You look so healthy!

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By
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In the early 2000s I appeared to be a healthy mother of three working full time and approaching fifty. In addition to my family and my job as a self-employed consultant and educator in art and antiques, I also did a variety of rewarding voluntary work. And I had time over for singing in particular, as well as a certain amount of dancing. My life was very much like that of many people my age who live in a big city with everything big cities have to offer. In January 2002 it was time for the annual meeting of one of the societies of which I was chair. The previous week I had had a fever and an irritating cough. I had found myself increasingly prone to coughing, not only when I had a cold. Coughing, short of breath and fortified with antibiotics and painkillers, I went to the meeting. Afterwards life went on as usual, with various meetings, work, travel and the family. Self-employed people do not usually allow themselves the luxury of being ill. Naturally, I worried about not being able to get rid of my cough and that my condition was getting worse, but it is easy to get used to a condition to the point where it feels normal. For me it was normal to always have chest discomfort, to cough, and to become out of breath just by going up a few stairs. This had been the situation my entire adult life, although now it was more pronounced. My doctor, whom I had gone to see over the years, and I usually agreed that my condition was a result of prevailing circumstances. One example was when I had a check-up at the age of 35 and suddenly had to stop in the middle of the exercise bike test. Something had gone wrong with my heart and my breathing. The doctor, who was monitoring the test, looked at me and said “these things can happen when you work a lot and have three small children”. The same thing happened when I did a similar test 12 years later. On that occasion the doctor wrote to the physician who had referred me saying that further tests might be advisable. But no further examinations were made. The samples taken over the past few years had not revealed anything untoward – my blood count (Hb) was good, and “I did look healthy”. The possible sign that something was not as it should be was that my PEF was about 50% of the expected level. (PEF= peak expiratory flow). This was interpreted as a sign of asthma, and I was given medicine for it. During the spring, after the annual meeting I mentioned earlier, I was at a conference in the US. There I met people whom I had met the previous year, who had seen me in a performance of music and dance. Several of the participants inquired after my health, having seen how I was dragging myself around the conference area. I confirmed that I was not feeling well, but told them that tests and examinations had shown I was in good health because after all, “I did look healthy”. I had also had a cough and breathing

difficulties during my three pregnancies 20 – 25 years earlier, but was told the same thing. When visiting an ear, nose and throat specialist during my first pregnancy my problems were explained thus: “that’s how it is when you’re expecting a child, but it will pass eventually”, and “you do appear to be in good shape”. Also: “Bricanyl will most likely be of some help”. And it was of some help. During the years between my pregnancies as I approached my fiftieth birthday I also sometimes experienced other acute symptoms in various parts of my body. I was usually told “that’s how it is sometimes at your age, but you really do look healthy”. When I saw specialists in different fields I described my symptoms and asked whether they might be related. But no – everyone focused on their own specialist field and did not consider it reasonable that such diverse symptoms as mine could be linked to one another. Samples taken on a number of occasions when I was suffering from various conditions, revealed nothing unusual; the answers I received were that “the tests do not show any signs that anything is wrong”. Mention is seldom made of the fact that we still do not have complete knowledge about possible ways of taking samples, making examinations and interpreting their results. It might be a good idea to bear in mind that we have not yet discovered everything. Our knowledge is incomplete when it comes to health care/the human body and all other areas. Will we ever be able to know everything? My own search Some months after the US conference the situation worsened. I began searching for information on the internet about various lung conditions and rheumatic diseases. I discussed the issue with some friends who work in the health sector in Sweden and elsewhere to find new approaches and explanations for what was happening to me. I felt that time was running out on me at an ever increasing rate. I could really feel how I was getting worse day by day. And I was not the only one who noticed the deterioration – my family did too. Sub-consciously I got around to doing things that should have been done a long time ago, but that had been easy to put off. I wrote a will, reorganised and ended some of my professional commitments. It had become quite clear to me that time was starting to run out. At the same time as I was putting all my affairs in order I nonetheless continued to work as usual and make commitments. On a choir trip during the autumn one of my fellow choristers came up to me and said “Anette – you’re not well”. “There seems to be something wrong with your breathing”, and “Do you have a good doctor?” I then reflected that friends whom I did not see very often, like those at the conference in the US and now my fellow chorister, could easily see that something was wrong. They had not had any test results on which to base their opinions. They merely made day-to-day observations, noticing the difficulties I was having in walking and talking. I was “assessed” on the basis of what I had previously been able to do without hindrance. I said that I had been to see my own doctor, who had made the examinations he considered to be relevant. My friend from the choir offered to call her sister, who was a lung specialist at one of Stockholm’s hospitals. I had a

