LAM: lymphangioleiomyomatosis

The patient and healthcare professional perspective

Patient perspective: Iris Bassi

Iris Bassi, President of LAM Italia and European LAM Federation, was diagnosed with LAM in 2007

I remember experiencing shortness of breath several months, or even years, before my lymphangioleiomyomatosis (LAM) diagnosis. Then, in 2007, I started to have major symptoms, including dry cough, dyspnoea and a high fever, and I was hospitalised for X-rays and a CT scan. The doctors saw that my lungs were filled with cysts, and a bronchoscopy revealed LAM cells in my tissues.

My LAM diagnosis was confirmed in September 2007 at Dr Harari's hospital in Milan. Before receiving my final diagnosis, I had a lot of panic attacks due to the fact that I did not know what was wrong with me. Doctors told me that my condition was stress-related and they did not understand that I had an illness, which made me feel very anxious.

Finding out that I had LAM was frightening but, in a way, it was good to finally have an explanation for my symptoms. It surprised me, as I've always led a very active lifestyle and have never smoked. When I was told that my lungs were like those of someone who smokes 100 cigarettes a day, I was very shocked. I felt despair but tried to seek a solution. I told myself that, just because there is currently no cure for LAM, it does not mean that there will not be in the future. I founded LAM Italia and the European LAM Federation, through which I try to promote research and therapy for the disease.

After my diagnosis, I carried on experiencing cough and fatigue, and was quite weak. I tried to improve my symptoms by taking care of my body: eating a healthier diet rich in fruit and vegetables and doing mild exercise like yoga and lifting light weights. I was also prescribed bronchodilators. Since my diagnosis, my condition has been relatively stable. My daily symptoms are cough and anxiety due to the fact that I have a rare disease, but I have not experienced the more severe effects, like pneumothorax.

I have worked hard to manage my negative state of mind, which can affect breathing and worsen the effects of LAM. I speak to many patients who get stuck in a vicious cycle of fear and panic, which causes them to not look after themselves properly and leads to more physical weakness stopping them from being able to face everyday life.

As the President of LAM Italia and the European LAM Federation, I am very often in contact with people newly diagnosed with LAM. The first thing that I tell them is to make sure that they have ongoing support from a national centre with experience of LAM. I also try to give them a message of hope. LAM is not the same in everyone, there are both moderate and aggressive forms, so I tell them not to compare themselves with other people with LAM as this causes unnecessary stress. I also advise them to stay as healthy as possible, and recommend that they share their experiences with other patients through patient organisations and support groups.

I think that it is very important to raise awareness of LAM among family doctors, who are often the first point of contact for women with the disease. And it would be great if, in the future, everyone were to know about LAM.

It is also vital to promote trials of new potential drugs at a European level and to

communicate information to patients about these new therapies. This could accelerate the process of getting a cure. As LAM is a rare disease, it requires more effort from the LAM community and physicians.

The healthcare professional's perspective: Sergio Harari

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LAM is a rare multisystem disease that mainly affects women of childbearing age. It is characterised by progressive cystic destruction of the lung, lymphatic abnormalities and abdominal tumours. LAM is a rare disease, although its profile has grown in recent years, and we are, therefore, becoming aware of more people that are affected.

I saw my first LAM patient in 1989. She was a young woman with a history of emphysema; she was receiving oxygen therapy and was sent to me for a lung transplant evaluation. I saw that she had unexplained emphysema and angiomyolipomas, and I diagnosed her with LAM. Since then, I have been interested in the disease. Iris's experience is very typical of someone with LAM.

Suspicions of LAM should be raised in any young woman with a history of emphysema, unexplained dyspnoea, angiomyolipomas or pneumothorax. The course of LAM and symptoms vary greatly from patient to patient and according to the severity of the disease. The symptoms include dry cough, dyspnoea, pneumothorax and chylous effusions. In almost 50% of women with LAM, pneumothorax is the first manifestation of the disease.

Until a few years ago, many people with LAM had to undergo invasive procedures to receive a diagnosis. In most people, this can now be done using a noninvasive procedure. We now know about biomarkers which are very useful for diagnosis, and for monitoring the disease, like vascular endothelial growth factor D. Invasive procedures should only be performed in a select number of patients and in specialist referral centres, if possible. Biopsies could be taken when a person is receiving treatment for relapsing or major pneumothorax, for example.

When treating someone newly diagnosed with LAM, I first try to calm them down. People tend to read about the disease online and become very desperate and anxious. I explain that LAM is rarely aggressive, that there is a treatment to manage the symptoms and that lots of studies for new drugs are underway. I know patients that received their diagnosis 25 years ago, who are still living with a significant quality of life. I then talk about LAM and how we can manage it. Usually we don't start treatment at the first evaluation, but follow the patient to see if the disease is progressing or stable; we need to detect progression really early to best manage it. Not every patient needs to be treated with drugs and there are also roles for rehabilitation and bronchodilators.

We now have effective treatment to manage LAM: an mTOR inhibitor called sirolimus. The Multicenter International LAM Efficacy of Sirolimus (MILES) study, published in 2011, showed that the vast majority of people with pulmonary LAM were stabilised when taking the drug. Sirolimus is the only treatment for LAM: hormonal treatments have no role in managing the disease. However, the problem with sirolimus is that you lose the benefits of the drug when it is stopped, so you have to keep taking it. There are also patients who do not respond and those who have major side-effects; we need an alternative therapy for these patients. And we need to find a definite cure.

We need to concentrate on having experienced LAM centres that cover large geographical areas and that collaborate with each other. In my centre, we try to maintain strong relationships with doctors with a LAM patient outside of our region and advise them to use a follow-up protocol (*e.g.* performing a pulmonary function test every 6 months and regular blood level checks if the patient is on sirolimus), and we survey the data. However, I think that patients should also be seen by the national reference centre at least once a year. If a patient has other complications, *e.g.* haemorrhages related to angiomyolipomas, these should be managed by those with experience.