A patient's Journey: Life with LAM (Lymphangioleiomyomatosis)
Havi Carel, Simon Johnson and Liz Gamble

Dr Havi Carel was diagnosed with Lymphangioleiomyomatosis (LAM) in 2006. In the four years since she has learnt much about the adaptability of the human body and about some clinicians' insensitivity to the quality of life issues that can be so important to patients with chronic illnesses. She has also involved herself actively in LAM research. This is her story. It is accompanied by a clinical perspective provided by her treating physician and information about LAM provided by a UK LAM specialist.

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I need to walk my dog. She is delighted to be heading out. She is a runner. I, on the other hand, have a cystic lung disease that has damaged my lungs. Running is out of the question. At best, I can walk at a measured, matronly pace. I can only dream about running.

Four years since my diagnosis with LAM, a rare lung disease, during which I experienced dramatic decline in my lung function, followed by a transplant assessment. The transplant team said I was too well to go on the list and have kept an eye on me since. I then asked – begged – to be put on an experimental drug, sirolimus, which stabilised my condition. I went from being a healthy, fit, 35 year-old woman, with a wonderful husband and a great job as philosophy lecturer, to being ill and scared. My diagnosis felt like everything was taken away from me suddenly, unfairly and with no prospect of compensation.

For quite a while I had been feeling breathless. I could feel my lung capacity reduce. Eventually, I went to my GP. She shrieked in horror when she saw my spirometry results. ‘I’ve never seen anything like this,’ she said, ‘I have no idea what this could be’. I was alarmed and asked my father, a director of a medical screening centre, to arrange a CT scan for me. I had the scan in the morning, and returned to collect the results in the afternoon.

The radiologist clearly did not want to break the bad news to me in person. He said ‘sit down. I’ll let you read about what you’ve got’ and handed me a heavy diagnostic manual. It was open at a page, the top of which said: ‘lymphangioleiomyomatosis’. I read the description of this strange disease, my illness, and got to the bottom of the page: ‘prognosis: ten years from onset of respiratory symptoms’. I could not speak or move. My only thought was: 45; I will be dead by the time I am 45.

The first month was terrible. I tried to reach out to my friends, but many of them mumbled that they didn’t know what to say and disappeared. A new awkwardness entered my life. The awkwardness of nurses doing my breathing tests showing a sharp decline; the awkwardness of my parents, paralysed by their inability to help; the awkwardness of the healthy, to whom illness is a foreign and exotic land.

In the months that followed I learned that illness is multifaceted and complex; that it is a process, not a static entity and that it is possible to go on living well and experiencing wellbeing even within the context of a terrible and incurable illness. This surprised me, as I had always thought of health as the *sine qua non* of happiness. And yet, all of a sudden, I found myself changing, responding to constraints, learning to make sense of my life in light of my illness. The work of realigning my life, its values and the meaning I gave its different elements, surprised me.

**Amazing adaptability**

At first, I still tried to move around briskly, run for the bus, cycle up a hill. After nearly fainting a few times, I realised I had to slow down. But there was a more subtle change instigated by my body. Whenever I tried to do something that was no longer
possible, like chew gum while walking, or lift and swing my nephew, my body quietly registered the failure and removed the action from my bodily repertoire. This change happened slowly, subtly, but cumulatively. The result was quite amazing: I stopped sensing, and therefore no longer think much about, what I cannot do.

I started seeing that the lived experience of illness was very different from its textbook descriptions or third-person view of it. I realised that the first-person experience of illness was varied, changed continuously, and shifted from being at the foreground, as during my diagnosis, to being in the background. When it is in the background I think I am no less happy, but a lot wiser, than I was before I was ill.

This first-person perspective became important to me. I felt that during my frequent dealings with medical and healthcare professionals it was neglected. No one asked me what has changed in my life; what have I had to give up because of my illness. Overlooking the lived experience of illness is a mistake, because there is so much important knowledge to be gleaned from it. For example, knowing what the most effective intervention might be; being able to help the patient retain their life-world despite their illness. The ultimate aim of medicine is to help those who are ill regain their life, habits and activities. But it is impossible to do this without knowing what these are and how they have been affected by illness.

Helping researchers help me

As it turned out, the 10-year prognosis is now out of date. My family and I did a lot of research into the illness and made contact with clinicians, researchers and LAM organisations to get the best advice. I joined two LAM organisations, which are a source of support and information but also gave me the opportunity to assist the search for a treatment. Joining a mailing list and meeting other women with LAM was of great help, as with rare diseases in particular, a lot of information is held by patients who have had the condition for many years. But raising funds for research and coordinating tissue retrievals is even more important to me. I am now the European tissue coordinator for the LAM Treatment Alliance, a LAM research organisation that collaborates with another US-based organisation, the National Disease Research Interchange (NDRI).

