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PRACTICE

A PATIENT'S JOURNEY

Gitelman syndrome

Peter Park describes the not always easy interaction between the informed patient with a rare disorder and the medical profession, which he calls "a critical quadripartite relationship"

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

> General practitioner: "No, you cannot possibly take that quantity of magnesium, and besides, magnesium glycerophosphate is not on the formulary on my NHS computer."
>
> Me: "But Doctor, as a Gitelman syndrome sufferer, that is, in fact, the dosage and formulation prescribed by the consultant nephrologist, and, in actual fact, Doctor, you will need to hand write a prescription for this drug to be specially ordered and manufactured."
>
> GP: "Gitel what? …"

The above exchange is one variant on how I, as a fully diagnosed Gitelman syndrome patient already under the care of a nephrologist, began my relationship with each of the five general practitioners in the four surgeries I have been assigned to as I have moved around the UK. Despite early frustration, with a little patience and education by myself and the occasional "enlightened guidance" from the consultant nephrologist, each of these GPs save one (who was inexplicably reluctant to contact the nephrologist for advice) came to be a full participant on the proactive team that keeps me on track today, adjusting drugs over time.

A journey that began in the 1950s

When I was 10 years old, lack of energy and what I now know was tetany were the initial warning signs that, along with low potassium levels, began my medical journey. It quickly led to Dr C Everett Koop, a pioneering children's surgeon, and Dr Wallace McCrory, a paediatric nephrologist, at the Children's Hospital of Philadelphia in my native Pennsylvania. In the absence of today's technology, the tentative diagnosis was a probable tumour on the kidneys or adrenal glands, and an exploratory laparotomy was performed, with negative results. I went on a course of (then) poorly coated and foul tasting potassium chloride tablets, and Dr Koop went on a medicopolitical path to become Surgeon General of the United States (1982–89).

Managed for several years solely with large doses of potassium chloride, I often felt lethargic, but my condition began to come into sharper focus shortly after 1962, when Dr Frederic Bartter described the syndrome that bears his name. The addition to my drugs list of Aldactone (spironolactone) to regulate potassium wastage was an immediate change, followed later by the addition of magnesium to the mix by successive endocrinologists. I grew up, married, and my wife and I had a child (who, now aged 29, exhibits none of the signs of Gitelman syndrome). I ran, biked, and sailed in my 20s and 30s, albeit never with the stamina of my peers, and I generally felt OK.

I always thought I had Bartter syndrome, and it was not until I nearly died in 2005, after a severe stomach bug that caused kidney failure and, paradoxically, a dangerously high potassium level, that I was referred to a specialist clinic at Addenbrooke's Hospital, Cambridge. There, the true Gitelman syndrome variant was diagnosed through genetics, and a comprehensive and well monitored treatment regimen began. Since then, I have twice had knee replacement surgery without incident (though I suspect I had to have a lot more blood tests than most patients), and I feel well most of the time, except when I miss a dose of a key drug such as potassium chloride for eight or more hours.

GP meets informed patient with a rare condition

When someone such as myself first presents to a doctor for a related, or unrelated, ailment, either at a surgery or at a hospital, and claims to be a Gitelman or Bartter syndrome patient, several possible reactions can be expected. The most likely one is a vague recollection by the doctor of the syndrome name, perhaps

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from a single lecture during medical training years ago, but no further immediate knowledge.

Another reaction (this one a bit tedious for the patient) is the doctor whose face tells you that, as a "walking medical curiosity," you have just brightened up an otherwise boring day spent treating ingrowing toenails and hard to diagnose itches. You just know this doctor will be late for the next patient as he or she fairly leaps for the computer to access Wikipedia the second you leave the treatment room.

A third reaction is the (thankfully quite rare these days) physician who believes that an "informed patient" is an oxymoron and is thus inclined to dismiss your description of the condition and its treatment unless and until it is confirmed by a consultant medical professional in the subject. This reaction is especially likely for juvenile and young adult patients.

The good news is that almost certainly each of these reactions by a GP can be guided with a little effort toward the fully supportive role that is essential for optimal care of a rare and sometimes unpredictable condition. It is important to note that, at core, I think that is what our GPs want as well.

Equally important is for us, as Gitelman/Bartter patients, to understand that, although the consultant nephrologist may be the pinnacle of knowledge on the disease and in the best position to guide GPs and ourselves, he or she simply cannot be called on by patient or GP for every small event in our treatment. As informed patients, we need to be able to take responsibility, shared with our GP and coordinated with the pharmacist, for the day to day management of the syndrome, and of syndrome related aspects of other ailments.

While the relationship with the consultant nephrologist (or endocrinologist) is based on far more shared knowledge and therefore much easier, it too needs to be managed for good outcomes. The patient presenting for perhaps a semi-annual appointment with the specialist can maximise the precious minutes of this encounter by such simple things as working with the GP to ensure recent blood work is available before the appointment, and writing down questions before the day, including reporting on experience, good or bad, with drugs and other healthcare professionals.

