Conclusion

The mass also compressed the interazygos esophageal system, the SVC, and the left brachiocephalic vein (Figure 1B). Thoracic computed tomography confirmed the presence of an expansive solid mass of the neck area and a well-defined net of superficial calcifications and renal failure.1,3,6

Interestingly, though our patient has the same compound heterozygous mutation as was found in 2 other cases,2,3 their clinical presentations are not identical. Indeed, neither LMNA or ZMPSTE24 mutations were found in 3 other patients with MAD type B,1 suggesting that undiscovered mutations may modulate the clinical phenotype.

Dermatologists may be consulted to evaluate dyspigmentation but can be instrumental in the diagnosis of MAD type B by recognizing its earlier multisystem constellation of symptoms. Earlier diagnosis would alert physicians of the particular need, among others, for early renal monitoring. However, disease heterogeneity suggests that management should be tailored to phenotypic severity.

Edema and Telangiectasia of the Chest Caused by Neuroendocrine Carcinoma

Superior vena cava (SVC) syndrome includes a constellation of signs and symptoms resulting from SVC obstruction that produces elevated pressure in the afferent veins and increased blood flow through collateral vessels.

Report of a Case | A man in his 60s was referred for a subcutaneous nodule that had appeared in the right supraclavicular region about 1 year earlier. The patient, who had a 5-year history of chronic obstructive pulmonary disease, complained of hoarseness, cough, and tachycardia. At physical examination, the nodule, measuring about 4 cm in diameter, was round and fixed with irregular ill-defined margins. Edema and erythema of the neck area and a well-defined net of superficial dilated vessels in the thoracic and epigastric areas were also observed (Figure 1A).

Blood tests showed elevated levels of lactate dehydrogenase (1180 U/L; normal range, 250-500 U/L) and slightly raised levels of carcinoantigen (CA) 15-3 (29 U/mL; normal range, 0-25 U/mL). Levels of CA 19-9, carcinoembryonic antigen, and α-fetoprotein were within the normal range. Otolaryngology consultation revealed paralysis of the right vocal cord. Chest radiography showed an opaque thoracic mass approximately 12 cm in diameter located on the right apical side that caused left tracheal deviation. Thoracic computed tomography confirmed the presence of an expansive solid mass in the mediastinum infiltrating the right side of the trachea, the SVC, and the left brachiocephalic vein (Figure 1B).

The mass also compressed the interzygous esophageal system, with vascular shunts detectable in the right paramediastinal side.

A punch biopsy of the nodule revealed a large-cell neuroendocrine carcinoma showing a proliferation of atypical medium and large undifferentiated cells and areas of necrosis and hemorrhage (Figure 2A). Immunohistochemical staining was positive for the neuroendocrine marker synaptophysin (Figure 2B). The diagnosis of mediastinal neuroendocrine carcinoma of unknown origin associated with SVC syndrome prompted referral for complete surgical excision. The patient died a few weeks later after multiple postsurgical complications.

Discussion | Historical causes of SVC syndrome are usually reported as infective, such as tuberculosis or syphilis. Cur-
Currently, in 60% to 90% of cases, the cause is determined to be a malignant condition (~85%), whereas causative benign conditions are less commonly reported and include mediastinal fibrosis, retrosternal goiter, lipomatous hypertrophy, and transvenous cardiac pacemakers.1,2

Superior vena cava syndrome is related to extrinsic compression or intraluminal stenosis of the SVC. Consequent collateral circulation depends on the level of the obstruction that may be located (1) just at, (2) just above, or (3) just below the outlet of the azygos vein. In the first case, as in our case, dilated superficial collateral vessels are evident in the skin, involving internal mammary veins, epigastric veins, and intercostal veins. Signs and symptoms of SVC syndrome include facial or neck plethora, arm swelling, dilated chest veins, chest pain, dyspnea, light-headedness, dizziness, or syncope; presence of orthopnea and facial plethora may be exacerbated when the patient is supine.3,4

Figure 1. Superior Vena Cava Syndrome

A. Clinical view of edema and erythema of the neck and presence of a well-defined net of dilated vessels in the thorax and the epigastric region. B. Computed tomography of the chest showing an expansive solid mass involving the mediastinum and infiltrating the right wall of the trachea, the superior vena cava and the left brachiocephalic vein; mass dimensions indicated by green lines.

Figure 2. Neuroendocrine Carcinoma Biopsy Specimens

A. Histopathology specimen showing a proliferation of atypical and undifferentiated cells of medium and large size and areas of necrosis and hemorrhage (hematoxylin-eosin, original magnification ×100). B. Under synaptophysin immunohistochemical staining, specimen tested positive (original magnification ×200).
Mediastinal neuroendocrine carcinoma is one of the possible, although unusual, reported causes of SVC syndrome, with most patients showing a progressively worsening facial edema. A case of SVC syndrome due to mediastinal carcinoid tumor has been previously reported in a 36-year-old man who presented with facial swelling, mild dyspnea, pain in the right arm, slurring of the voice, and deviation of the tongue to the left side. However, development of cutaneous varicosities with multiple dilated venules on the anterior thoracic wall, as in this present report, has not been previously described to our knowledge in patients with mediastinal neuroendocrine carcinoma.

Recognition of SVC syndrome may be crucial for revealing a possible underlying malignant condition. In this regard, the dermatologist may play a decisive role because cutaneous manifestations may suggest this diagnosis when systemic symptoms are still lacking or negligible.

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Conflict of Interest Disclosures: None reported.


Massive Subcutaneous Masses on the Back Related to β2-Microglobulin Amyloidosis

Patients undergoing dialysis are unable to properly eliminate β2-microglobulin, a component of major histocompatibility complex, type 1, which is catabolized in the kidney. Accumulation of β2-microglobulin favors the formation of amyloid, the deposition of which causes dialysis-related amyloidosis (DRA). Symptoms are initially osteoarticular, but progression of the disease involves systemic manifestations including cutaneous ones.

Report of a Case | A woman in her 50s presented with a history of subcutaneous masses on her back that she had noticed 8 years previously and that were producing postural discomfort. History included hepatitis C infection 23 years earlier and bilateral nephrectomy requiring treatment with dialysis for the last 31 years. After 20 years of hemodialysis, she developed bilateral carpal tunnel syndrome and flexion contractures on her fingers; hence, she was diagnosed with DRA.

On physical examination, she was found to have 3 massive nodules projecting from her back, each with an elongated shape (Figure 1). The largest (measuring about 31.0 × 5.5 × 4.5 cm) was located over her spine, and the others (each measuring about 27.0 × 2.5 × 1.0 cm) were located symmetrically on either side of the spine. These masses had firm consistency and were covered by normal skin. Computed tomography revealed infiltration of subcutaneous fat without evidence of calcification. Examination also revealed macroglossia and some yellowish nodules (about 2-4 mm each) on the sides of her tongue.

Skin biopsy of the central mass of her back was performed demonstrating subcutaneous deposition of amorphous eosinophilic material (Figure 2A). The deposits stained positive with Congo red, showing apple-green birefringence under polarization. Immunostaining of the amorphous material was positive for β2-microglobulin amyloid (Figure 2B).

Discussion | Up to 65% of patients undergoing dialysis for more than 10 years develop DRA, and the prevalence increases over the years. Unlike other types of systemic amyloidosis, amyloid deposition in the skin or subcutaneous fat is extremely rare. However, 3 types of cutaneous manifestations related...