

Rare Disease

A Family's Experience



Introduction

Graphic novels offer a powerful way to communicate what it means to live with a rare condition. These conditions shape daily life in ways that clinical language alone struggles to capture. Although each rare disease is individually uncommon, together they affect millions of people worldwide. Many of the challenges they bring — limited and often complex treatment options, social misunderstanding, and the sense of being set apart — are shared across countless rare disease communities. What we present here reflects the realities of living with two rare conditions, Phenylketonuria (PKU) and Dihydropteridine Reductase Deficiency (DHPRD), but their experiences will feel familiar to many others affected by rare conditions.

Visual storytelling bridges the gap between clinical description and lived experience. It conveys the emotional texture of growing up with a rare disorder: the uncertainty surrounding diagnosis, the vigilance of dietary management, the strict medication routines, the relentless burden of care, the constant undercurrent of anxiety, the weight of stigma, the search for normality, and the resilience that individuals and families cultivate over time. By combining imagery with narrative, this set of graphic novels makes complex experiences accessible to readers of all ages and backgrounds. A single panel can communicate frustration, fear, or isolation far more immediately than paragraphs of text. For children with PKU and DHPR deficiency and their caregivers, this format validates feelings that are rarely acknowledged in medical settings. For clinicians, educators, and policymakers, it offers insight into the human realities behind biochemical targets and treatment protocols.

These stories invite readers to notice the moments that define everyday life with a rare condition, and to connect with the person behind the diagnosis. In doing so, they broaden understanding, challenge assumptions, and help build a more compassionate and informed community.

Anita MacDonald

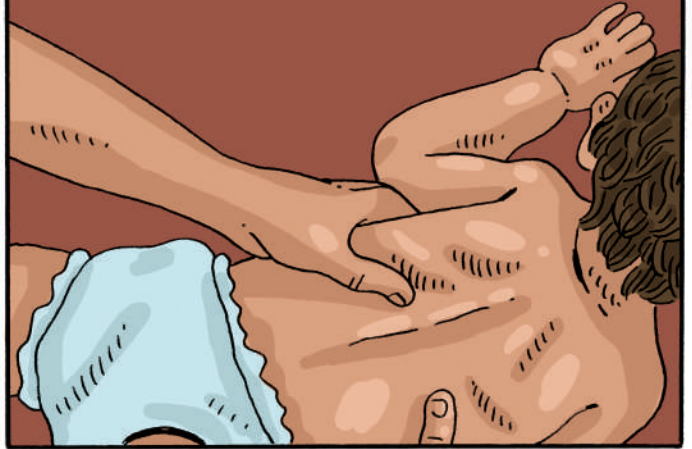
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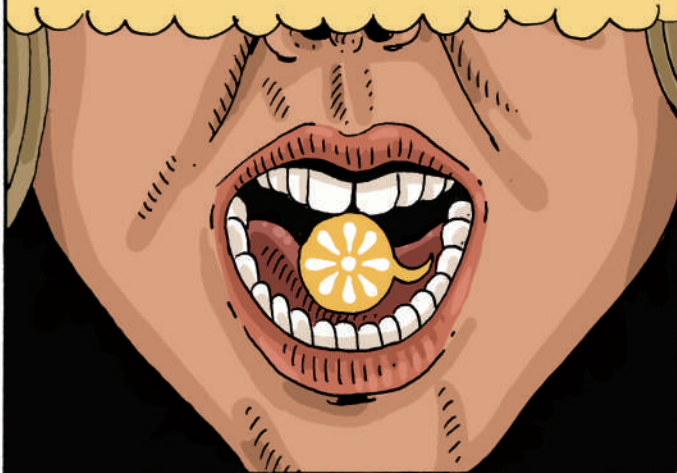
I was told...
"You have to come tomorrow to have a lumbar puncture done on your baby. We suspect that your son has a rare condition. DO NOT Google it."



