By the time the diagnosis is made, a lot of time has passed by. This time is necessary though to understand the disease, to be able to cope with it and to begin palliative therapy.

My two sons and I are affected by the mutated gene *COL6 A3. According to information from the genetics laboratory (database) the degree of our mutation is so extreme as to have not been recorded or registered elsewhere*. Even in our family the disease manifests itself very differently. My youngest son has been affected badly since the age of three, my eldest son is scarcely affected and my own limitations started to develop strongly only beyond the age of 30.

For years I suppressed, put off or paid little attention to the first symptoms. Many factors have been connected together only recently. The disease began insidiously. I began to walk when I was 1 ½ years old. My mother insists that I was an extremely tranquil child. The real problems began in my schooldays. I was always exhausted, had weak muscles, difficulties climbing stairs and back problems. At the time, the orthopedist said I had weak bones and prescribed a muscle enhancing product. Due to my physical constraints, I did not train for my preferred profession. When my first son was born, the constraints became worse. I could no longer open bottles. I was chronically exhausted. I had constant pain due to overworked muscles. The symptoms intensified after the birth of my second son. I had three knee operations (grade 4 osteoarthritis), a movement constraint of 90 degrees and could not build up my thigh muscle. No one could explain the poor healing and by the spring I could no longer move my hands and feet. Numerous stays in clinics and rehabilitation measures were unsuccessful. Muscle building and electrical muscle stimulation caused the condition to worsen. There were many diagnoses: undifferentiated collagenoses, burnout, fibromyalgia… Many specialist reports followed, many unpleasant comments too. With my youngest son, it all started at the age of three. One morning he had a limp, which was not greatly significant. No one could have imagined that his condition would deteriorate in such a short time. Something similar happened to the other leg – he developed pes equinus. Some weeks later he could no longer walk due to extreme pain. His hands changed and he could no longer grip properly and would fall over, always with his hands closed. The next shock was he was no longer hungry, not even for his favourite dishes. If they were chopped small, he ate with great relish. It was only when he was brushing his teeth that I noticed his small jaw opening. Light activities exhausted him. His skin became red and blotchy very quickly. At night he sweated heavily and it was noticeable that he slept a lot. This was followed by many doctors visits, stays in clinics, operations, diagnoses such as: club foot, spasticity, undifferentiated collagenoses, Achilles tendon lengthening, Botox treatments, jaw stretching, skin biopsy, muscle biopsy… we finally had a result with this one. The mitochondria (power plants in the muscles) were altered but no disease could be attributed to this. The pediatrician and the orthopedic surgeon provided him with aids and regular physiotherapy and occupational therapy. Through continuous treatment three times a week, his condition is presently stable. Cold and a lot of effort lead to a significant deterioration. I found that my constraints were

similar to those of my son and pored over all the medical books that I could get hold of. Then I came to Bethlem myopathy and thought: that fits. I approached the issue at my next appointment. The doctor compared the symptoms with those of my son and confirmed to me that we both have the same disease. The next step was to send a blood sample to a genetics laboratory. After 12 weeks we had a positive result. Following that the whole family was tested. After six weeks we got the news. That I was affected was no great surprise. That my eldest son is also affected was a huge shock. Now, after a year of knowing, I can handle it better than I could when I did not know what to expect. For my youngest son we get the necessary support in almost every respect, though some applications and their implementation through the authorities can sometimes be associated with a one-year wait. I now receive regular physiotherapy and occupational therapy, which does help me. The long period of no treatment or inappropriate treatment has naturally left its marks – I am under pain relief treatment. Physical exertion and the ability to work are severely constrained. What is also distressing is seven years of continual specialist reports during which the experts did not want to admit that their medical assessments were wrong. Even doctors specialized in second opinions and professors were doubtful despite the confirmed diagnosis.

The most important thing is this: after seven long years of ups and downs, we have a diagnosis and a rough direction. Our family is more closely knit and we'll manage the rest as well.

I want to thank the many kind people: the doctors, our wonderful physiotherapy and occupational therapy, all those who have been supportive and, of course, the kind people affected by BM whom we have already got to know.