Living with a rare disease: A personal perspective

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doi: 10.56012/otyj6941

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Working in rare diseases especially, it is so important to just sit back and listen to the individuals who live with rare disease every day. As part of this edition of *Medical Writing*, guest editor Sarah Milner sat down with Richard Farquhar, who has a rare metabolic condition, phenylketonuria (PKU). He spoke about his life and his experiences and offered up some motivational works.

***Medical Writing* (MW):** So, Richard, can you just tell us a little about yourself?
Richard Farquhar (RF): Hi, it's lovely to speak to you. I'm currently in my early 40s, have a career in the higher education system in the UK, and I have 2 children who do not have the PKU condition.

I've been fortunate enough to have lived in the UK for my whole life, which has meant I've had access to PKU treatment and medical professionals. Over the past 5 years, I've taken a very focused approach to my PKU condition and overall health, fitness, and general well-being.

MW: For those who don't know, could you explain briefly what PKU is?
RF: PKU is a genetic condition, so it's something I've had to deal with my whole life. It's an inherited metabolic disorder in which an amino acid called phenylalanine (which is a building block of protein) builds up in the body and causes quite serious issues. When I eat foods with protein in them, which is pretty much nearly all food, phenylalanine enters the body. A person without PKU will be able to discard excess phenylalanine, but I cannot. Once phenylalanine increases to harmful levels it crosses the blood-

brain barrier and causes developmental issues and impacts the brain. Someone who is not treated will suffer intellectual disabilities and other serious health problems. There are different variants of the condition from mild to severe. Treatment is a lifelong, low-protein diet.

MW: Are there any medications you use to treat the disease?
RF: About a year ago, I started taking a drug called Sapropterin, which lowers the levels of phenylalanine in my blood. Since being on Sapropterin, I can tolerate 30 grams of protein from food daily. This still means I cannot eat meat, lentils, nuts, eggs, cheese, pastries, to name a few. Before Sapropterin I couldn't eat normal rice, or pastas. My tolerance for those foods is now quite good. But many PKU patients have very low tolerances, so I would guesstimate 80% of foods are not allowed on their PKU diet. The reliance then on medical-based food products is very high, and choice is very limited.

MW: Looking back, have you ever discussed with your parents how they felt when they found out their baby had PKU?
RF: My parents were very young when they had me, which I believe made it harder for them. I was also born in the early '80s, which was a very different time and still early in the treatment of the condition. They have told me they were terrified and fearful of the future. There were a lot of unknowns for them. They had to rely heavily on the medical professionals who were supporting people with the condition.

MW: As a follow-on, knowing what you know now, what advice would you give to someone who has just had a family member diagnosed with a rare disease?
RF: Firstly, don't panic. Time has moved on and medical science and knowledge about many condition has progressed. Also, there are a lot of people with my condition living amazing and full lives. It is a challenge, I will not lie, but I've lived a life full of family, friends, holidays, adventures, and fun. It's scary, but seek out up-to-date information, network with people with the condition to learn more, and access tailored care, if available to you. For me and my parents, the medical professionals I've had in my life have quite literally been a lifesaver.

MW: How do you feel having a rare disease has impacted your life (physically, mentally, socially)?
RF: There have been times in my life when it did impact it in a negative way. Food plays such an important part in how humans' bond; we socialise, and food plays an important part in that bonding process. How many times, when you've been with friends, is food part of the activity or process? How many times have you gone on holiday and wanted to experience the culture through food? I've had to find ways to adapt to so many situations, I've got better at that with practice and age, but in the early days, I missed out on some opportunities. Food science has progressed a long way in recent years, so this helps a lot. The biggest issues physically are that if I'm not tracking my food consumption well enough, and my phenylalanine levels are higher than they should be, I will experience headaches, brain fog, and a cognitive decline. It also impacts my overall mood and how I engage with the wider world. I'm at my best when I'm in control of my diet; controlling my phenylalanine levels is, for me, the foundation of everything.

MW: You are clearly a big advocate of exercise; do you feel like this helps you manage your condition physically or mentally?
RF: Yes. I think it has been one of the best decisions I've ever made. When a lot of people think of exercise, we just think it helps us get fitter and maybe stronger, but the effects go much deeper than that, and I'm living proof of this. Through focusing on exercise, I've become more mindful of my nutrition, my PKU condition, and my overall health and wellbeing. Exercise literally rewires our brain, for the better. It has a positive effect on our endorphins, which can cross the blood-brain barrier and make us feel better and happier. It's hard at the start to be motivated and to be consistent, but once you are, the positive effects are incredible. If medical science could take those positive effects and put all of them into a pill, not only would it have no side effects, but it would also be the best-selling pill ever created, forever. I've become psychically stronger and mentally stronger, it's impacted my life so much and is now starting to affect the lives of people around me (friends and family), and that's an amazing thing to see.

