DENISE'S STORY:

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Denise
Scientist
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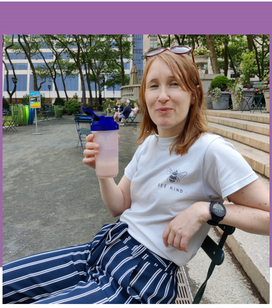

How did you come to work at Nutricia?

I currently work as Scientist Ketogenic Diet / Epilepsy within the Metabolics and Epilepsy. After finishing my master's degree, I decided I would like to do a PhD, but only on a topic close to my heart: PKU. I applied for several grants to set up my own PhD. I moved to the UK and lived in Leeds for four years whilst I undertook my research together with adults with PKU who struggled with adherence to their low protein diet. **As I (luckily) rarely struggled with the low protein diet myself, I was motivated to work with (and hopefully help) those that do.** After completing my PhD, I moved back to the Netherlands and started working in specialised nutrition at a dairy company, knowing that someday I would love to get back into the field of Metabolics.

When there was a job opening for a Clinical Study Researcher (CSR) Metabolics and Ketogenic Diet at Danone Nutricia, I applied straight away and got the job! In addition to my role as CSR, I supported Ketogenic Diet in the capacity of scientist. At some point, both functions were split. As PKU became more prominent in my life (see below) and I'm more of a scientist at heart, I moved to the scientist role, whilst staying close to the Metabolics colleagues in my team – best of both worlds if you ask me!

What is your experience living with PKU, your challenges and what gives you strength?

When I was born, I was diagnosed with mild hyperphenylalaninemia (HPA) and a low protein diet, with protein substitutes, was not needed at the time. At age 3, I was hospitalized with a fever and high phenylalanine levels. **After 3 years on a normal diet, they adjusted my diagnosis to phenylketonuria (PKU) and I had to start the low protein diet with protein substitutes.**

My first protein substitutes were cans of amino acid-based protein substitutes, which we mixed with tomato juice. Whenever I got fed up with my substitutes and tried different ones or different things to mix the powder with, I would always go back to the same combination.

As I reached my teenage years and protein substitutes developed a bit more, I switched to forest fruit-flavoured protein substitutes in sachets that could be made up into a drink. **During puberty, these would often disappear underneath my bed simply because I was too lazy to prepare them.** If they were mixed for me, I would drink them without issues. Once my mum finally got through to me regarding her worries of long-term effects of not sticking to my diet, I decided to adhere to the diet and never looked back.

In addition to having a relatively mild form of PKU I have been very lucky with my family, classmates and friends growing up. My parents never thought in challenges, but always in possibilities. They never shied away from big trips or eating out because it was "difficult", and I was never made to feel that PKU was a burden or something that should hold me back from doing what I wanted to do. **Parents from classmates would just consult mine before birthday parties (I would often bring my own pancake batter or something like that) instead of not inviting me because it was too difficult to take PKU into account.** I realise that this is not the experience of every individual with PKU and count myself very lucky.

When I was born, my responsiveness to BH4 (a co-factor to the enzyme that is affected in PKU) was already tested and about 20 years later, Kuvan (synthetic version of BH4) was approved for prescription in Europe. I responded so well that I was able to come off my low protein diet completely. This was a challenge at first: going from not eating too much natural protein (15 grams) to eating enough (~60 grams) took some getting used to! I think it took me about 2 years to fully get into the swing of eating enough natural protein!

After 15 years of being off the PKU diet, I returned to the low-protein diet because my partner and I had a wish to start a family and my metabolic clinic is hesitant about the use of Kuvan during pre-conception and pregnancy as there is little information about long-term effects on the offspring.

When I returned to the PKU diet, I opted for GMP-based protein substitutes, which taste better than the amino acid-based protein substitutes. I'm so glad that this option is now available, because after not having had any protein substitutes for 15 years, I'm not sure how amino acid-based ones would have gone down!

What is your dream for the future of PKU?

Let me start by saying that I think the knowledge, management, and treatment options for PKU have come a long way since it was first discovered. During the time I was off diet, the offering, convenience, and taste of protein substitutes as well as low protein foods has massively improved.

As I mentioned before, except for the occasional "off-day", I never really struggled with the low protein diet, although I recognize my life was definitely easier whilst on Kuvan. **I know many individuals with PKU and their families do struggle. Therefore, I hope things will continue to improve so that all individuals with PKU can live their best lives!**

In addition, there are still a lot of unknowns, such as the impact of PKU and available treatments on ageing. Again, hopefully with time and research, insights will improve, which - in turn - could support improvements in management / treatment options.