# A SUCCESSFUL CASE OF HEART TRANSPLANTATION FOLLOWING RESTRICTIVE CARDIOMYOPATHY DUE TO SYSTEMIC MASTOCYTOSIS

**Ana Braga, Giovanna Koukoulis, José Calquinha, Bruna Ferreira, Celina Afonso, Rosa Gouveia, Martinha Chorão, Sância Ramos, Maria José Rebocho, Miguel Abecasis, Miguel Mendes, José Pedro Neves**

**Hospital de Santa Cruz – Centro Hospitalar de Lisboa Ocidental**

## INTRODUCTION:

Systemic mastocytosis (SM) is a rare heterogeneous disease characterized by the accumulation of neoplastic mast cells (MCs) in the bone marrow and other organ organs/tissues[1].

Most adult patients have indolent forms of disease with no organ dysfunction but in advanced variants of the disease the malignant expansion and accumulation of MCs can lead to organ damage[1]. MCs physiologically reside in the interstitial space between the cardiomyocytes in close proximity to nerves and may be associated with the generation and progression of arrhythmias[2].

Activated cardiac MCs release potent proinflammatory and profibrotic mediators and had a demonstrated role in congestive heart failure animal models but case reports of heart failure in SM patients are rare[2]. Available evidence describes usually no pathologic alterations in systolic left ventricular function and systolic and diastolic left ventricular diameter[2]. However a diastolic left ventricular dysfunction and left ventricular hypertrophy may be associated with SM but symptomatic chronic heart failure appears to be not more prevalent in SM patients than in the general population. Treatment guidelines for SM patients with chronic heart failure are the same as the general population and may include transplantation[2,3].

## CASE REPORT:

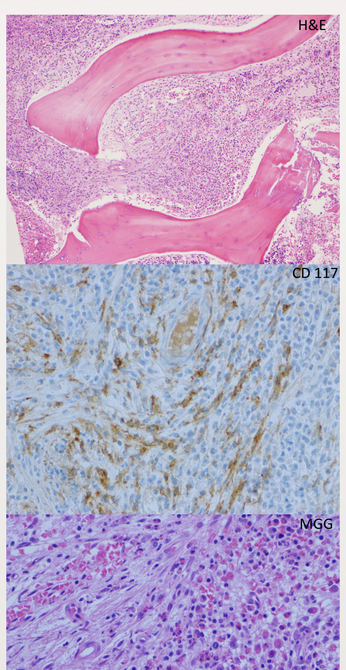
56 year-old man referred to our department due to cardiogenic shock secondary to heart failure in a patient with a previous diagnosis of systemic mastocytosis and hypereosinophilia.

**One year** — **Haematology** — **Six months** — **FIP1L1-PDGFRA negative, No MCs in BMB**

- fatigue and malaise
- mild splenomegaly

- anemia (8.6 g/dL)
- thrombocytopenia (48x10$^9$)
- hypereosinophilia (6 × 10$^9$/L)
- Serum tryptase 24.7 µg/L(<11.4)

