Personal Paper

A rare type

ELINOR GRAHAM

“You just can’t do that, Elinor!” said my husband. His illness had just been diagnosed at King’s College Hospital liver unit. I had suggested to Alfred that we should make arrangements for his tissues to be offered after his death to researchers into his rare metabolic disorder. “We know the names of people who have written articles,” I had said. “Let’s write to them and get the post mortem organised with a ‘shopping list’ of everyone’s requirements.” Alfred disagreed: “You can’t mention post mortems to doctors who are striving to keep me alive. It would be an insult. But if I die away from King’s, find out whether they would like a post mortem.” And that is what I did... and more.

A general practitioner’s hunch

The story began during a holiday in France in 1983. Alfred, normally so even tempered and placid, became strangely irritable. He said his urine was dark and his stools pale. I attributed that to the rich food and wine of Provence and the dry heat. I knew nothing of hepatology. Back at home he was ill for a few days with a flu like illness and nosebleeds. Our general practitioner arranged for a blood test, followed by another one: the first might have been affected by the illness. Then, vampire like, he wanted a third. Alfred asked no questions; he did not believe in harassing people. “Ask what is wrong with you,” I urged. He was told it could be his liver. An ordinary general practitioner had made an almost instant diagnosis without any sophisticated tests, but we did not know that then.

Alfred had been remarkably fit for 55 years, but he soon became familiar with the local hospital as test after test was arranged. The doctors did not tell him what they were doing, and again he asked no questions. “They will tell me what is wrong when they have found out,” he said. A year passed before we really knew.

Alfred made light of his illness and hid his discomfort. His bile ducts were under suspicion. Radiologists were getting frustrated because they could not get a clear ultrasonographic scan of his liver. Alfred laughingly told the tale of the barium enema: the quick frog march with a nurse to the nearest, but seemingly so distant, toilet; they made it, but only just. Then there was the morning when they injected him with iodine for an investigation. Later that day he chaired a management meeting (he was assistant comptroller of accounts in the Department of the Environment) and told his colleagues not to worry if he looked odd. Another colleague walked in and exclaimed, “What is wrong with you?” Those at the meeting had obediently said nothing as Alfred turned beetroot red. In the casualty department he had an injection for the reaction. They kept him for an hour in case there was a reaction to the injection to stop the reaction to the first injection: it was beginning to get complicated.

Alfred then developed an umbilical hernia. He underwent a laparotomy and hernia repair just before Christmas 1983, which he spent in hospital. Within an hour after discharge he began to drain fluid where the operation drain had been. That was when our troubles began in earnest. Two days later he was back in hospital “just over the weekend.” With his right lung congested and his right ankle swelling alarmingly he spent the New Year in hospital. He emerged blue and gasping from his first bath after the stitches had been removed. With oxygen he soon recovered, but I had never seen him so ill. We had been given scant information about his operation. An overworked house surgeon had borne the brunt of bank holiday crises. The regular consultant was on holiday. There was no registrar directly responsible in post. The house surgeon told me that I was not to worry, which was very worrying. At that point a senior registrar who specialised in the chest appeared: he reappears later in this story. He answered our questions, showed us the x ray films, and explained that Alfred had a pleural effusion. Three litres of fluid were aspirated from his right lung in the next two days. The house surgeon talked of another operation and an “ERCP.”

Clearly, someone had to find out more. I began to read the Oxford Textbook of Medicine in the local library. I learnt the terms for my husband’s symptoms. I realised that questions to doctors had to be specific. From that moment on no one would inform me I would inform myself. I began to brief Alfred on the questions to be asked. It was the beginning of his quest to find out what was going on in his body. It was the beginning for me of the realisation that medical knowledge is not the exclusive preserve of doctors. And that led, later, to doctors gaining more knowledge.

They then transferred Alfred to medical treatment at a different hospital. He left the strict atmosphere of the surgical ward and entered a new environment, grim and old from the outside but inside full of tender, loving kindness with few restrictions. He spent a happy month there while they made him reasonably well again. I even took him out one sunny day. The consultant broke away from his retinue to talk to me alone in the day room, giving the impression that he had all the time in the world, though I knew he had not. My husband had cirrhosis, possibly from an infection. The liver had great capacity to regenerate. Alfred would be able to go back to work. They would continue to keep an eye on him.

Alfred had more tests—a radiosotope scan and further ultrasonography. His bile ducts appeared to be clear, but the cholestatic symptoms persisted. Three attempts at liver biopsy in the next six months failed because of ascites. His condition was fairly stable, and he soon returned to work. Diuretics controlled the fluid, which seemed to come in cycles of four to six weeks. Weighing him daily, we adjusted the dose to the amount needed to keep his weight steady, and it worked most of the time. He was on a diet of no added salt with minimal amounts of salt in cooked food, and he enjoyed his meals. He tired easily and was unwontedly irritable when his weight was down. Although he was uncomfortable when the ascites was worse, he was his usual cheerful self.