He was too young to be given a general anaesthetic. So I had to hold him down while they put a needle into his back. I was crying, but I just kept thinking to myself *this is the only way to keep him safe.*



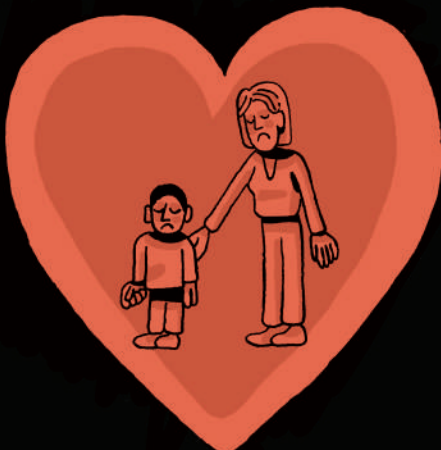
I don't have any recollection of what I heard in that consultation. You're just kind of traumatised. And you don't know how to articulate what's happening, because you're not sure yourself.



Appointments come with anxiety. Six years later he still has lumbar punctures to manage the condition; these are for life.

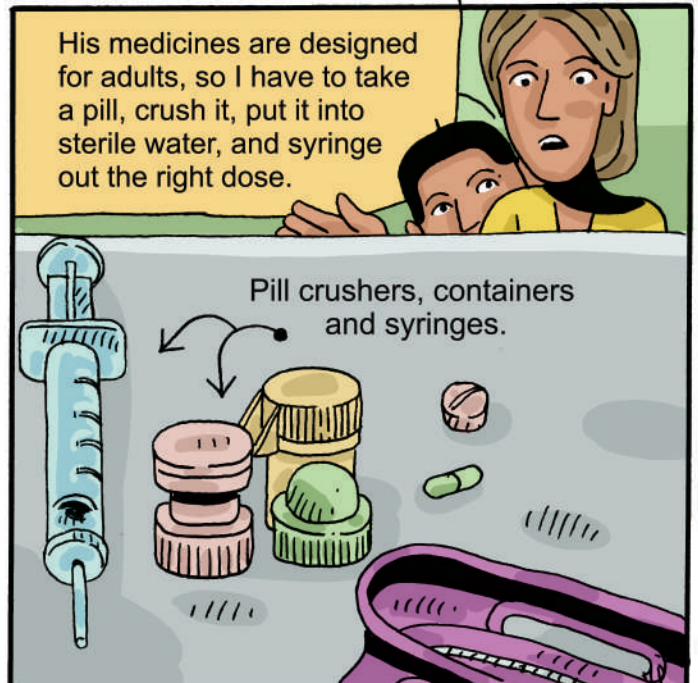
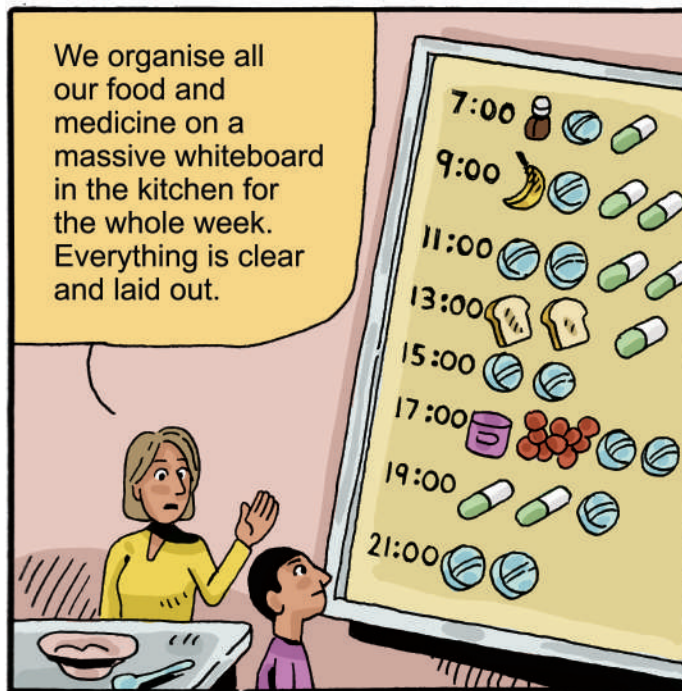


I tell myself the same thing each time, *this is what it takes to keep my child safe.* You have to go in there with full trust, because there is no alternative.