short telephone conversation with her during which I described the state of my health, upon which she told me I was welcome to come in to the clinic after the weekend. Once there, I described how my health had been since my teens. This might seem somewhat superfluous, but, as usual, I wanted to give as complete a picture as possible. Samples were taken and relevant examinations made at this first visit: a lung X-ray, blood samples and breathing tests. The X-ray and blood samples revealed nothing out of the ordinary, but the spirometry test showed that something was wrong. The diagnosis was probably untreated asthma. Several examinations were made, but everything else was normal. After a period of treatment for asthma I was no better; my condition had continued to deteriorate. Now it was a tremendous effort for me to walk more than a few metres. I almost fainted sometimes and the pressure on my throat was unpleasant to say the least. Friends working in the health sector told me it was possible to consult other lung specialists. I asked one of them if I could have oxygen. The answer I was given was “no, you can become addicted to it”. I also asked about a lung transplant, but received no reply. Perhaps I asked too soon. After all, I was told I did look healthy. I asked another doctor whether it could be LAM (Lymphangioleiomyomatosis), a disease I had read about on the internet and elsewhere. The doctor replied “no – it’s so rare”. One of my thoughts was then – who gets a rare disease? My specialist at the hospital, my good friend’s sister, continued to ponder my situation, and sent me for a number of examinations. A CT scan of my lungs revealed something strange that could not be identified. I now know that she considered the possibility of LAM, but that her colleagues did not agree with her about the diagnosis. It must have been too unusual. Since the resources at that hospital had been exhausted, I was referred to the next “level” at the next specialist hospital. The scan images would be examined there and I would be contacted shortly. By this time I had also had to capitulate on the work front. I was put on part-time (50%) sick leave. As for my lung capacity, which was one of the many criteria to be considered, my diffusion capacity was 27% of the expected level. It was now also time for me to deal with matters that I had no knowledge or experience of, namely the Swedish Social Insurance Office, occupational therapists and later on welfare benefits officers. Diagnosis – asthma again? Time passed and I waited for an appointment at the next specialist clinic. Months went by without an appointment. Contact was made with specialist clinic No. 2 to speed things up. Some time later I was given an appointment. When I finally met my new doctor I told my story. I was quickly told that this was “untreated asthma”. I protested; that diagnosis had already been tried. “Haven’t you seen the scan images from clinic 1”? The answer was no. What scan? Well, no-one there could help me. We agreed that there was no point continuing at that time. I would be given a new appointment in a week or two after my new doctor had looked at the CT scan images. The next appointment went better. My doctor had looked at the unusual images of my lungs. This time

the diagnosis was COPD/emphysema, even though I had never smoked. We discussed various possible reasons for my status. One issue that came up was my work and working environment. I was referred for new X-rays and tests, including a treadmill test. Meanwhile, the images were also sent to the Sahlgrenska Hospital in Gothenburg for further comments and, if possible, an alternative diagnosis, since I was highly sceptical of the diagnosis I had been given. I received the answer in mid-June 2003. My doctor called and confirmed the diagnosis on the basis of the opinion given by Sahlgrenska Hospital. It was LAM, which I already knew from other help I had received and my own investigations. My friend's sister, a doctor, had been right in her diagnosis theory, but her colleagues had not supported her view. The prognosis in perspective With the benefit of hindsight, I have glad that I was not diagnosed with LAM 25 – 30 years ago when I experienced my first symptoms. (I had probably had a minor pneumothorax (which healed on its own) when my youngest daughter was born in 1984.) Since the disease was then (and still is) rare, the advice I would have been given on the basis of knowledge at the time would have been not to have any (more) children. My body's production of oestrogen would have been stopped by chemical or surgical means because it was considered that oestrogen caused the disease. My family life, now with three children, would definitely have been different and poorer. Moreover, it is unlikely that I would have been able to commit as I have done to my profession or other interests in the arts, in music and ZONTA (a worldwide non-political and independent organisation of women in executive positions whose aim is to advance the status of women in the world). In the past it was considered that LAM sufferers survived for 5 – 10 years from diagnosis, and no treatment in the form of a lung transplant was available, as it is today. When one is in one's early twenties the future seems infinite, with infinite possibilities. These would in all probability have quickly been extinguished if I had been diagnosed back then. Sometimes it is good not to know. For me it would now have been more than 35 years since the first signs of the disease. I have lived a good, active and rewarding life and made the most of the opportunities before me, which I am sure I would not have done or been able to do if I had lived as an invalid and waited for the end at the tender age of 25. Receiving the answer to a question – a diagnosis – gave me some peace of mind, but naturally also led to new questions. True, I could plot a new course. I realised that I could no longer work as I had done. I would have to make the best possible use of the energy I had. I prepared my customers and everything that running a business entails for a long period of absence, perhaps never to return. But my aim was, and still is, to keep some of my activities going to allow me to continue at some later date. When I went for an appointment with the doctor now in charge of my case I took with me the information about the disease that I had obtained from the LAM Foundation in the US. I got into the habit of always being able to present the basic information and latest findings about the disease to

