EPISODE 14: Prader-Willi Syndrome (PWS)

Transcript

I remember sitting in Medical Nutrition Therapy class learning about so many different conditions but one of them stood out above most of the others. I was truly fascinated by this condition. I couldn't imagine never feeling full. To eat and eat and never reach satiety was something so mind-blowing to me especially because I'm on the other end of the spectrum. I often don't get hungry or I get hungry quickly after I eat. So never feeling full is something I just couldn't or can't relate to. The condition responsible for this is Prader-Willi syndrome. People with Prader-Willi syndrome are often followed by Genetics because it's a genetic condition. I'll get into that in a minute but I'll never forget my first few experiences and encounters with patients who had Prader-Willi syndrome. Stay with me.

[Music and Intro]

Well hey there. In today's episode of the RD Exam Made Easy Podcast, we're going over a case study. But first, I wanted to thank you for listening today. I'm so grateful for the feedback and topic ideas I've been getting because that's why I'm here for. This podcast is to help you get across the finish line. But beyond that, I've heard from Registered Dietitians about how helpful the podcast is as a refresher on topics they haven't used in years. I want to thank you for being here with me, for tuning in, and sharing out to share your wins and stories with me. Since we're on the topic of sharing wins…when you pass the RD Exam, I'd love for you to send me an email and let me know you passed so I can give you a big shout-out and a warm welcome into the RD community. Check out the show notes on how to share your big win with me! And if you have a question you want featured or a topic you want covered on the RD exam made easy podcast, let me know.

So let's get started by reviewing Prader-Willi syndrome, what it is and how it impacts a persons life who's been born with this condition. I'll also go over a couple case studies and share how I approached and helped these patients when they came to me for nutrition intervention.

First let's go over what Prader-Willi syndrome is. It's a congenital disorder which means it happens in utero during fetal development. In the early stages of development, chromosomes pair up but sometimes, things don't pair up as they should and a fetus can end up with chromosomal abnormalities. Prader-Willi occurs due to an abnormality in chromosome 15. There are a couple reasons it can occur but the most common reason is due to a

deletion in Chromosome 15 from the father. People with Prader-Willi syndrome never feel full or reach satiety. The condition affects the hypothalamus which is where hormones that control growth and appetite are produced. So that means people with this condition struggle with appetite regulation and growth which are 2 of the main characteristics of this genetic condition. The characteristics of Prader-Willi change throughout the lifecycle. During infancy, the 3 big red flags or indicators that there should be further work up are hypotonia which is when the baby is described as being floppy or lacking tone, failure to thrive, and difficulty feeding due to low tone. Hypotonia can happen for other reasons too but anytime a baby is diagnosed with hypotonia, further work-up is necessary to determine the cause. None of these symptoms are very specific but once the genetic tests are ran, Prader-Willi syndrome will show up on the results.

As the child grows, the characteristics change. The child will start eating everything, appearing to never feel full or satisfied. The medical term for eating excessive amounts of food is hyperphagia. This excessive intake then leads to obesity especially since there are also often issues with growth. Also, the poor tone causes decreased energy expenditure and nutrient needs. It's harder for the child to play and exercise like other kids due to the child getting tired more easily because they don't have the muscle strength or lean body mass to play as hard as the other kids. Physical therapy can help. The complications that arise from obesity such as type 2 diabetes and respiratory issues also affect people with Prader-Willi syndrome. They aren't immune to the additional complications and comorbidities. Another characteristic of Prader-Willi syndrome is learning disabilities. Also, they have hypogonadism which can cause delayed puberty. Most people with PWS are infertile. They are also at increased risk of osteoporosis. I previously mentioned short stature as one of the characteristics of PWS. Thankfully, because science is amazing, early intervention can include giving growth hormone which can help with promoting growth in these patients. Some other characteristics these patients have are behavior difficulties, anxiety, frustration, low mood, shouting and screaming. I was listening to a girl who was born with Prader-Willi syndrome and she did a great job explaining these behaviors. She said it feels like a tug-o-war inside, where her Prader-Willi is pulling her to act out and get frustrated even though she knows she shouldn't. I thought it was such a beautiful way of explaining it. She knows it's a part of her but not all of her and she has the awareness of what's happening even if she can't fully control it.