My pharmacist: a VIP

An often overlooked part of managing Gitelman or Bartter syndrome is the role of the pharmacist. This medical professional, who in the UK has completed not only a four year Master of Pharmacy degree but at least one extra year before registration, can be the linchpin in care, and a most welcome and informed advocate with the prescription clerk in the GP's surgery, and often, via the clerk, with the GP. I have also learnt that, though in a lesser role, the surgery prescription clerk is another person the patient will want to have "on side"—well worth a quick word when you are at the surgery.

In my case, a committed young pharmacist, after researching the drug, spent hours convincing the surgery that, by ordering magnesium glycerophosphate in quantities starting with 5000 capsules, amazing savings in NHS costs would follow for this specially formulated drug from the economies of a larger production run.

Any patients who fail to get to know their pharmacist and discuss their prescriptions with him or her, and any GP who fails to listen to this professional on drug formulations and interactions, are missing good information. Like GPs, pharmacists may initially question prescribed doses and medications (a fellow patient described this as the "policeman" role), but all will benefit from the time invested in education and discussion, as this medical professional becomes a member of your personal care team.

My life today

Although I live with a condition that most people have never heard of, and have had to become an "expert patient," I am pretty lucky: although the treatments I must take don't taste good and I almost need an extra bag when I travel just for them, they and my care team have meant that I have had enough energy to hold down a job and now look forward to retirement.

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A clinician's perspective

Me: "Thank you for completing the urine collections and blood tests after our first meeting, when your GP asked me to look into your low potassium. The results show that your potassium and magnesium levels are both still low, and there's too much of both in your urine. So I think we can put a name to the symptoms of tiredness, weakness, pins and needles, palpitations, cramps, nausea, and chest pain that have made your life a misery for a long time. I think you have Gitelman syndrome."

Patient: "I've never heard of that."

Me: "It's quite rare, but there are probably at least a thousand people with it in the UK. Tell me, when you were a child did you prefer sweets or crisps as treats?"

Patient: "Oh, always crisps. I used to eat jars of pickled cucumbers as well."

Me: "That's a very familiar response. Your body was telling you it needed salt. OK, let's start talking about how you are going to manage both your diet and the potassium and magnesium tablets you will need to take for the rest of your life."

This is a highly truncated summary of some essential features of Gitelman syndrome. As Mr Park describes, Gitelman syndrome and a similar disorder, type 3 Bartter syndrome, are rare, autosomal recessive renal tubular disorders that usually emerge after childhood and affect the kidneys' ability to conserve potassium or magnesium, or both. The usual scenario is of someone with rather non-specific symptoms, perhaps "off sick" from work a lot or reluctant to do sports at school, who is found to have a low blood potassium level and lowish (sometimes disablingly low) blood pressure. Sometimes, however, life threatening arrhythmia may be the presenting feature.

In most cases both potassium and magnesium levels are low. Treatment, with potassium and magnesium supplements, is at best unpalatable and at worst intolerable, mainly because the quantities required may be very large. There are additional options—including amiloride, spironolactone, angiotensin receptor blockers or ACE inhibitors, and β blockers—but use of these may be constrained by hypotension. Every patient's needs are different, so protocols cannot easily be prepared.

Among my patients, there is a wide variety of knowledge. Those like Mr Park, who was diagnosed in childhood, are, of course, the easiest. He has a pretty comprehensive understanding of his own pathophysiology and is able to self manage to the same extent that a well informed person with diabetes does. Newer or less able patients may rely much more on their specialists or GPs to interpret their potassium, magnesium, and blood pressure levels and advise them when they need to change treatment. We try to make sure that our patients' GPs are "in the know," but of course this must be balanced against their other commitments. This is particularly relevant for unlicensed treatments, of which magnesium supplementation in this context is a good example: there is almost nothing at all in the *British National Formulary*.

One thing unites my cohort of close to 50 patients with Gitelman or Bartter syndrome (apart from a general feeling of diagnostic isolation and their long term symptoms): most have been doubted by someone in a healthcare role at one time or another. A GP friend commented that she thinks that patients with this type of disorder (or their carers) should always be the ones in the driving seat—carrying with them paperwork on hospital headed notepaper that can provide authoritative confirmation that the treatment being proposed by the patient is valid, and also inform other healthcare staff. This seems like a good goal to work towards: above all, patient and family empowerment and good communication are key.

Fiona E Karet Frankl

Resources for patients and clinicians

The Bartter Site (http://barttersite.org/)—Provides information and support for Bartter and Gitelman syndromes

RareRenal.org. Hypokalaemic alkalosis (www.rarerenal.org/diseases/hypokalaemic-alkalosis/)—Provides information for patients and professionals, and a forthcoming opportunity to enrol in a national patient registry

Cambridge University Hospitals, Renal Genetics and Tubular Disorders Clinic. Information about Gitelman syndrome (www.cuh.org.uk/resources/pdf/patient_information_leaflets/PIN1446_gitelmans_information.pdf)

British Kidney Patient Association (BKPA) (www.britishkidney-pa.co.uk)—UK registered charity working to improve quality of life for patients with kidney disease. It provides information, advice, and small grants to help patients and families with kidney disease and financial support to kidney units throughout the UK