Chapter 2

A patient’s journey:

Polycystic kidney disease

‘The road ahead is empty. 
It’s paved with miles of the unknown. 
Whatever seems to be your destination. 
Take life the way is comes, take life the way it is.’

(City to City: The Road Ahead)

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Polycystic kidney disease is inherited, so most patients have seen how it has affected their relatives. Young adult patients have no symptoms and need no drugs, but kidney function will slowly deteriorate.

My family and PKD

My journey on the “PKD path” started when I was a child, but my family’s journey began earlier. My grandmother died of polycystic kidney disease (PKD), and my mother was diagnosed as having it shortly after my birth, after having a cerebrovascular accident, which she surprisingly survived. The doctor told her that she shouldn’t be afraid: “She would be able to raise her own kids.” He was right; she only started dialysis after several years of having nausea and tiredness.

When she started dialysis I was 16 years old. I always knew that my mother had a hereditary kidney disease and that I too might have it. But I hadn’t understood the consequences of the disease and its impact on life. I only knew that my mother needed to take her daily medicines and I always thought: “If this is PKD, then I can handle it. It’s not that bad to have a kidney disease.” I hadn’t known that haemodialysis and transplantation might be needed, and because of this I was deeply shocked when my mother started dialysis. I was worried about her: would she survive? But I was also confused, sad, and angry: “Why didn’t my parents tell me this before?”

Suddenly I was confronted with the impact of having a kidney disease and uncertainties about my own future. Probably my ignorance related to the fact that my parents didn’t like to talk about the disease. Their guiding philosophy was: “Don’t talk and complain about your problems, but get on and live your life. Emotions don’t help you to survive, so stay strong and go on.” Later my mother told me: “I didn’t want to know the truth, it was too intense. By putting it away, the disease wasn’t there and because of that I was able to survive and go on.” My parents always wanted us simply to be like other families. Looking back I now think this was impossible; we were not a normal family. Having a relative with a chronic disease changes the child-parent relationship and the family.

After three years of dialysis my mother received a cadaver donor kidney. The months before she received the kidney were exciting and emotional. She was very ill, and dialysis was more and more problematic as her blood pressure was so low. Every day we were happy she was still alive.
People might think that her journey along the PKD path ended after the transplant. Unfortunately it did not: her patient journey goes on, on a path with side effects of medicines and sometimes fears for the future: “Will I be able to keep this kidney for a long time?” Her path has become easier now, but it still has difficulties.

**Deciding to be tested**

Because of the confrontation with the real impact of PKD I wished to know if I had it myself. After many conversations with my parents and professionals, however, I decided to postpone “the examination.” If tests showed I had the disease, I would undoubtedly have problems with my future—with insurance, for example, and with getting a job or a boyfriend. At that age it was difficult to get a reliable result; I was advised to wait until I was at least 18 years old.

The next few years were filled with doubts: did I want to know or not? When I was 20 I decided to have the test, but the day before the appointment I cancelled it. I was too afraid of the outcome. The uncertainty didn’t disappear, though, and a few years later I decided to undergo the test after my GP said: “You have lived with doubt for more than 10 years; I think the time has come for clarity.” My mother and boyfriend accompanied me to the appointment, to support me. I was nervous and knew the next few days would be emotional, as we would be hearing the results from my GP a week later. The day of the test I expected only to have the scan, not to hear the result. But it didn’t work out like that. The scan was done in a small, dark, inconvenient room. I was shaking and trembling, and the ultrasonographer suddenly said: “I definitely see a lot of cysts.” When I asked if this meant that I had PKD, he answered: “I’m not allowed to give you any information.” I am still angry that he gave information and then refused to clarify it. Desperately upset, I told my mother the news in the waiting room. A few hours later, I gathered myself: I should be strong. After all, getting on with life was our family coping strategy. A week later my GP gave me the formal diagnosis of PKD. He admitted that he didn’t know anything about the disease and had had to read up on it in one of his books, which said I should visit an urologist. I told him I wanted to visit my mother’s internal medicine specialist, not an urologist, and despite his lack of knowledge he tried to support me and accepted my wish.
On a rollercoaster

After the diagnosis I used my parents’ “put it away and go on” coping strategy: I wasn’t ill, and I didn’t want people to see me as ill. During the early months this strategy worked quite well, but after my first visit to the specialist I collapsed. In this appointment it became clear that my mother’s cerebrovascular attack was related to the PKD, and that I also have a greater chance of getting aneurysms, which may cause a stroke. I work with people who have had strokes and the idea of being affected in this way was awful. Stunned by the known risks, I found the examination and period following it traumatic. At the moment everything is all right, but I have to be checked every 5-10 years, and I have to avoid taking risks. Hearing this news from the specialist, I was unable to “be strong and just go on” any longer. My emotional state changed; I felt as if I was on a rollercoaster. But the support I received enabled me to handle the situation—people listening, just being there, accepting my emotions, and telling me I am somebody to love despite my disease.

Recurring difficulties

I haven’t yet had serious health problems because of the PKD. I get tired more quickly than before, but I am still able to work. I expect eventually to develop serious problems and need renal dialysis and transplantation. I don’t often think about my future and remain hopeful about it, but sometimes I also feel sad.

One difficult situation is my annual visit to the hospital. My body tells me a story but I don’t know whether it is the true story—the one told by the blood values. I know there are people who suddenly become more ill and have to start dialysis far earlier than expected. Of course there are also people who do better than expected, but at the annual hospital visit my fears are stronger than my hopes. So every year there is tension and fear when I am told my blood values.