I've had several patients throughout my career with Prader-Willi syndrome. Two patients stand out the most to me. Both have Prader-Willi syndrome but had very different management.

The first patient, let's call her Samantha, was born to a mom with low education level. The mom was illiterate and had a very limited understanding of her child's condition. Samantha was 7 years old the first time I saw her. The mom had been educated many times before me by doctors and dietitians but she continued to have poor

understanding of how to manage her child's Prader-Willi syndrome. When I saw her the first time, the mom told me she never stops eating, she goes through all the cupboards in the day and at night time when people go to sleep, she hides food, and has also been found eating food out of the trash when there's no other food for her to eat. The mom said she needs to take out the trash before bed to try and stop Samantha from eating out of the trash. This was a food safety concern to me. She was at risk of food poisoning by going through he trash to find food that was sitting in the temperature danger zone. She also had no control of her eating and the mom wasn't sure how to implement any behavior modification to help Samantha's excessive intake.

After speaking with the mom and gathering a history of how she had been eating at home, I spoke with the mom about some strategies she could do to help her daughter. A couple strategies include having set meal times and working with Samantha so she understands when she can have meals. Otherwise she'd continue to eat all day. She was already obese for her age so the goal was to prevent further weight gain. I also recommended locking up the refrigerator and cupboards. Since Samantha was already obese, the mom asked about foods she can eat. At the time I saw Samantha, MyPlate wasn't yet available - it was still the MyPyramid so I used the pediatric pyramid as a guide to help mom review the foods and appropriate portion sizes. But remember how I mentioned the mom was illiterate? Well, that added another layer and was a little challenging since my artistic abilities are not the best. I tried my hardest and had to draw things out for her so she had something to reference at home that she could understand especially in her frustrating moments. I saw Samantha numerous times but unfortunately, the mom was not successful in implementing any of the strategies we discussed. I asked her about the barriers she was experiencing. She couldn't identify anything that was getting in the way of any of the recommendations. She wasn't motivated or wasn't able to make any changes. I answered her questions and really focused on letting her know she had support and could reach out if she had any questions. Mom's level of understanding and lack of ability to implement strategies made it a very challenging case. In the end, the recommendation was to follow a low calorie diet, eat at set mealtimes, eat veggies in between meals, drink water, take a calcium and vitamin D supplement, play and participate in fun exercise activities such as running around at the park or riding her bike and for the mom to lock the refrigerator, freezer, and cupboards. So that was Miss Samantha.

Now moving on to a completely different case. I'll call this patient Julius. Julius was a teenager, around 15 years old and he also had Prader-Willi syndrome. His mom was very proactive. From birth, she advocated for her son, getting him support and resources he needed, implemented set meal times and helped him understand the importance of eating during meal times. If he needed a snack, she'd give vegetables and other lower calorie, high nutrient foods. He had good growth due to growth hormones and his weight and BMI were both within normal limits. He was plotting around 70th percentile on the pediatric growth chart. Julius was getting assistance in school and had a couple good friends. He said he feels hungry but knows how to control his eating and I didn't

need to do any nutrition intervention other than to continue to encourage the great work they were doing and let them know they could contact me anytime with questions.

So as you can see, two people with the same condition but very different nutrition recommendations.

So let's review the nutrition recommendations for people with Prader-Willi syndrome.