The NDRI keeps a database of pre-consented patients with a particular disease and of researchers working on that disease. When a patient has a procedure or dies, her tissue is retrieved and shipped to researchers around the world. Since the scarcity of tissue is a big obstacle for LAM research, making sure no tissue is wasted is crucial. I explain to LAM patients about the importance of tissue donation and coordinate individual shipments from patient to researcher.

Finally, I also wanted to incorporate my illness into my work as a philosopher. I began to write and talk about illness, which linked to my work as a philosopher interested in embodiment. Philosophers spend much of their time thinking about consciousness and its relationship to the body. Through my illness I realised that illness is one such relationship, the importance of which has not been recognised by philosophers. I now head a research project (funded by the Arts and Humanities Research Council) on the concept of illness. I also published a book, *Illness*, which ties together the philosophical ideas around illness with my experience of it. It is my hope that by talking about the experience of illness with medical professionals I can make this experience and its dramatic affects on the patient’s life better understood.
Box 1
Background and clinical data
Dr Simon Johnson

Lymphangioleiomyomatosis (LAM) is a rare disease which almost exclusively affects women; generally presenting before the menopause. The disease can occur sporadically or in association with the autosomal dominant disease tuberous sclerosis complex (TSC). Sporadic LAM occurs in only 1 in 400,000 adult women but LAM is present in up to 40% of adult women with TSC when examined by computerised tomography\(^1\). In LAM an abnormal clone of benign cells (called LAM cells) metastasize and proliferate diffusely in the lungs and axial lymphatics and can also form angiomyolipomas\(^2\): a mixed lineage benign tumour which occurs in the kidneys of almost half of patients with LAM\(^3\). In the lungs, LAM cells form nodular proliferations causing multiple lung cysts, probably by secreting proteolytic enzymes. The cysts replace the lung parenchyma and can rupture leading to pneumothorax.

Respiratory features are generally predominate with the majority of patients presenting with either dyspnoea or pneumothorax. Occasionally, bleeding from angiomyolipomas or enlarged abdominal lymphatic masses are the presenting problems\(^1\). The clinical spectrum of the disease is highly variable with some developing progressive respiratory failure punctuated by recurrent pneumothorax, although some patients may stay stable for many years. On average, in 10 years from the first symptom, over half of patients are breathless walking on the flat, one quarter use supplemental oxygen and approximately one in ten will have died\(^4\). Diagnosis is either made by lung biopsy or the combination of lung cysts visible on high resolution computerised tomography and angiomyolipoma or TSC. Treatment has been mostly aimed at complications including pneumothorax and symptomatic control of dyspnoea with lung transplantation an option for patients with advanced disease. Impressive progress in the molecular pathology of LAM has demonstrated that LAM cells have constitutive activation of the mTOR pathway (a pivotal cellular kinase governing proliferation) resulting from bi-allelic loss of either TSC-1 or more commonly TSC-2; the genes abnormal in TSC\(^5\). This finding has lead to trials of mTOR inhibitors in LAM which have caused regression of angiomyolipomas and possible improvement in lung function\(^6\)\(^7\).

References


**Box 2**

*A physician's perspective*

**Dr Liz Gamble**

Havi was referred to my clinic at the age of 35 with exertional breathlessness. There was no wheeze, no trigger factors and it was not episodic. She had never smoked and was fit and active. Her lung function showed airflow obstruction with reversibility after bronchodilators. Her chest X-ray showed marked hyperinflation. She was given inhaled corticosteroids and beta 2 agonists. After some discussion about the X-ray appearance a CT scan was arranged, which showed LAM. Caring for patients with rare conditions is a challenge. They may take longer to diagnose and once a diagnosis is established there is often little good quality data available to inform management decisions. Sharing care with a national expert in the condition is helpful, allowing both patient and physician to benefit from specialist knowledge. The psychological impact of diagnosing a life-changing condition particularly in a younger person should be remembered.

**Box 3**

*Resources for patients and clinicians*

The following websites of LAM organisations provide information for clinicians, patients and their families, as well as supporting research.

The LAM Treatment Alliance, founded in 2005, aims to fast-track research focused on LAM by driving, fostering and funding collaborative, high-impact research and patient partnerships. It is based in Boston Mass. ([www.lamtreatmentalliance.org](http://www.lamtreatmentalliance.org))

The LAM Foundation, founded in 1995, seeks an effective treatment, and ultimately a cure, for LAM through advocacy and the funding of promising research. It serves the scientific, medical and patient communities by offering information, resources and a worldwide network of support. ([www.thelamfoundation.org](http://www.thelamfoundation.org))

LAM Action is the UK charity for women suffering from LAM, with 160 patient members. It provides patient support, fundraising for research and information for clinicians and researchers. It is based in Nottingham, UK. ([www.lamaction.org](http://www.lamaction.org))

LAM Australasia Research Alliance is based in Australia and working to strengthen links across the Asia-Pacific region and throughout the world. LARA aims to improve diagnosis of LAM, educate medical practitioners, support women with LAM, and fund research to accelerate the discovery of a cure for LAM. ([www.lara.org.au](http://www.lara.org.au))