I sometimes feel sad and angry when people ask: “When are you going to have children?” The medical risks aren’t that great, and there are some new options like genetic selection, but for me this isn’t an option so the risk of heredity remains. We decided to give up our wish for children. People have tried to convince me of their
own opinion, rather than respecting our choice. Also, people say: “Why are you upset, in the future there surely will be new technologies which will cure you” or “Everybody has problems, perhaps you will be fine.” They are trying to help, but it has the opposite effect, making me feel misunderstood and miserable. Accepting my feelings and just being there helps far more than offering “good advice.” On my sad days, I find it difficult to understand why my boyfriend would want to stay with me—he would be better off when finding a new, healthy girlfriend. He helps me most by giving me a big hug and not saying anything at all at these moments.

<table>
<thead>
<tr>
<th>Helping hands along the way</th>
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<td>• Support from friends and family.</td>
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<td>• Contact with fellow sufferers—both for empathy and for advice.</td>
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<td>• My specialist in internal medical and my neurologist took me seriously and are respectful, straight, open, concrete, and understandable, taking time and being stringent or sensitive, depending on what I need at the moment.</td>
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<td>• The appreciation of my experience of the illness in my temporary job as research partner; I can relate to other patients with the condition, and thanks to the disease I’m now a PhD student.</td>
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Gradually I have learnt that “putting the illness away and getting on with life” doesn’t help. In my research job I can talk to many fellow kidney patients and have developed a much more realistic picture of living with a kidney disease. Talking to fellow patients and relating their stories to mine has changed my perspective; I have become more aware of the negative and positive aspects of the disease. I’ve stopped trying to change irreversible things, and this makes me happier.

I used to tell people "I seem to have a kidney disease" or “They have told me I have a kidney disease.” The disease wasn’t part of me; I tried to keep it at a distance. It was an enemy, something negative, not a part of me that I had to accept and cope with. With the passing of time, the disease has become a part of me. Through my research, my self-esteem has grown and I use the knowledge gained through my experience within academia to help fellow patients.
A doctor's perspective

Autosomal dominant polycystic kidney disease (ADPKD) is a common cause of renal failure, occurring in one person in every 500. Progressive renal damage leads to renal failure, at which point dialysis treatment or transplantation is inevitable. In families with a mutation in the gene coding for the protein polycystin-1, localised on chromosome 16, renal failure occurs at about age 50. In families with a mutation for the gene coding for polycystin-2 on chromosome 4, the disease may have a more lingering course.

Hypertension is common in ADPKD. Indeed, antihypertensive agents are currently the only accepted treatment. Other symptoms such as flank pain or fever are related to bleeding or infections of the renal cysts. The many extrarenal manifestations of the disease include early and severe diverticular disease of the colon, mitral valve prolapse, and intracranial aneurysms. The aneurysms, which occur in a subset of families, may lead to cerebral haemorrhage.

Until the 1960s, patients with ADPKD died at a young age because of uraemia or a ruptured intracranial aneurysm. Since then, renal replacement treatment has improved markedly: haemodialysis is much more bearable than it was 30 years ago and the results of (preferably pre-emptive) kidney transplantation have improved greatly. Furthermore, prophylactic, nearly non-invasive coagulation of the aneurysms has become feasible.

As ADPKD is an inherited disease, most patients are conscious of its course and complications in their affected relatives. ADPKD is an autosomal dominant disease, so offspring having a 50% chance of being affected. Though the disease can be confirmed at any age by means of DNA analysis, the diagnosis is generally made by ultrasound in the late teens or early adulthood.

Subsequently, many anxious years follow, even though young adult patients usually have no symptoms at all and need no drugs. Slowly, renal function deteriorates, drugs become necessary to treat hypertension and the progressive abnormalities in calcium-phosphate metabolism. A stage of life with haemodialysis or (pre-emptive) renal transplantation looms for ADPKD patients, more so than for other patients.
with progressive renal failure, who have not witnessed these dramatic events in their affected relatives when they were still children. Consequently most ADPKD patients are worried about their future. Many feel as if they are in a long tunnel, with renal replacement treatment waiting behind a closed door at its end.

**What can medical professionals do?**

- Provide information and support for the patient and the whole family, including partner and children, starting from the onset of the disease.
- Information includes medical knowledge about the disease, but also practical knowledge of how to deal with the disease in daily life. Fellow patients are particularly helpful to share creative solutions for daily problems.
- Medical specialists can provide an automatic referral to an association for renal patients to meet fellow patients.
- Support also entails emotional support for accepting the disease and giving it a place in one’s life. Listening to the patient’s story is important to help them find meaning again, and to revalue their life with the disease.
- Information and support also covers sensitive topics such as the desire to have children in relation to hereditary diseases.
- Taking the patient seriously (concerns, fears, ideas), creating enough time, and a respectful and open attitude is helpful for patients.
- Supporting the wish of a patient to remain active in society and life; letting the patient evaluate the risks.

What are the perspectives in the 21st century? The role of angiotensin converting enzyme inhibitors is still unclear. The HALT-PKD study is designed to test whether ACE inhibitors are superior to other antihypertensive agents in slowing down deterioration of renal function, but its results are still pending. Fundamental research indicates that cyst formation is caused by ciliary dysfunction, followed by accelerated proliferation and dedifferentiation of the epithelium. Growth of the cysts is related to deterioration of renal function. This process may be slowed down by drugs inhibiting fluid and ion transport into the cysts (vasopressin receptor antagonists and basolateral K⁺ channel inhibitors). Another approach is inhibiting cell proliferation, for instance with rapamycin. Finally, gene therapy may correct
inborn errors of metabolism. Studies are underway, but adverse effects of these potent drugs are inevitable. Because they influence different mechanisms, combining some of these agents may effectively slow progression, while allowing dose reduction and hence the incidence of side-effects. So, there may be a bright light at the end of the tunnel: ADPKD could one day become a treatable disease.

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