No better

He was not, however, getting any better. At that stage I began to read about liver diseases: a difficult task for a layman. I was trying to understand abnormal processes with no knowledge of the normal. Continual references to a medical dictionary were necessary at first, but as I acquired the specialised vocabulary I speeded up. Alfred wanted to plan for the future. In financial affairs this was, of course, his daily work. For his personal life he needed to know the likely duration of his disease. He tried, without success, to find out what his expectation of life was. The doctors seemed reluctant to discuss it. If they would not help him, I would have to. I had one great advantage: as a graduate of London University I had access to all its libraries, including those of the medical schools. In the following months I read more and more. Sherlock was my bedtime reading. Later I borrowed Schiff too, although it was much too heavy to read in bed.

In summer 1984 we needed a holiday. We planned to drive slowly across France and have rail “runabout” tickets for a week in Switzerland, coming back through France again. I dipped into Sherlock and Schiff and wondered about the risks. I confirmed with our general practitioner that the outlook was indeed serious, but he told me to go on holiday. So we went and were
glad we did. Alfred’s ascites worsened with the well salted food and the heat, and drowsiness became a problem. We went on the famed Gornegrat cog railway among a polyglot company of excited tourists. On the way down, when everyone was gazing at the Matterhorn, Alfred slept soundly. I could sense the incredulity of our fellow passengers but I gazed at the view as though it were quite normal for my husband to sleep on a trip worth three stars in the Michelin guide.

Soon after our holiday Alfred became breathless. Over a litre of fluid was removed from his right lung. Then he developed a flu like infection, which subsided with antibiotics. He asked to go back to work before his certificate for sickness expired. I went with him to outpatients, where the registrar mentioned two possible causes for his cirrhosis: a rare enzyme disorder or non-A, non-B hepatitis. It was a difficult conversation for a pneumonic drip outside the open window drown the replies to my questions. Alfred was waiting, almost stripped, in the examination room. “What enzyme disorder?” I asked. “Alpha Pi ZZ.” The registrar replied, “Alpha what?” I repeated. “Alpha, anti bzzzzzzzzzz”. In the intervals between the noise of that wretched pneumonic drip I understood that it was alpha anti something-beginning-with-t. I wrote it down quickly while the registrar examined Alfred. I had been there somewhere in Sherlock. And so back home and to Sherlock. Yes, there it was, alpha antitrypsin deficiency. I looked in other books but no much information was available. Anyway, they only thought it might be that.

Alfred soon had another infection and became disorientated with rapid, laboured breathing. Antibiotics worked, but alpha antitrypsin deficiency remained. We got used to it, and after the medical schools in Hastings. Secondly, I was used to making contact and organising as I had once worked for cabinet ministers. Thirdly, I was dealing with a small, discrete subject. Fourthly, the delay between the death and the signing of the postmortem consent enabled me to collect my thoughts and control my grief, and, fifthly, I knew that I was carrying out Alfred’s wishes. This kind of thing should happen more often. It is up to the medical profession to ensure that it does. Why not establish an organisation to do what I did when postmortem material has value for research? What about donor cards for people of interesting rare types, with lists of doctors working on such cases, researchers who would like material, the tissues that are of interest, and whom to tell before the body is disposed of? It could possibly be done by the United Kingdom Transplant Service. And the patients? Just tell them how interesting and valuable they are.

I should be particularly interested to hear from anyone doing research into alpha antitrypsin deficiency.

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Postscript

I was able to make Alfred’s rare tissues available to members of the medical profession for several reasons. Firstly, I had access to the knowledge that had been built up over many years: I had spent many hours in the libraries of the medical schools at King’s and University Colleges, London, and I had much help from librarians in Hastings. Secondly, I was used to making contact and organising as I had once worked for cabinet ministers. Thirdly, I was dealing with a small, discrete subject. Fourthly, the delay between the death and the signing of the postmortem consent enabled me to collect my thoughts and control my grief, and, fifthly, I knew that I was carrying out Alfred’s wishes. This kind of thing should happen more often. It is up to the medical profession to ensure that it does. Why not establish an organisation to do what I did when postmortem material has value for research? What about donor cards for people of interesting rare types, with lists of doctors working on such cases, researchers who would like material, the tissues that are of interest, and whom to tell before the body is disposed of? It could possibly be done by the United Kingdom Transplant Service. And the patients? Just tell them how interesting and valuable they are.

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What physical exercise could be advised for a 50 year old non-smoking man with a right four rib thoracoplasty done about 30 years ago?

Such a man should, like any other man of his age, exercise daily. That exercise, according to the weather conditions of the day, should be enough to get him almost breathless from time to time while he is taking it. Fortunately, he does not smoke cigarettes. Should he be able to swim, then swimming with vigorous walking is certainly the best exercise that he can take. Even if there is some slight deformity because of the thoracoplasty, it would now, perhaps, be too late to correct this. It is assumed that his weight is normal, otherwise this should be corrected.—L. H. CAPL, consultant physician, London.