- Research has shown that a calorie intake of 8-11 kcal/cm can help maintain body weight and prevent weight gain. In adults, most people should not exceed 1200-1400 kcal/day.
- Consuming an adequate amount of calcium and Vitamin D can help prevent osteoporosis. These patients may also need Calcium and Vitamin D supplementation.
- Eating a well-balanced diet that is low in processed foods or sugary foods helps decrease excessive calorie intake.
- Regular physical activity, including weight bearing activity not only helps manage their decreased muscle tone, it's also helpful for weight management.
- Higher intake of healthy fats, fiber and protein along with lots of vegetables that are low in calories, high in nutrients can help prevent weight gain and promote weight maintenance.
- Encouraging water to drink instead of high calorie beverages such as juice or pop helps maintain a low calorie diet.
- Providing appropriate portion sizes is critical since they can't self regulate. You can teach patients what appropriate portion sizes look like. I like using simple strategies such as using the palm of the hand, the thumb, and fist to recognize the right portion size since this can be used anywhere. Also, teaching portion sizes with food models are great for visual learners as well as patients who are illiterate - like in the case with Samantha.
- And lastly, locking up food in the cupboard, pantry, and refrigerator and removing the trash is a good intervention to prevent excessive intake.

I have so much compassion for people with PWS. It must be so uncomfortable to always feel hungry and have a need to constantly eat.

Studies have shown that people with Prader-Willi syndrome have elevated levels of Ghrelin. What's Ghrelin again? Well, it's a hormone produced mainly in the stomach. Ghrelin rises with fasting and is suppressed by food intake. This could be the reason they eat so much which can lead to obesity in people with this condition.

Let's do a quick review of Prader-Willi syndrome.

In infancy, people with PWS present with hypotonia or low tone, failure to thrive due to feeding difficulties because of low tone. The infant may need a special nipple to help with feeding or may need garage feedings. The difficulties with feeding are temporary.

In childhood, around the age of 2, the child begins to show inability to stop eating. They eat excessively, also called hyperphagia, and experience excessive hunger. As they continue to age, they can have hypogonadism which causes delayed or incomplete puberty. Most people with PWS are infertile. They also have short stature due to low levels of human growth hormone which can be treated with growth hormones.

We talked about Prader-Willi syndrome in this episode but I wanted to remind you to look at the big picture and think about the mechanism behind the condition. I want you to connect the dots and to truly understand the process because it can help you think critically when other conditions or scenarios present themselves. So what do I mean by that? Well, low tone in patients with PWS can also happen in other conditions. Having low tone also means having less lean body mass. Muscle burns more calories than fat. This is true for anyone. The more muscle you have, the more calories you burn. So with people who have PWS, they don't burn as many calories because the condition causes low tone which decreases the energy expenditure and therefore, means they need less calories in the day. When estimating nutrient needs, you need to keep this in mind. Instead of memorizing why these patients have decreased needs, make sure you understand the condition, the characteristics of it and the way you'd address the nutritional concerns.

Lastly, there is a separate food guide pyramid for PWS which recommends approximately 800-1200 kcal/day. The largest food group at the bottom of the pyramid is vegetables which as we know, are low in calories. The next level on the pyramid has 2 food groups - fruits and grains. Then the next level is milk and meats which recommend fewer servings and then lastly, the top level of the pyramid is fats and sweets which should be eaten sparingly. Vegetables are at the base of the pyramid and recommended as the primary food group because they're low in calories especially the non-starchy vegetables. In 1 serving, there's 5 gm CHO and 25 calories. This is easy to estimate when using the exchange list to calculate. So for someone who needs less calories, eating more low calorie foods when feeling constantly hungry can help with weight management in people with PWS.

I shared a lot about Prader-Willi syndrome including a lot of the characteristics that we as dietitians don't intervene on. But I think it's important to understand the syndrome as a whole. One things forsure, patients who have PWS do best when they receive early intervention and receive regular follow-ups by the interdisciplinary team including the dietitian who actually has a big role in guiding the patient and the family on how to manage calorie intake, eating behavior and weight.

Stay on top of your study game. There's no limits to achieving the success you so deeply desire. Until next time.

[Music and Outro]