A patient's journey: our special girl

Sandra Dunkelberg

Bergkoppelweg 2, D-22335 Hamburg, Institute for General Practice, University Hospital Hamburg

Sandra Dunkelberg *researcher in general practice*

Correspondence to: dunkelbe@uke.uni-hamburg.de

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How it began

When my daughter, Mathilda, was 5 weeks old our lives changed. It began with a seizure, probably caused by a late form of newborn hypocalcaemia. The seizure brought us to hospital for 10 days, to injections, infusions, hard, ugly knots in the head caused by calcium deposits, even to lumbar puncture. It brought pain to Mathilda and fear to her parents. There she stopped growing. There we got to know that, for unknown reasons, she had a slightly raised liver enzyme activity and anaemia. There I encountered the medical system from the other side.

I am a general practitioner. For many years I have worked mainly as a researcher at the university department of general practice. I already knew a lot about problems in the German healthcare system (and others) and shortcomings in doctor-patient communications. Now I experienced them. I met them in the form of ward physicians who changed almost daily and who were unable to answer our questions on causes and prognosis. One of them asked the senior physician to speak to me. The senior physician said, somewhat reproachfully: "You asked to speak to me." I had not, and the suggestion that I had made me feel like a difficult patient of the sort that until now had only been the subject of my research.

A difficult time

Mathilda gained little weight, even after a course of oral calcium and magnesium seven times a day. She did not grow, though I went to a breastfeeding counsellor and tried to change the way in which my milk was fed to her. I saw myself as the main problem and sought the advice of a psychologist, but Mathilda did not grow any faster. Of course I had more than one problem: feeling unable to nurture my baby, worrying about the future, and spending so much time in hospitals and surgeries instead of caring for the rest of my family.

When Mathilda was nearly 2 years old, she was still able to sit in her doll's bed

Mathilda rapidly diverged from all weight and height centiles. She also temporarily developed high liver enzyme activity, apparently in connection with trivial infections. Her maximum aminotransferase concentrations exceeded 4000 IU/l, resulting in additional stays in hospital and many diagnostic procedures, including liver biopsy. And I continued to experience the medical system in many ways, mainly negative but some positive.

The bad news: experiences with doctors we would like to have missed

Our GP paediatrician (in Germany the family doctor for children is a paediatrician) was caring but appeared uncertain and anxious. This was understandable because the condition was rare, but her anxiety reinforced my own fears at a time when I most needed reassurance. She sent me to several specialists.

While we waited anxiously, most of the specialists took many weeks to send their reports with the results of the examinations and tests, perhaps because they were reluctant to admit that they had no explanations. We waited months for the report on the electron microscope examination of liver tissue and got no further than being told that the investigation showed nothing to explain the unspecific changes seen in light microscopy. This happened in the university hospital where I work and where I know many of the people who tried to help. I wonder how such things work out for parents with no such privileged access.

One particular specialist repeatedly suggested a whole series of new diagnostic tests. But when I asked him in the same consultation about the results of previous tests and their interpretation, he was unable to tell me.

As far as I know the different specialists involved in the care of Mathilda never came together and discussed the case. Each seemed to have his or her own theory and told me something different. A geneticist suggested it might be a congenital disorder, such as Hallerman-Streiff syndrome. The pictures I discovered when I looked into this syndrome were so distressing that I hid them from Mathilda's father. We were glad that other doctors rejected the hypotheses.

The good news: doctors we love

Two physicians stood out from the crowd of those mainly offering disappointment. The first was a retired geneticist. He agreed that Mathilda's illness could be a congenital syndrome but admitted frankly that he had never seen such a case and had no idea which syndrome it might be. I was relieved that someone with his reputation and experience was able to admit that he did not know, the more so as he also explained the limited usefulness of simply knowing the name of a rare syndrome. As a general practitioner, I am familiar with the fact that many diagnoses remain unknown or uncertain and that coming to terms with this is important for the whole family.

At the university hospital where I work I met another excellent doctor, the one who helped us most, an experienced and friendly professor. The only problem from our point of view was that he retired two years later and was no longer available to us. He surprised me from the outset by addressing me by my surname, which is different from Mathilda's. He thought aloud about possible diagnoses, excluding most of them through reasoning rather than by ordering tests. He was also good at emphasising the positive aspects of Mathilda's case—her satisfactory development in all respects other than growth. He made me feel she was not retarded, just small; that she was not abnormal but special. She became our wonderful little girl again.

My own role

I started to acquire my own expertise in the specialty, but stopped when I found that my role as a mother was already hard enough to fulfil properly. I felt the pressure of responsibility. I had to find the right doctors and to be insistent but not obstinate. On the one hand, I had to save Mathilda from unnecessary diagnostic tests, often saying "no" and "why?" On the other hand, I had to ensure that she received everything she needed. We quarrelled with the insurance company about meeting the costs of attempted treatment with a growth hormone as they were unwilling to pay for its use off licence. We disputed issues around quality of life with a representative who had never met Mathilda but was sure that her quality of life was unaffected. I experienced the limitations of evidence based medicine, which was, of course, an important part of my academic career. But how many studies and reviews deal with undiagnosed health problems?

Mathilda today—small but perfectly formed

What was important for us?

- To be protected from specialists who propose more and more tests but cannot admit they do not know what is wrong
- To accept that we don't have a diagnosis and will probably never have one
- To have the opportunity to try a treatment, even though there is no evidence for its efficacy
- To clarify my own role as doctor, co-therapist, or mother
- To be taken seriously and treated with respect
- To shift from the focus on pathology and abnormality to Mathilda's health and to the positive aspects of her life

And her father? Well, he did everything he could to support me in practical issues. For example, he learned to handle the pen injections of growth hormone immediately. But he left decisions and negotiations with the physicians completely to me, partly because that's my role in the partnership anyway, reinforced by my medical background. For long periods he seemed to be in denial about the problems. I often heard him tell others on the phone that everything was fine with Mathilda when my answer had been different. When Mathilda had to undergo medical procedures, however, his attitude changed and he became upset and anxious. Our relationship grew closer during these difficult times. We are to be married in September.

Today

Mathilda had her third birthday in January. She is a lovely girl, very special—the best known child in the kindergarten, where she gets much attention because of her smile, her vigour, her ability to be noticed and to achieve what she wants, and, last but not least, because of the discrepancy between her short stature and her abilities. At 82 cm and 10 kg, she is markedly too small for her age and still far away from the third centile, but with growth factor treatment she seems to have started to catch up. Her liver enzyme activity has remained normal for the past 18 months. She still has constant marginal anaemia with haemoglobin concentration around 100 g/l. As she is in her "terrible twos" we call her our little domestic tyrant. We are all reasonably comfortable with the uncertainty of undiagnosed illness, the hypothesis of a rare congenital syndrome, and the effort involved in daily hormone injections. We get support from a self help organisation. We have found a paediatrician for day to day problems who tends to be sparing with diagnostic tests and measurement of liver enzymes and can reassure us when new problems cause new anxieties. And we found a paediatric endocrinologist, who knows my name and struggles to obtain funding for the growth factor therapy.

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