Life can be incredibly hard when you're living with a condition like this. You feel powerless.





We can get extra help but ultimately we are responsible for our son's health. We are the only constant forces in his life. Even though everyday cannot be perfect we still blame ourselves when things get missed or forgotten.



I had to quit my job. It's changed the way we live our lives.



Home is our safe space, which is why it's so important to get the daily routine and who has responsibility for what, right.



When you live with a rare disease it can be really tricky when you go out, but it's important to bring some normality into his life.



When he was three I took him for a babyccino and it became a Sunday tradition.

It has to be oat milk, it has to be this amount, and no chocolate powder please.



I'd make excuses saying he has an allergy, as I can't explain to a barista that he has a low protein diet. It felt lovely sitting and sipping our drinks together.



My son's condition requires complex treatments that relate to essential human needs, like food. This can affect participation in societal glues; things that help people connect and understand each other.



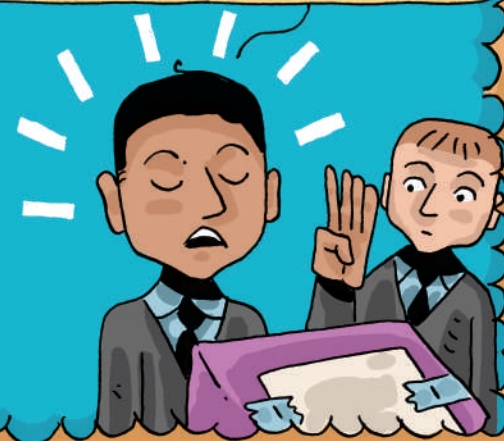
They make fun of me all the time about my food at school.



Why is there always a list on the top of your lunchbox? You're such a baby because your mum has to write what you have to eat.



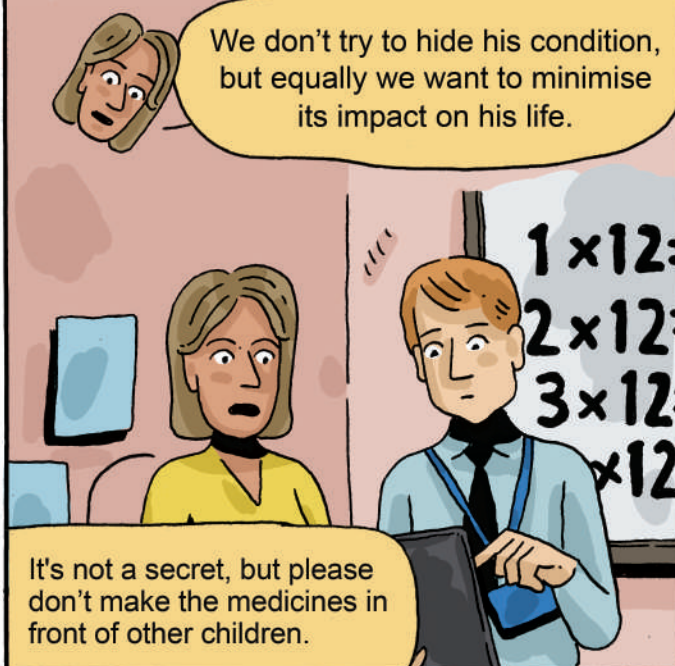
I need a list because I have to eat very specific amounts of protein. And anyway, I have to have lumbar punctures, and everyone tells me I'm really brave!



When I heard this I thought to myself, *I don't think you realise the gravity of what you have just shared; but good for you.*



We don't try to hide his condition, but equally we want to minimise its impact on his life.



