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A PATIENT'S JOURNEY

Acromegaly

Jon Danzig

Jon Danzig patient, Hemel Hempstead
jondanzig@aol.com

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Acromegaly may be a rare condition and difficult to diagnose. This patient's story highlights the importance of inquisitive questioning in cases with no obvious diagnosis

Something strange started happening to me around 10 years ago. My shoe size grew from 9 to 11, my income permanently dried up, and the girlfriend I was going to marry left after we stopped having sex. It wasn't just the sex, or lack of it. I also dramatically changed, both personally and physically. My nose grew bigger, my forehead enlarged, and my face and hands became puffy and coarse. But these were all gradual changes that I only realised in retrospect. Some friends noticed more acutely. One, who hadn't seen me for four years, looked shocked when I opened my front door. "You've rearranged your face," he blurted. My younger sister was blunter. She said I looked like I'd been in a fight.

I went from being dynamic and creative to someone who was apathetic and exhausted. My moods became widely variable. I was more anxious and easily fatigued. Inexplicably, my voice grew deeper. On many nights I had weird, out of body experiences. I woke up sweating, scared, and gasping.

I had been a successful freelance journalist and film maker, but things began to go wrong. Increasingly I went to the office late or not at all. Frequently I did little or nothing at work. Then I'd go home and do nothing there. Often I fell asleep at inappropriate times—at my desk or in the middle of having a cup of tea. Sometimes I tried hard to snap out of it by starting a grand new project, but it didn't work. I soon ran out of steam and mourned the loss of my former vibrant self.

From 1997 to the summer of 2001, I saw all manner of medical experts. My general practitioner collected almost 100 pages of notes about me. The doctors found nothing fundamentally wrong. I came to think it was me. I must have just "lost it." My general practitioner sent me to the practice counsellor. I told him I was worried that I had stopped being creative. He replied that I'd got to an age when I couldn't expect to be creative any more. I was only 42.

An opportunity to discover what was wrong came and passed. A small lump rapidly grew in my right breast—gynaecomastia. "Stop taking vitamin pills," said one consultant. "Have surgery," said another. Neither doctor, though, arranged hormone tests to determine the cause. A fortnight before surgery, the lump disappeared. Later it reappeared in my left breast. Ignore it, advised a general practitioner.

My dermatologist missed the diagnosis by not discovering the true cause of chronic mountainous acne on my back that was extremely painful. My orthopaedic doctor lost an opportunity by not finding the reason for joint pains. My dentist missed a chance by not recognising the widening gaps in my lower front teeth and problems with my lower jaw overbite. The doctors in my general practice failed by not understanding the combination of all my complaints, including depression, apathy, and snoring. When my testosterone was discovered to be substantially below normal, one general practitioner told me to come back in 12 months.

So I struggled for years, undiagnosed and with symptoms that stopped me from earning a living. Even when at last I was referred to an endocrinologist, he was also unable to make a timely diagnosis. It was only by becoming a more informed patient that I eventually added up the sum of all my symptoms and spelt out acromegaly. My suspicion sent me into a deep shock.

My endocrinologist failed to confirm a diagnosis of acromegaly in the six months I was his patient. Instead, he proposed a "non-functioning tumour." Only belatedly was I sent for an oral glucose tolerance test, the standard marker for acromegaly. As the endocrinologist then went on holiday, I asked my general practitioner's receptionist to obtain the results for me but was met with reluctance. I became exasperated and said, "This could be a life threatening illness."

A general practitioner rang back: "Your growth hormone levels are completely normal, and you do not have any insufficiency. This is hardly a life threatening illness." I remember feeling disappointed. Maybe my suspicion was completely wrong. "I thought my growth hormone level might have been too high," I said. I was sweating and my heart was beating fast. "Well," said the doctor, "as part of the test, you had taken a growth hormone accelerant, so I expect the

This is one of a series of occasional articles by patients about their experience of traumatic medical events that offer lessons to doctors. The *BMJ* welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance

Doctor's perspective

Jon Danzig has been one of the most challenging patients in my 32 years of general practice. I was initially frustrated, and probably irritated, with his frequent attendances for what seemed at that time to be a disparate collection of symptoms. I never seemed to be able to find any treatable cause for any of them, neither could any of the various specialists to whom I referred him. I reserved a drawer in my consulting room just for the paperwork that he generated—both from his own researches and the endless stream of hospital letters.