doctors and nurses. It was, after all, the only way of being able to discuss and adopt a stance on what should be done with me in various situations. Given that LAM is such a rare disease, I could not expect other people to know about it. Nor is it possible to make good decisions based on limited knowledge. Perhaps earlier experience of receiving the wrong treatment, as happened to me at a hospital many years ago, was part of the reason that I wanted to be sure that the health service would deal with my case in the best way. Following the diagnosis my doctor referred me for further tests and examinations, some of which I myself had requested. In some cases I waited patiently to be given an appointment. When I had not heard anything within what I considered to be a reasonable period, i.e. somewhere between one and two months, I contacted the clinics myself. The answers I received included “we have not received any referral”. Maybe they were lying among piles of paper, maybe not. I found it draining to drive clinical knowledge of my condition forward. For my part all I knew was that my condition was deteriorating. Preparing for a lung transplant Once a decision had finally been taken – to refer me to Sahlgrenska Hospital in Gothenburg – things happened quickly. The same day I was notified that an appointment was to be made I received a call from a nurse who asked what date would suit me. “You can come as soon as next Monday if you want”, I was told. I was pleasantly surprised by this swift response. We quickly agreed a suitable time and procedure. I was not used to not having to take the initiative and ask questions and ask to be examined, as I had had to do for almost my entire adult life. There had not been many times that I had felt welcomed by the health service; one often feels in the way and that one should make way for other people whose need is even greater. I know that many people feel the same way as I do. Oxygen I took oxygen with me when I went to Sahlgrenska Hospital. The samples and treadmill test before my departure had shown that I needed it. This was about six months after I had asked one of the doctors I had met along the way whether oxygen could help me. With a more open-minded approach, I could have been examined then to determine my oxygen saturation level at the time. This would also have opened the way for a discussion on the possibility of being given oxygen later on. My oxygen saturation level would not have deteriorated much in the interim, even though it had most probably been slightly better some months previously. I now know that when I took a few very slow steps the level of oxygen saturation in my blood fell to 78%, and my oxygen absorption capacity to 19%. It was exhausting for me to take these few steps, but after all, I had “looked so healthy”. After five days’ examination of my heart, lungs, kidneys and liver, along with infection and tissue profiles and other tests, I was told I was a suitable candidate for a lung transplant. I accepted this fantastic offer the second it was made. For me there was no doubt, but the normal procedure is that one should consider the matter for a time before having a meeting attended by family members at which reflections and questions can be aired. I felt at peace

after years of uncertainty and the knowledge that there are no alternative treatment methods for LAM. Now I knew that I would at least have a chance of reaching my fiftieth birthday and hopefully seeing my children begin their careers and start families. Hearing my daughters give musical performances together. Seeing my son wakeboarding and water-skiing in the archipelago, and all the other joyous moments I have shared with my wonderful children. Once I was on the “list” I was well aware of, and prepared for, the hard work that lay ahead, both before the transplant and afterwards.

