

Doctor, what's wrong with my tongue?

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A 75-year-old male presented with a history of generalized weakness, fluid retention and weight loss of approximately 50 lbs. over the last year. Within the last 10 months, he also noted a change in his voice and a significant enlargement of his tongue. He complained of shortness of breath on minimal exertion and persistence in his leg edema. He did not report chest pain or palpitation, and there was no history of any peripheral paresthesia or focal weakness. He had good control of his bladder and bowels. There were no musculoskeletal complaints. He bruised easily and did not have a tendency to bleed.

As for his lifestyle habits, for over 30 years, he used to smoke a pack of cigarettes a day, but finally quit the habit 15 years ago. He is a non-drinker, and is married with two children who are in good health. Family history is positive for heart problems.

For the physical examination, his pulse rate registered at 88 beats/min. Blood pressure was 98/64, with a slight jugular venous distention. His tongue was markedly enlarged (see Figure 1) and there was prominent subungual hematoma, as well as subungual petechia and bruising of his lips.

Heart sounds were distant and systolic murmur were present, while palpable peripheral pulses decreased.

As for his lungs, there was poor air entry bilaterally and dullness consistent with a double-side effusion. Bi-basilar crackles to approximately 1/3 of the lower zones were noted. Abdomen was soft with slight hepatomegaly. Lower limb showed both ankle edema. The neurologic exam was within normal limits. Chest X-ray showed bilateral pleural effusions and evidence of congestive heart failure. ECG showed sinus rhythm with low voltage and with a left anterior hemiblock Poor R wave progression was noted (see Figure 2).

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Figure 1: Markedly enlarged tongue.

What's your diagnosis?

Discussion

The patient was diagnosed with primary amyloidosis.

The natural history of amyloidosis is poorly understood and the clinical diagnosis is often not made until the disease is far advanced. The following classification is clinically the most useful:

1. Primary amyloidosis, meaning no evidence for pre-existing or co-existing disease.
2. Amyloid associated with multiple myeloma.
3. Secondary amyloidosis associated with chronic infectious diseases (*i.e.*, osteomyelitis, tuberculosis, leprosy) or chronic inflammatory diseases (*i.e.* rheumatoid arthritis and ankylosing spondylitis).
4. Heredofamilial amyloidosis-associated with familial Mediterranean fever and a variety of neuropathic, renal, cardiovascular and other syndromes.
5. Local amyloidosis – local often tumor-like, deposits occur in isolated organs without evidence of systemic involvement.
6. Amyloidosis associated with aging.

Primary amyloidosis is a disease in which major organs become damaged by homogeneous material

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that probably results from the interaction of light-chain fragments with tissue polysaccharides.

The organs most commonly affected are the kidneys (nephrotic syndrome), heart (congestive failure), joints (carpal tunnel syndrome), tongue (macroglossia), nerves (peripheral neuropathy) and gastrointestinal tract (malabsorption syndrome). About 90% of patients with primary amyloidosis and without evidence of myeloma have a small monoclonal peak of serum or urine electrophoresis. Fragments of light chains are present in amyloid fibrils, both in myeloma-associated and in primary amyloidosis without myeloma. Therefore, primary amyloidosis must be considered a monoclonal gammopathy.¹

Involvement of the heart is a common finding and is the most frequent cause of death. Cardiac amyloidosis occurs more commonly in men than in women (except the senile form) and is rare before the age of 40.²

Involvement of the cardiovascular system by amyloidosis occurs in one of four general patterns:

1. The most common is congestive heart failure due to systolic dysfunction.

2. Restrictive cardiomyopathy-right-sided findings dominate the clinical presentation, with peripheral edema a prominent finding while paroxysmal nocturnal dyspnea and orthopnea are absent.

3. An abnormality of cardiac impulse formation and conduction may result in arrhythmias and conduction disturbances.

4. Orthostatic hypotension is the fourth mode of presentation. Although most likely due to amyloid infiltration of the autonomic nervous system or of

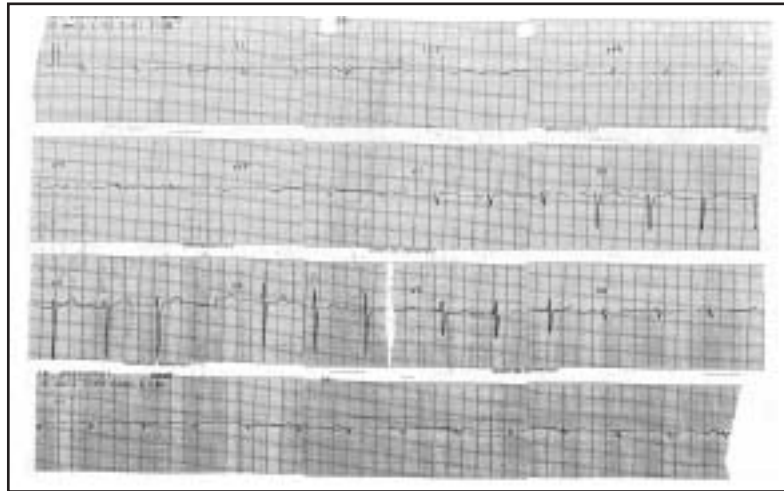


Figure 2: ECG-low voltage and poor R wave progression is noted.

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blood vessels, amyloid deposition in the heart and adrenals may contribute to this manifestation.

Physical examination often reveals findings of congestive heart failure with systolic murmurs due to atrioventricular valvular regurgitation. Particularly in patients with restrictive cardiomyopathy, jugular venous distention, a protodiastolic gallop, hepatomegaly, peripheral edema and a narrow pulse pressure are present. The chest rentgenogram usually shows cardiomegaly in patients with the clinical and hemodynamic picture of congestive cardiomyopathy, although heart size may be normal in patients with restrictive form. Pulmonary congestion may be prominent in patients with congestive heart failure. Pleural effusions are common.¹

The electrocardiogram is frequently abnormal and the most characteristic feature is diffusely diminished voltage, occurring in approximately half the patients. Myocardial infarction is often simulated because of small or absent R waves in right precordial leads or less frequently, by Q waves in the inferior leads. Left-axis deviation is seen in more than half the patients. Arrhythmias are common, particularly atrial fibrillation. Various forms of atrioventricular conduction defects are often seen and have been found in one-third of patients with cardiac amyloidosis.¹

Echocardiography most commonly reveals increased thickness of the walls of the ventricles and increased left ventricular mass. The left ventricular cavity is usually normal or small in size, and wall excursions are often reduced. A pericardial effusion is common, but rarely results in tamponade. The appearance of the thickened cardiac walls is often distinctive on two-dimensional echocardiography, demonstrating a granular sparkling texture, presumably due to the amyloid deposit.¹

The specific diagnosis of amyloid with appropriate stains with Congo red, and the presence of green birefringence should be sought by polarized microscopy.

Rectal biopsy has been the single most useful diagnostic procedure, combining the attributes of relative ease of performance, sensitivity and safety. **Dx**

References

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2. Braunwald E. *Heart Disease. A textbook of Cardiovascular Medicine*. Second Edition. W.B. Saunders, Philadelphia. 1984, pp. 1422-25.