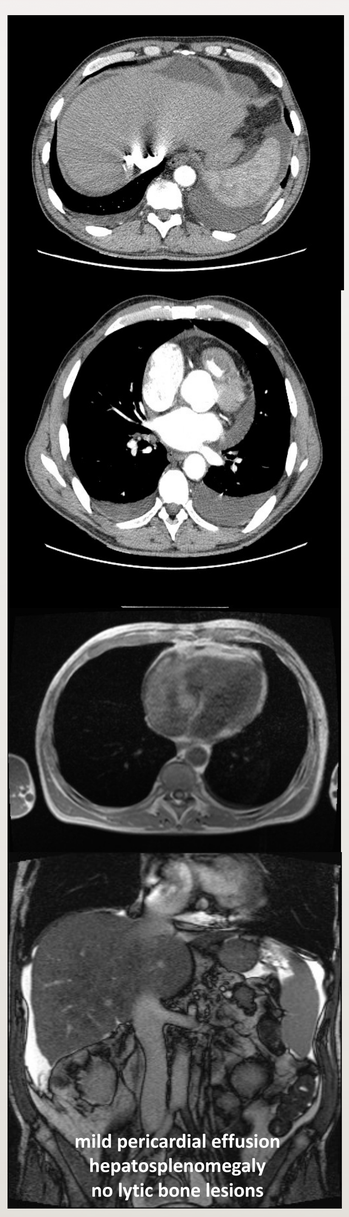
### Bone marrow biopsy



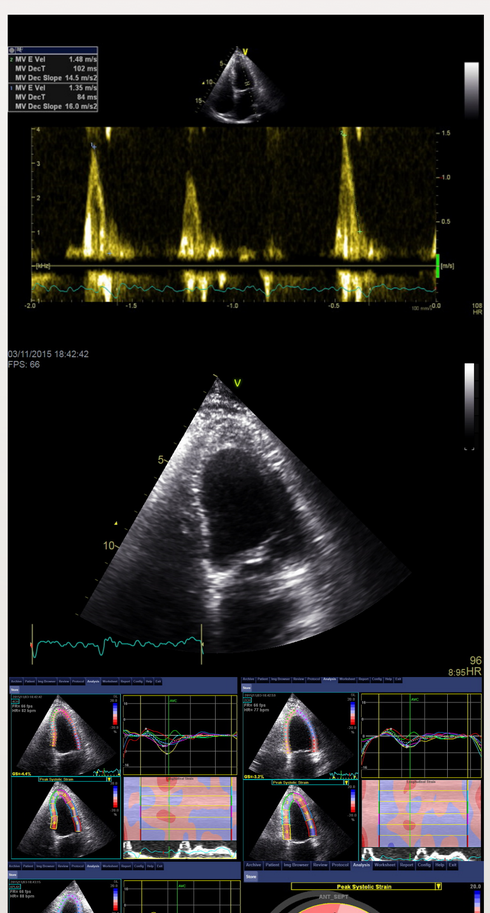

**Positive screening for FIP1L1-PDGFRA associated with myeloid neoplasm (systemic mastocytosis)**

**Imatinib mesylate**
(100 mg/day -> 200 mg/day)

### CT-scan and MRI




### Echo

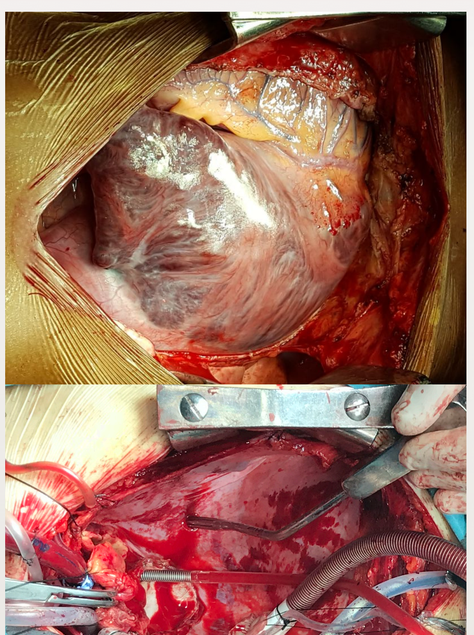


Mitral inflow pattern with severe dyastolic disfunction, tall E wave and short deceleration time;
Spherical shape of LV with apical thickness;
Severe reduction of GLS.

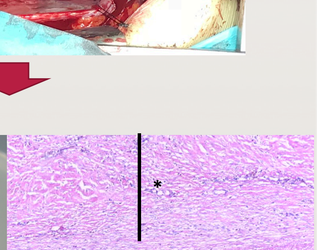
**Maintenance of Restrictive miocardiopathy**

**Accepted for heart transplant under Immunosuppression**

### Transplantation



Macroscopic right atrial view showing endocardial fibroelastosis (*) and thrombosis (T).




Microscopic right atrial view showing the myocardial wall (M) and the thick endocardial fibroelastosis (*).

**18 months free from disease in Tacrolimus + Everolimus + imatinib (2xW)**

## CONCLUSION:

Despite the pathophysiologic possibility of heart transplantation need in SM patients only one case was published and due to dilated cardiomyopathy probably not directly associated with SM[4]. We describe the first case of restrictive cardiomyopathy secondary to SM with heart failure and need for heart transplantation.

**References**

1. Ustun C, Arock M, Kluin-Nelemans HC, et al. Advanced systemic mastocytosis: from molecular and genetic progress to clinical practice. *Haematologica.* 2016;101(10): 1133-1143.
2. Kolck UW, Haenisch B, Molderings GJ. Cardiovascular symptoms in patients with systemic mast cell activation disease. *Transl Res*. 2016;174: 23-32 e21.
3. McMurray JJ, Adamopoulos S, Anker SD, et al. ESC Guidelines for the diagnosis and treatment of acute and chronic heart failure 2012: The Task Force for the Diagnosis and Treatment of Acute and Chronic Heart Failure 2012 of the European Society of Cardiology. Developed in collaboration with the Heart Failure Association (HFA) of the ESC. *Eur Heart J.* 2012;33(14): 1787-1847.
4. Moro JA, Almenar L, Jarque I, et al. Heart transplantation in a patient with systemic mastocytosis. *J Heart Lung Transplant.* 2008;27(6): 689-691.