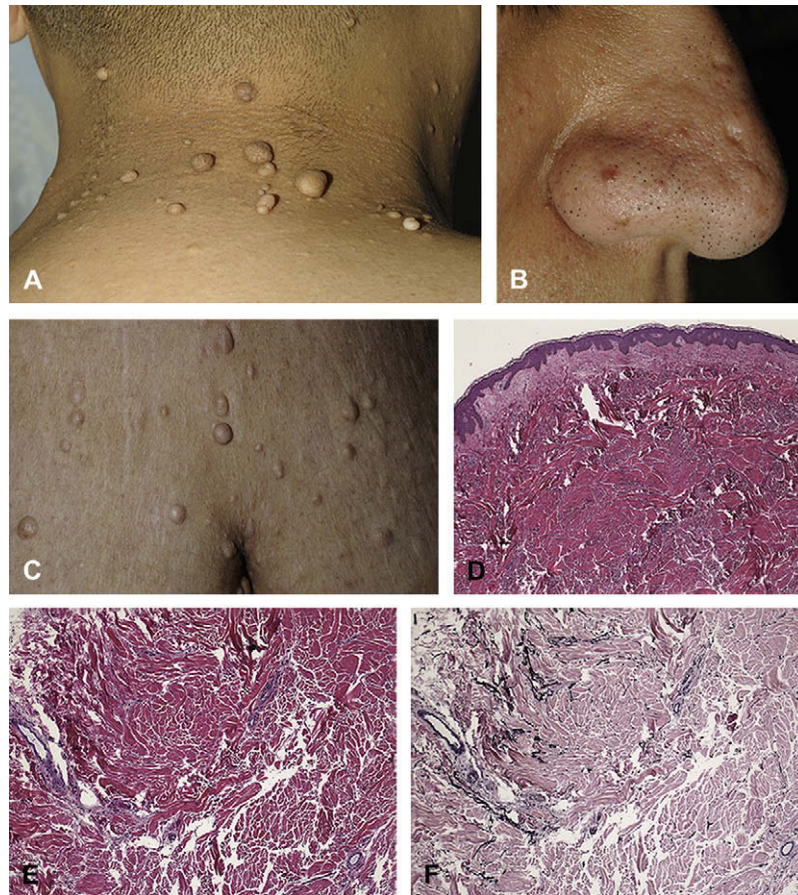


Fig 1. **A**, Numerous pedunculated papules and velvety hyperpigmented hyperkeratosis on posterior neck. **B**, Multiple telangiectatic papules on the nose. **C**, Periumbilical skin-colored papulonodules. **D** and **E**, Skin biopsy specimen from the abdomen demonstrates widened collagen bundles and loss of adnexae. (Hematoxylin-eosin stain; original magnifications: **D**, $\times 2.5$; **E**, $\times 5$.) **F**, Verhoeff–van Gieson elastin stain of similar field shown in **E** demonstrates normal elastin staining at periphery of the papule (left side) and decreased elastic fibers at the site of the collagenoma. (Original magnification: $\times 5$.)

firm, nontender, skin-colored papules ranging from 0.1 to 1 cm in diameter (Fig 1, C). There were two soft subcutaneous nodules on the ventral aspect of both upper extremities, suggestive of lipomas. Linear atrophic plaques consistent with striae were present on the lower abdomen and upper thighs.

Histopathology

Histopathologic examination of 3 anterior abdominal papules demonstrated large areas of coarse dermal collagen without adnexal structures (Fig 1, D and E). Verhoeff-van Gieson elastin stains of these regions revealed markedly decreased elastic fibers compared with adjacent dermis, consistent with collagenomas (Fig 1, F). A biopsy specimen of a polypoid neck lesion demonstrated lobules of mature adipose tissue admixed with abundant fibrous tissue typical of fibrolipoma.

Other significant diagnostic studies

Laboratory investigations were significant for the following: parathyroid hormone, 128 pg/mL (reference range, 16-87); venous ionized calcium, 1.56 mmol/L (1.12-1.32); prolactin, 705 $\mu\text{g/L}$ (3-29); 24-hour urine calcium excretion, 17.8 mmol/24h (1.25-7.50). A serum gastrin level of 98 pg/mL was at the upper limits of normal (<100). Other normal laboratory study values included plasma fractionated normetanephrine, 51 pg/mL (18-112); plasma fractionated metanephrine, 23 pg/mL (12-61); adrenocorticotropic hormone, 24.8 pg/mL (0.0-46.0); serial serum cortisol levels (30 minutes apart) 7.5 $\mu\text{g/dL}$ and 5.3 $\mu\text{g/dL}$ (5.0-25); thyroid-stimulating hormone, 1.00 $\mu\text{IU/mL}$ (0.40-4.00); thyroxine, 7.9 $\mu\text{g/dL}$ (4.5-12.5), free thyroxine, 0.9 ng/dL (0.8-1.9); and triiodothyronine, 180 ng/dL (90-215).

Indium In 111 octreotide scintigraphy demonstrated increased uptake in the pituitary region and

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