After the penny finally dropped and acromegaly was diagnosed, Jon said: "If only you had asked me if my shoe size had changed." Well, of course we can all be wiser after the event, and because I saw him so regularly, I failed to notice the asymmetrical enlarging of his jaw—unlike his friend who saw him for the first time in years.

I have come to respect Jon's quiet persistence in pursuing his quest for a diagnosis. Even since the diagnosis, he has had to challenge the rigidly held views of experts, getting them, as well, to listen to his observations and the results of his own researches. He has taught me the value of listening to my "persistent" patients more carefully, not just of giving a weary sigh on seeing their familiar name on my appointment list with the consequent shutting off of my receptive faculties.

Stephen Cohen, senior partner at The Nap Surgery, Kings Langley WD4 8ET Steve.Cohen@gp-e82129.nhs.uk

levels would be a bit higher." "No," I replied, "the glucose I took should have suppressed growth hormone if my pituitary had been functioning normally."

"Hold on a minute," said the doctor. "I'll check the test results again." She returned to tell me my growth hormone levels. "That's high above normal," I exclaimed. "Surely it means I have a condition known as acromegaly?" "I'm afraid you're now talking beyond my depth," admitted the doctor. "I think it's best you wait for your endocrinologist to come back from holiday."

I didn't want to wait. Instead, I wrote to an acromegaly expert, Professor John Monson, at St Bartholomew's Hospital, London. I enclosed my test results and told him I had acromegaly. The next day he phoned to ask, "Who made the diagnosis of acromegaly?" I paused. "I did," I said, somewhat hesitantly, waiting for his rebuke. "Well," said the professor, "you're absolutely right." When we met a few days later, in August 2001, Professor Monson said he could tell immediately that I had acromegaly just by looking at me. The test results confirmed that my growth hormone levels were around 2500% above normal. There was no doubt I had acromegaly.

The diagnosis at last provided an explanation for all my strange and incapacitating symptoms, which was a huge relief. I also felt let down. Surely any one of my doctors should have been able to recognise what was wrong with me much earlier? Apparently, the typical delay in diagnosis for acromegaly is 10 years or more. Isn't this a failure of medicine? The challenge for doctors, and patients, is to catch the illness early, years before the extensive damage and deformities occur.

Naturally, I wanted a cure. Yet I knew that with a large tumour my chances were low. The position of the tumour also caused concern as it was dangerously close to the carotid artery. This made me determined to find the most experienced neurosurgeon. That led me to Professor Rudolf Fahlbusch, the then director of neurosurgery at the University of Erlangen-Nuremberg in Germany. He had performed over 4000 pituitary operations, probably more than anyone else in the world. I was right to seek the best. I needed two operations, and afterwards Professor Fahlbusch wrote that the transsphenoidal surgery on me was "one of the most difficult and risky of my surgical life."

After the operations my face started going back to normal and my testosterone levels increased. Later, prolactin and thyroid levels returned to normal. My sleep improved and the out of body experiences stopped. The cystic acne on my back disappeared. Most importantly, the growth hormone levels also went down considerably but, alas, not enough. Blood tests showed I still had acromegaly.

Eventually, I sought the help of a leading acromegaly expert, Professor John Wass at the Churchill Hospital in Oxford. He fought hard for me to be treated on the NHS with octreotide (Sandostatin Lar, Novartis) injections, costing almost £1000 a shot, which I now receive every six weeks. My growth hormone levels are now at last in the normal range.

I feel I've made considerable progress. Unfortunately, though, I am not yet back to my work. Although I have my good days, I still get easily and unpredictably fatigued, a typical symptom for acromegaly patients, even after treatment. After my second operation I also developed a deep and debilitating headache in the centre of my forehead that, five years later, still visits me most days. Joint pains have also recently required surgery to both knees. Clearly the years of untreated acromegaly did some damage, not all of which can be undone. Nevertheless, I'm an optimist and remain hopeful that I may eventually be able to resume my career and life's plans in general.

My relationship is greatly improved with my general practitioner, Stephen Cohen. We've been on a long journey together and learnt considerably from it. From a difficult start, ours is now a true doctor-patient partnership. Dr Cohen has become my strongest ally through the medical maze and has helped to restore my confidence in the profession. Recently he told me, "I've come to the conclusion that any doctor who's not prepared to learn may as well retire." All the doctors at the practice have met to try and learn from my case. That, I applaud.

RESOURCES FOR PATIENTS

Pituitary Network Association (www.acromegaly.org)
Pituitary Foundation (www.pituitary.org.uk)