The protean manifestations and vagaries of primary systemic amyloidosis are well known and its ability to imitate many disease complexes by involvement of various organs of the body may lead to erroneous diagnosis. The purpose of this case report is to reiterate the importance of a high index of suspicion in the unusual clinical presentations of primary generalized amyloidosis.

Case Report

A 45-year-old housewife was admitted to hospital with complaints of shortness of breath and anaerobic since 2 months. She had history of dysphagia and swelling in neck since one and a half months, dysphagia initially to solid gradually increasing to and now taking liquids with extreme difficulty. The patient had weight loss and anorexia since 1 month, difficulty in swallowing and inability to open mouth fully. At physical examination, she was pale and showed flattening of facial expression. Her blood pressure was 120/70 mmHg, pulse 80/min regular, temperature 98°F. Coarse rales were heard in both lung bases. Her abdomen was tense and liver enlarged 2 cm below right costal margin. The spleen and the kidneys were not palpable. Oral cavity showed severe glossitis, stomatitis, and aphthous ulcers on tongue, floor of mouth was hard and fibrotic. JVP was raised and generalized thickening of skin noted.

Investigations

Urinalysis: specific gravity: 1.010; Fasting blood sugar: 90 mg%, non-protein nitrogen: 34 mg%, uric acid: 3.5 mg%, total serum protein: 5.4 gm%, albumin: 2.9 gm%, haemoglobin: 9.3 gm%, ESR: 50 mm at the end of one hour. Total leucocyte count(TLC) and Differential count were in normal range. X-ray examination of chest showed left middle zone consolidation and right pleural effusion. The pleural fluid examination revealed it to be transudate. ECG showed a non-specific pattern. Biopsy of stomach showed moderate chronic active gastritis. Biopsy material from skin revealed deposits of hyaline material within blood vessels. Due to these findings along with clinical presentation, clinical impression of scleroderma was strongly considered. She was frequently nauseated and vomited on many occasions. Her pallor became more marked. Eventually patient became weaker and emaciated and died. A complete autopsy was requested in view of death within 24 hours of hospital stay.

Scleroderma – An Unusual Presenting Feature of Primary Systemic Amyloidosis


Abstract

The case presented in this report is one of 45-year-old female with dysphagia and swelling in the neck since 2 months, breathlessness, history of inability to open the mouth and course of events resembled scleroderma and diagnosis of primary systemic amyloidosis could not be established until autopsy. At autopsy skin, oesophagus, salivary glands, tongue and all the parenchymal organs like spleen, kidney, liver, pancreas, lungs and thyroid showed amyloid deposition which exhibited apple green birefringence by congo red stain and was resistant to potassium permanganate test (KMnO4). It was thus labelled as primary systemic amyloidosis. The purpose of this case report is to reiterate the importance of a high index of suspicion in the unusual clinical presentations of primary generalized amyloidosis.
Autopsy examination

Gross findings: The general examination of the body revealed marked oedema of the left leg and foot. Her skin everywhere was pale and slightly firm in consistency. Marked generalized muscle wasting was demonstrated, in particular of the upper extremities and hands. Right lung showed lower lobe consolidation. Her heart weighed 350 gm, the right ventricular muscle was thickened and there was diffuse firmness of the myocardium with moderate pallor. Her liver weighed 1200 gm and demonstrated no gross abnormalities. Spleen weighed 300 gm and firm in consistency and cut surface was homogeneous. The wall of the oesophagus demonstrated a diffuse pallor and was thickened (Fig. 1). The gastrointestinal tract was unremarkable. The kidney section (coronal) showed prominent stiff thickened vessels at the corticomedullary ratio (Fig. 2).