To Gothenburg When I finally got the call after a ten-month wait, what was to follow was an overwhelming experience on many levels. I was flown in a specially chartered plane from Stockholm to Gothenburg, which took around 50 minutes, and was then taken by ambulance from the airport to the hospital. I arrived approximately two hours after taking the call from the coordinator at Sahlgrenska. Once I was on the ward and had been signed in, the last checks were made to make sure the lung I was to receive was in good condition and that it would suit me. Preparations were made for the operation. I was nervous and excited; it was show time... The last things I remember before losing consciousness in the operating theatre were the friendly eyes of the medical team behind their masks, that I was terribly cold and that a number of attempts were made to insert a needle in me, which was no easy thing after months of needing oxygen around the clock, virtually unable to move around without a wheelchair and assistance. Nine and a half hours later I was woken up by intensive care staff and it felt like my birthday. My whole family was standing in front of me. It really felt like being born again. I had been given a new chance. Everything had gone well and exactly as planned. Now it was time to begin the process of returning to life, i.e. as normal a life as possible. Although I was connected up to various machines in intensive care, the staff there wanted me to get out of bed so I could sit in a chair. This was to avoid unnecessary complications. With great effort I managed to meet this first challenge. I remember feeling so grateful to the family that had so generously given me a new chance by allowing the donation of a lung from a young family member whom they had just lost. It was a fantastic thing to do. Less than 24 hours later I rang my doctor in Stockholm to tell her that it had been done. She sounded surprised and pleased. I had had an appointment with her a couple of days earlier, and we had spoken on the telephone about some test results the same day I received the call from Sahlgrenska Hospital. I thought it was important to give feedback, something I myself would have appreciated – knowing what had happened to the patient. She told me later that she thought she had seen me for the last time. Recapturing life and getting to know oneself again after a lung transplant is a process of small steps. A physiotherapist helped me to regain my balance, strength, and not least to get the new lung to work, which I also did using exercises of my own from

the world of singing. From an early stage I took over responsibility for taking the medicines I needed (and still need). This is fairly uncommon. For me, this was an aspect of my recovery and a way of gaining knowledge about my new situation. During my time on the ward my rehabilitation continued, albeit not entirely without complications. Solving them required good communication between me, doctors and other medical staff. There are recognised methods of treating some problems, such as rejection. But for other conditions that arose there were no definitive answers as to the right approach/treatment, since there was limited experience in Sweden of the underlying disease from which I was suffering. In addition to the symptoms I had, I tried to describe what had happened to me. I tried to communicate as much as I knew of the latest findings in reports published in the US and UK in particular. A New transplant It is now almost eleven years since my first single lung transplant. Exactly one year ago, just after my sixtieth birthday, I had another single lung transplant. The reason was that my lung function had deteriorated again, probably because of my underlying LAM. Unfortunately, during my time with the first new lung I suffered pneumothorax in my remaining original lung a number of times, and also in the transplanted lung. In 2006, two years after the transplant operation, I also suffered chylosis, mainly in my remaining original lung, which then needed to be drained at regular intervals. The chylosis problem was resolved with a low-fat diet, simvastatin (Zocor) and treatment with Rapamune for a number of years, something I had suggested having read about ongoing studies on the LAM Foundation website. Unfortunately I suffered troublesome side-effects from Rapamune and after a time went over to traditional transplant drugs, followed by Certican/Everolimus after another pneumothorax. The new transplant was a success in purely technical terms. I recovered quickly from the operation and, for the first time in almost 20 years, I could take lengthy walks, talk at the same time, and most of all, not become short of breath. Yet the current situation is that I suffer from recurring infections that require treatment. Since it has only been a year since I received my second new lung, I must be patient and hope that everything will eventually settle down. I anticipate having another ten years, despite have LAM and undergoing two lung transplants. Over the past ten years as a transplant patient I have had the privilege of experiencing new family events, such as my children entering adulthood, embarking on careers and getting married, seeing the arrival of two small new family members, now five and two years old, and being able to watch their progress. I have also had the opportunity to work with the LAM Academy (www.lamacademy.org), which at the suggestion of Maryam Fathi, a consulting physician, I founded with her in 2008. The LAM Academy network is a joint project run by the Lung-Allergy Clinic at Karolinska University Hospital in Solna, LAM patients and their families. Our goal is to support patients and their families, disseminate knowledge of the disease and encourage research into LAM. One of the aims we have is to

establish a LAM Centre, preferably at Karolinska Hospital, where the interest and expertise is to be found. Doctor Fathi has started a LAM register, and Professor Gunnar Norstedt is ready to test his research, which is of interest to LAM patients, and which may also offer synergies in other areas of treatment. The LAM register has provided useful information: 42 women in Sweden have so far been diagnosed with LAM, out of a population of 10 million, of whom some 5 million are women. This figure suggests that the diagnosis is more common than had previously been thought. The LAM Academy also maintains contact with other LAM organisations and international LAM networks. This keeps us abreast of developments and enables us to exchange information about progress in the health sector and in the field of LAM research. We also arrange meetings for members and distribute newsletters to them with the latest news on LAM. In addition, we are endeavouring to find channels for distribution of information in health care areas where women suffering from the disease might be found. If my rehabilitation progresses well, it is my hope that I will be able to attend the LAMposium, as well as other LAM conferences in the future. If I can share my experience of anything of relevance to LAM and make any contribution to LAM research in particular, I will naturally do so.

Anette von Koch, June 2015