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MW: As your condition is essentially "invisible" to the outside world, I am interested to understand what, how, and when you tell people. Do you think that there is a stigma attached?
RF: When I was younger, I was not great at telling people, I would either avoid it or downplay it. I think that approach was not helpful. I'm quite open and honest about it now, with practice I've become better at explaining it too. I'm not ashamed of the condition, I have it, yes, but it's not who I am. I am much more than the disease. I will normally tell people when and if they ask, which can normally be around situations with food, which is all the time. Telling people is much easier now, there are a lot of food allergies that people throughout the world are faced with, though this is not a food allergy, it's much more severe than that, and it does make having the conversation easier.

MW: If this isn't too personal, did having a rare disease come into consideration when deciding to have children?
RF: Honestly, no. I knew that if my wife was not a carrier my children would not be born with the condition. Plus, it is a rare condition, so I treat it as such. I will not lie though, when both of my children were born, I was full of anxiety, especially when the heel prick blood test was taken. This is how they check newborn babies for the condition (and other conditions too). It was a very nerve-wracking time.

MW: I hear a lot that having a rare disease can be isolating. There is a lot of hate for social media, but I wonder if you see the positives of connecting with others in the community? Do you think it is useful for younger people too, I imagine in your teens, having a rare disease can be incredibly challenging socially.
RF: It can be very isolating yes. I spent 40 years of my life never meeting anyone face to face who had my condition. I messaged with a few people for years prior to that, thankfully because of social media platforms, but never met anyone until I went to my first PKU event in Birmingham, England. I've come to learn I get great strength from engaging with others with the condition, we can help, support, and understand each other, in a way that others can't. There is something very

healing about that. Social media gets a lot of bad attention, and rightly so, but I use social media to access the PKU community. I use it in ways to empower, to motivate me, I'm incredibly thankful for it and for the PKU community that engages with me. I think for everyone, no matter what their age, if they use it correctly, they can find people with the condition and they can potentially become lifelong friends. Having that type of support is amazing.

MW: Are there organisations that have helped you over the years and provided support and advice?
RF: Yes, the National Society for Phenylketonuria (UK NSPKU) is incredible. I didn't access the organisation for most of my adult life, and that was a mistake. The people who help run the organisation are amazing individuals, I have so much respect for them. The help and guidance they've given me and other people with PKU is incredible. I'm also thankful for medical professionals and organisations that create products, support research, and share information so that living with this condition can be a bit easier.

MW: OK, pivoting to a different topic and experiences with the healthcare system. Tell me more about the good and challenging experiences you have had with your care, and do you think this has changed over the years?
RF: Without access to the National Health Service (the UK's public health provider), I would not be the person I am today. I would not be living the life I'm living, and for that, I will be forever grateful. I know my parents are incredibly grateful for the support they had in those early years too. That doesn't mean everything is perfect though, there are always ways we can improve. The care across our country is equal in many ways, but not in many ways too. I'm aware there is a huge cost to treating this condition, for any healthcare provider, but the consequences of not, in my view, are much larger. I hope that across the world care for PKU becomes equal, it is needed. Also, there needs to be a more tailored approach to treating the PKU condition, and I hope to see more of this in the future. I have an amazing team of professionals around me now, and I am so thankful for having them in my life. There needs to be more access to products and the correct amount of supplements given to people who live with the condition, if these decisions are just made based on financial costs, the treatment will never be optimal, or even acceptable.

MW: Charities like Medics 4 Rare Disease do such important work educating medical students and doctors, but their work is limited by resources. There must be recognition of the patient/caregivers as the experts in their condition. Have you experienced this frustration where healthcare professionals (HCPs) don't listen to you?
RF: In the past yes. I've become very articulate and understand my body very well. I'm able to explain how and what I feel, but there have been times when this has not been listened too, or even interpreted as anxiety. Sometimes decisions made were purely financial, and I don't think that is putting the patient first. We live with the condition 24 hours a day, every day, and we have done for our whole life. In the past 5 years I've learned more about myself, the condition than I ever have. I'm proud of the learning I've gone through and how disciplined I've been, I've changed my life for the better, but there have been moments in the past where access to knowledge was limited, access to protein supplements were denied, and resources not good enough. I'm aware that medical staff have very limited time and resources themselves, and the pressures that they face to meet the requirements of all their patients very difficult. I'm thankful for all the treatments that I've had throughout my life, but treatment could be better for so many individuals, in the UK, and across the world. Thankfully, as I've said before, I have an amazing team of professionals around me now, and I'm so incredibly grateful for that.