Microscopic findings

There was diffuse infiltration and vascular involvement by the acellular, eosinophilic homogeneous material in heart, lungs, thyroid, and skin. The parenchymal organs like liver, spleen, kidney, and pancreas and showed extensive hyaline homogeneous deposition around the vascular channels (Fig. 3). The salivary glands and also thyroid gland showed interstitial and vascular involvement by hyaline material (Fig. 4). Congo Red staining was done and read under polarising microscopy, which showed apple green birefringence confirming diagnosis of amyloidosis (Figs. 1d, 2c, 4d). The potassium permanganate (KMnO₄) treatment was done and Congo Red was resistant to KMnO₄, ruling out AA type of amyloidosis and based on which the diagnosis of Primary Systemic Amyloidosis was made.

Discussion

Amyloidosis is of global relevance. Once
considered as a rare disease, incidence of amyloidosis has increased over last few years. Whenever, a predisposing disease was identified, the amyloidosis was classified as secondary. If no identifiable cause was found, the same was considered primary.

There are reports of primary amyloidosis affecting skin, presenting generalized morphea like scleroderma. The protean manifestations and vagaries of primary systemic amyloid are well known and its ability to imitate many disease complexes by involvement of various organs of the body may frequently lead to an erroneous diagnosis. The pattern of organ involvement by amyloid varies depending on type of amyloid. Primary amyloidosis tends to involve heart, gastrointestinal tract, nerves, skin, tongue, respiratory system and bladder. Secondary amyloid predominantly affects parenchymal organs like kidney, liver, spleen, lymph nodes, adrenals and thyroid. Symptomatic amyloidosis of the tongue, skin and oesophagus contributed to scleroderma like illness which is a rare but well documented condition. Amyloidosis involving the oral cavity and submandibular salivary glands in the form of a mass and presenting with neck swelling is rather uncommon was also seen in our case. Variable amounts of acellular, eosinophilic extracellular material stains pale red with Congo red stain, and exhibits a characteristic apple – green birefringence under polarized light which was seen in our case thus confirming the diagnosis of amyloidosis.

Potassium permanganate modifies the molecular conformation of secondary AA amyloid fibrils so that it no longer stains with Congo red. Therefore this technique is very useful in distinguishing secondary amyloid fibrils from other fibril types.

Since amyloid occurs widely throughout GIT, sites other than rectal mucosa have to be considered for biopsy, e.g. oral mucosal biopsy. Also abdominal fat aspiration technique is currently method of choice because of its sensitivity. (approximately 80% of patients known to have amyloid are positive) and because it is a procedure easily performed at bedside by any member of medical team.

Conclusion
Amyloidosis that was considered a rare disease entity a few years ago is not so uncommonly encountered these days. The need for a detailed and comprehensive patient evaluation cannot be overemphasized. So also is the role of a high index of suspicion and awareness of unusual manifestations of primary amyloidosis in adults, which would lead to more frequent recognition of this disorder during life.

References


HEART DISEASE : BREAKING DOWN BARRIERS

The benefits of tight blood pressure control are presented by Paolo Verdecchia and colleagues in the Cardo-Sis trial. David Holmes and co-workers from the PROTECT AF study assess the use of the WATCHMAN device for percutaneous closure of the left atrial appendage. And the value of albuminuria as a prognostic indicator in heart failure is highlighted by Colette Jackson and her team.

Around the world, people are sidelined on the basis of colour, ethnicity, sex, disability, illness, poverty, or age. This is epitomized by the example of mental illness: cardiovascular disease is the leading cause of death in people with psychotic illness or bipolar disorder, occurs more often in people with mental ill health, and the outcomes in this group are worse than in those of the combination of increased prevalence of cardiovascular risk factors, weight gain associated with some antipsychotic drugs, and issues relating to social exclusion, such as barriers in accessing care. Rheumatic heart disease, rare in affluent populations, affects 15.6 million people globally. Within-country variation is evident in Australia, with the Indigenous population accounting for 94% of all deaths due to rheumatic fever.

Discrimination comes in many different shapes and forms, some more obvious than others; however, cardiovascular disease affects populations without prejudice.