Kids' parties are particularly anxiety inducing for him. He can become very worried before attending that others don't understand his needs.



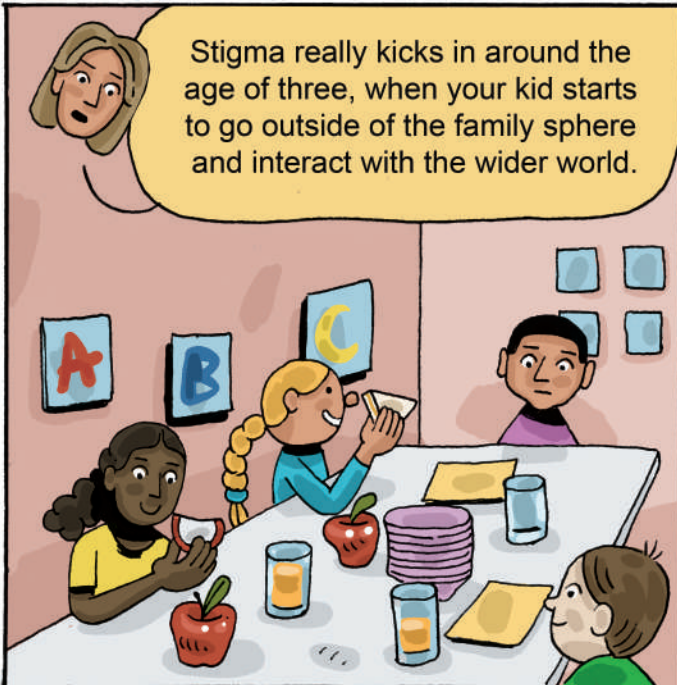
Will they know I can't have cake?

Due to his sensory issues he doesn't want to go to parties involving physical interaction, even if it's for his best friend. I worry he feels socially ostracised.



I can't keep up mum.

Stigma really kicks in around the age of three, when your kid starts to go outside of the family sphere and interact with the wider world.



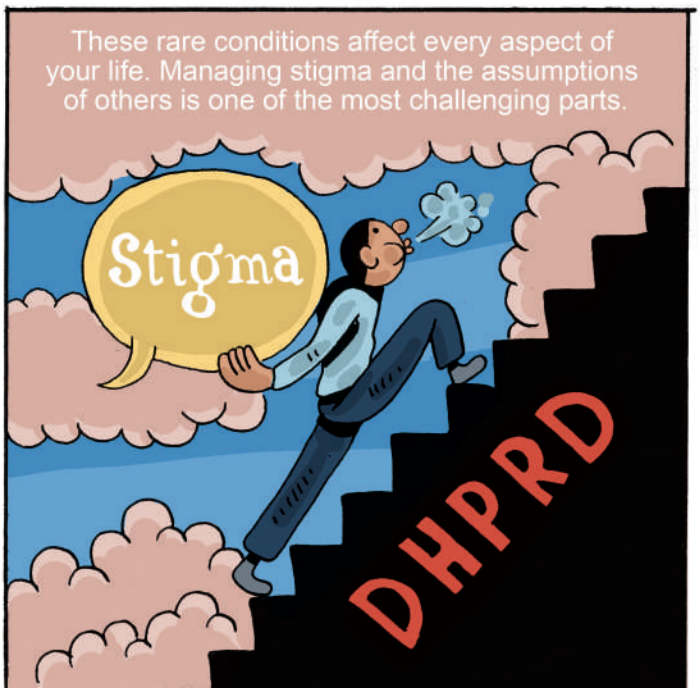
They don't feel different until then. And it doesn't get much easier as they get older.



I've heard of people telling their dates that they are vegan to avoid explaining their dietary restrictions. But when are they going to tell their date that it's a bit more serious than that!



These rare conditions affect every aspect of your life. Managing stigma and the assumptions of others is one of the most challenging parts.



Rare Disease - A Family's Experience

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