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MW: If you could offer up advice to healthcare professionals when working with a patient with a rare disease, what would that be?
RF: Try to ask the right questions and take the time to listen to their response. Be honest about any resource limitations you might have when dealing with any issues they are experiencing but show empathy too. The best advice I would give, is maybe try living on the [PKU] diet for one week yourself. It might be considered an unreasonable request, but it can be difficult to know the struggles of an individual until you've somewhat experienced what they are going through.

MW: Recently, I have seen a real move in pharma to develop treatments for patients with rare disease, which is so positive for the community. A lot of that is due to the regulatory agencies being more flexible around requirements for approval. Do you find it frustrating that, on the face of it, there isn't equal access for patients, and companies don't do work in rare disease because it won't make them lots of money?
RF: Yes, I think about this a lot. There are so many treatments coming through now that will make a clear difference in the lives of many, but finances play the biggest part in people getting access to them, and that's such a shame. There is a new drug called Palynziq which has the ability to help PKU patients progress onto a completely normal life, but there are some potentially risky side effects and the costs are huge. So far, I believe this is only being used in some states in the US due to cost. A lot of focus has been on the short-term implications of not treating rare disease conditions, but there are long-term implications of living on a diet that relies so heavily on processed food, I'm not sure that is being considered enough. Science is proving that this has long-term impacts for individuals, this needs to be considered more too. Approving these should be considered a priority.

MW: I also think that the regulatory agencies are really encouraging pharma companies to design trials that assess how a drug improves quality of life rather than just a finite endpoint like survival, etc. Do you see that as positive?
RF: Yes, in my view this is a huge positive step forward. Rare diseases effect every part of our lives, so assessing how quality of life is improved is just as important. A lot of managing the PKU condition focusses around phenylalanine levels, but it should be much more than just that.

MW: There is also a real move, especially in regulatory writing, to use more inclusive language in our documents I think the industry must move on from seeing clinical study participants as "subjects to observe". I know people generally go on trials to improve their health, but I was blown away but the people who go on clinical trials just to help generate

information on their condition. I think that's amazing.

Tell me about patient advocacy, why it is so important generally, and to you and why you share your journey on social media. What is your aim?

RF: I have a few aims and goals, firstly, it gives me a platform to connect with others, to share and learn new knowledge focusing on living with a rare disease. Also, there is limited research on rare diseases, exercise, and overall health, well-being, mental health and ageing with this condition, I hope that by sharing my journey in this area it will inspire people to share their voice and story and encourage new ideas and research in these topics. I hope to bring more awareness to the challenges we are faced with as a community, but to also be a positive role model for others.

MW: Looking to the future, what do you hope for in the rare disease community?

RF: Equal care and access to life changing treatments, across the world.

MW: Tell me what you want people to know about having a rare disease and how we can help your mission?

RF: I think helping patients have platforms like this is useful, so thank you for the opportunity, more opportunities like this would be helpful too. I want people to know that living with this condition is challenging for so many. It can either make or break you. So many people in general, who live a life without dietary constraints like the PKU diet struggle to follow a normal health or weight-loss diet plan for any period. It's why the fitness industry and the weight-loss industry are so big. People struggle, fail, then try again. People with my condition do not have this option, we must do this for life. It's more than that though, this is people's lives that are affected, their overall health, including their brain health harmed. Equal access to treatment that can change this would be amazing, and is needed. Think about that for a moment. Think of the strength it takes to be on a diet that for some, eradicates 80% of food options, forever. If this doesn't highlight that long-term treatment options need to be considered, and equal access to them, I don't know what does.

MW: Thank you so much for joining me today, Richard, and sharing your story. I really hope that in the future, there's a lot more research within this area.

Disclaimers

The views and opinions expressed in this article are the interviewers' and interviewees' own and are not necessarily shared by any employer or by EMWA.

Author information

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International PKU Day

International PKU Day is on June 28, a day on which we spread the word about Phenylketonuria (PKU). It also celebrates the legacy of Dr Robert Guthrie (1916-1995), and Dr Horst Bickel (1918-2000), who developed screening and treatment for PKU.

June 28 is also the deadline for the European Society for Phenylketonuria (ES-PKU) Sheila Jones Award, dedicated to patient advocates and organisations working for people with PKU.

Nominations for the award can be submitted at: https://www.espku.org/sheila-jones-award/sheila-jones-award-submission/.

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