My Amyloidosis Story

My name is Barbara, I am 53 years old and I live in Paris, France.

I was diagnosed with AL amyloidosis in June 2015. The first symptoms appeared around end of 2014. I often had marks of socks and shoes on my feet and legs and I started to feel a little tired. I am a very sporty person and used to be quite fit. Suddenly, I felt unable to get threw with my 2 km swimming distance and had to reduce it to 1km and then slow down more and more.

About March 2015, my ankles and legs really started to swell badly and my urine was very foamy. I felt more and more exhausted and short of breath. Of course, I went to see my generalist who could not find anything special, and my blood analyses seemed normal. In Mai, I had an appointment with a cardiologist who told me, I had a big « athlet heart », actually my heart was already thickened by amyloids, I started to have liquid, effusions everywhere in my body but still nobody really knew what it was.
In June, my condition was even worse, I suddenly gained 15 kg, and I ended up in hospital with pulmonory embolism ; and I had a nephrotic syndrom, 15gr/l albumines in my urine, a cardiomyopathy (thickened heart) with severe increase of cardiobiomarkers (Nt-pro BNP/Troponine). Detailed blood analyses showed a slight monoclonal gammapathy with lamdatype and then of course a very high amount of lamdalightchains. The hospital team suspected amyloidosis right away but it then still took some time to be prooved and to set the final diagnosis.

Unfortunately, my story seems to be quite usual. There is still too much time lost before the appropriate diagnosis.

I did not have genetic testing as it obviously was primary, systemic al amyloidosis. AL amyloidosis is the most commonly diagnosed form of this rare desease (85% of all cases)
In the contrary to hTTR it is not a heriditary desease, and the source of those misfolded proteins called amyloids is different. AL amyloidosis is due to a plasma cell disorder in the bone marrow, so a hematological problem and occasionally it can occur with Myeloma.

I had a biopsy of the bone marrow that showed an increased amount of monoclonal, abnormal plasmacells (9%, so just below the myeloma limit >/= 10%)

My test results from lip and tummyfat biopsies were negativ (no congo-red). Actually, there is no official diagnosis until you proove the existence of amyloid deposits in at least on of the organs.
As I was under anticoagulation treatment, no classical kidney or heart biopsy could be made in my case. Finally a transjugular kidneybiopsy prooved the amyloid deposits in my kidney. I finally had the diagnosis for AL amyloidosis with kidney and heart involvment stade III from the Mayoclinic standards. No good news of course.
I really felt horrible whis this diagnosis. The incredible thing about this story is that my father in law (my husbands father) had suffered from AL amyloidosis too and had died of its consequences in 2001.

Amyloidosis is definitely a disease that changes your life. In the beginning it was even lifethreatening for me.
I was lucky enough to respond to the treatments. I had 9 month of chemotherapy (standart drugs : melphalan, Velcade and dexametasone) with quite some side effects, like nausea, fatigue, burnt skin,….). During this time, I had to stop working and I only went back to work about 15 month after the diagnosis.

My family and friends were really a great support for me during this time but I think I contributed by keeping my positiv thinking all the time. Music was a precious help for me too. I am a hobby musician, and even if practising was more difficult at the first time, listening to music helped me a lot to cope.

Today, I am much better and had a complete hematological response. Unfortunately there has not been a very good organ response and I still have amyloid deposits in my kidneys and heart. However, my organfunctions have a little improoved.

From 2017 to 2018, I was inrolled in a clinical trial (PRONTO Neod001study) testing the effeciency of a monoclonal antibody (birtamimab). That took me some time and energy (every 28 days per IV in day hospital) Unfortunately, the study was discontinued in April 2018 in its extension phase as not concluding. It seemed a very bad surprise and disappointing for everybody.

Anyway, I am not on treatment anymore right now. I only take some diuretics to avoid edema and I try to avoid salty food but am not really on a special diet. I have a hematological and organ check every 3 months. Its a pluridisciplinary approach in nephrology, cardiology and hematology.

Most of the patients relapse sometime so it is always a difficult moment.

I only joined the french amyloidosis group in 2018, as I thought they were mostly concerned about hTTR forms and they are based in Marseille, which is quite far from Paris.

I do think it is a very good and important thing to communicate and share experiences with other patients.

I am a volonteer and frequently talk to new diagnosed patients and try to help them get threw.

Moreover, I really find the scientific aspect very fascinating and keep in touch with the latest research work about amyloidosis. Sometimes I am lucky to join hematological congresses which is always very exciting. We are several members of our support group to attend the annual amyloidosismeeting of french physicians which is useful in order to prepar another important date, our annual patient meeting.

Barbara Mazzolini (Paris, 12th of June)