INTRODUCTION

- Amyloidosis refers to extracellular tissue deposition of organized fibrils which are composed of low molecular weight subunits of a variety of proteins.
- Primary Immunoglobulin light chain (AL Amyloid) is typically caused by a plasma cell dyscrasia and is due to deposition of protein derived immunoglobulin light chain fragments.
- This can occur in association with multiple myeloma or less common Waldenstrom macroglobulinemia, non-Hodgkin lymphoma or chronic lymphocytic leukemia.
- We present a case of light chain amyloidosis secondary to multiple myeloma which presented in the setting of macroglossia and eventual cardiac failure.

CASE PRESENTATION

- A 47-year-old male with no past medical history presented to the emergency department with a one-week history of tongue swelling causing difficulty swallowing and a three-month sixty-pound weight loss.
- Computed Tomography (CT) Scan demonstrated macroglasia and a lytic lesion in the T4 spine.
- CT Chest/Abdomen/Pelvis showed multiple lytic lesions in the sacrum and femoral neck (see Figure 2).
- Laboratory studies were significant for a Hemoglobin of 9.4, Creatinine of 1.0, and Calcium of 9.6. Serum Protein Electrophoresis and Urine Protein Electrophoresis showed decreased total protein, albumin, beta and gamma globulins. Kappa Light Chain was 71.9, lambda light chain was 0.87 with an increased Kappa: Lambda Ratio of 82.6.
- Beta 2 Microglobulin resulted elevated at 4.2.
- Bone Marrow Biopsy showed limited subcortical marrow with focal diffuse involvement by a kappa restricted myeloma. Congo red stain showed extensive involvement by amyloid deposition in the soft tissue.
- During his admission, the patient developed ventricular trigeminy and EKG showed low voltage QRS complexes (see Figure 1).
- He underwent Echocardiogram which showed LVEF of 47%, global longitudinal strain at 11.6% with borderline global hypokinesis.
- He underwent Cardiac MRI which resulted with high pretest probability of cardiac amyloidosis (see Figure 3).
- Orthopedic Surgery was consulted, and the patient was started on Hematology and Oncology was consulted, and the patient was started on Cyclophosphamide, Bortezomib, and Dexamethasone (CyBorD) chemotherapy.
- He completed two cycles of chemotherapy.
- He underwent Cardiac MRI which resulted with high pretest probability of cardiac amyloidosis (see Figure 3).
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- Orphedc Surgery was consulted, and he underwent bilateral intramedullary nailing of the femur to prevent any future fractures.
- Hematology and Oncology was consulted, and he was started on Cyclophosphamide, Bortezomib, and Dexamethasone (CyBorD) chemotherapy.
- He completed two cycles of chemotherapy.
- Two weeks later he went into cardiac arrest and passed away due to cardiac failure.

Figure 1: EKG showing low voltage QRS complexes; prolonged QT interval.

Figure 2: CT Pelvis showing multiple lytic lesions in the sacrum and femoral neck.

Figure 3: Cardiac MRI showing diffuse subendocardial deposition of amyloid.

DISCUSSION

- Primary amyloidosis, commonly referred to as immunoglobulin light chain (AL) amyloidosis is a monoclonal plasma cell proliferative disorder which is characterized as tissue deposits of light chain fragments leading to organ dysfunction.
- The incidence of AL amyloidosis is approximately one fifth that of multiple myeloma (MM), making it an uncommon disease.
- We presented an interesting case of MM presenting as amyloidosis in the setting of macroglossia and cardiac failure.
- These cases are rare, and a high degree of clinical suspicion should lead to timely diagnosis and initiation of autologous hematopoietic stem cell transplant or initiation of a bortezomib-based chemotheraphy regimen.
- Prognosis depends on the organ systems involved and can progress rapidly at any time.
- Therefore, if the patient is not a transplant candidate and if the disease progresses at any time, then an alternative systemic therapy needs to be assessed and implemented.

REFERENCES

- Vaxman I, Gertz M. When to Suspect a Diagnosis of Amyloidosis