Endocrine Care in Prader-Willi Syndrome

Birth to Age 2

FPWR Family Conference • Dr. Sani Roy, pediatric endocrinologist, Cook Children’s
This transcript was lightly edited for readability. Brackets mark wording that is unclear in the source or where the source ends. Dr. Roy describes the neonatal consensus recommendations as a work in progress at the time of the talk.

Introduction

Annamarie Huttunen, FPWR board member

I'm Annamarie Huttunen. I'm on the board of FPWR and I'm so excited to see all these new faces and really excited for Dr. Sani Roy who's at Cook Children's. She's a pediatric endocrinologist and she has an awesome talk for us today.

Dr. Sani Roy

That’s a high bar, Annamarie. Good afternoon. It's good to see everybody here. I am really just so honored to be here and to be at this FPWR conference. For the families in the room, is this your first time here at this conference? I'm really excited for you all and I hope that this is a great opportunity for you guys to connect with other families. Whatever we’re all going through, having a community may not make your story easier, but it does help to have other people you can connect with and share experiences with.
And I really hope that we can share some of our experiences and some information that will maybe help you as you think about what these next years ahead look like. So, these are just some disclosures. I am a volunteer adviser for the Global PWS Registry and I've been involved in some clinical trials. So, there are several things I'm excited to talk about with you all today.
This is actually a very exciting time in the Prader-Willi community in the sense that there is a lot of mobilization of getting our expertise of different clinicians across the country together to really think about how do we learn from each other?
How do we understand from what different patient experiences have been like, how we can make care better? And there is actually this large collaborative group that's been meeting actually these past few days called the Clinical Investigators Collaborative that I'm lucky to be a part of. And so from that there are some recommendations for the care of babies with PWS that throughout this talk we'll bring up some of those points. We're going to talk about several of the endocrine issues in the first two years of life. Obviously, growth hormone is a really important topic.

So, we'll talk about that as well, too. Endocrine care, it connects with so many other systems in the body. And so, how does that all relate together? We'll talk about that. And then I want to leave you all with some practical tips, some questions that you can talk about with your clinicians to just, you know, better understand how to care for your baby. So, I want to start off with this beautiful picture. I love these babies. Throughout my talk, you're going to see lots of pictures of adorable babies with PWS that I've taken from all the different groups from Ireland and Australia and you know, Africa and these beautiful babies I think are just such a wonderful reminder that every single baby is unique, right?
Not a single patient with Prader-Willi syndrome that I take care of is exactly the same. They all have their unique personalities, their own strengths. They all have their own challenges. They also all have their own potential. And I think it's really important to keep that big picture perspective in mind. Even if the days may be long sometimes, I think keep that perspective in mind like all babies. That there are going to be unique joys, there are going to be unique struggles, but we keep that big perspective in mind.

Developing recommendations for newborn care

Very briefly, I want to introduce you all to this work so that throughout the talk as you see some of these recommendations you'll understand where this is all coming from. So I mentioned to you all that there is this collaborative group that is working together and what we're really aiming to do is to try to help use our expertise to help our neonatologists that are in your community hospitals. I would love a show of hands from this audience here. If you are a caregiver or a parent, when your baby was born, h, you know, how much did you feel like your doctors knew about Prader-Willi syndrome?
Did did they had they heard of Prader-Willi syndrome before? Do they know very much about it? In some cases, it may be yes. In some cases, it may be no. Because even though it is common for us in the community, it is still a rare disease. So what we are aiming to do is to come up with recommendations using our expertise and using whatever evidence is available to help the babies of tomorrow be able to have a better experience perhaps or use the new evidence that we have to help their care. So, so this group, this Delphi project is throughout this talk, you're going to see some recommendation points that this is all work in progress that hopefully in the next three to six months, you'll see this actually in print what some of those recommendations are.
So, you're going to see some early snippets of that. On the right side of this, you'll see some information about a call to action to our families. So, it's one thing to have the clinicians give their expertise on what needs to happen based on the evidence. An equally important piece of this is what is the caregiver experience? What do parents and caregivers think is important? Right? What kind of education would help you as you leave the NICU and as you go home? What kind of things do you need to be taught? What kind of experiences did you have with your baby?
So, there is a survey that is active in the Global PWS Registry that Lisa will talk to you all more about and we would absolutely love all of our families and every kind of community to complete this because this will all be very formative as we put these recommendations together.

How the hypothalamus connects growth, feeding, and sleep

Let’s talk about Prader-Willi syndrome from an endocrine lens. So, the hypothalamus This is a very important part of the brain that you may have heard of. We consider this basically the control center of the brain because it that's what it does. It sends signals to another part of the brain called the pituitary that then sends signals to other organs in the body. The hypothalamus is the control center. And there's lots of different things that you see on this slide. It seems like very separate, very distinct things. Growth, feeding, development, sleep, and body regulation. They all seem so different, but the hypothalamus is really the unifier between all of this, okay? Because it does a lot of things to regulate every one of the things that's shown on this slide. So, if if we ask ourselves why are there so many different things that are talked about in Prader-Willi syndrome, a big part of this is because of the difference, the genetic difference that exists. It affects that part of the brain. And another important question is, well, why is one child, one baby with Prader-Willi syndrome so different from another? That's a really important point.

By the way, it's so easy to look at another family and another baby and think, okay, well, their experience is my experience. That is not going to be the case. Every baby's experience is going to be very different. I would encourage every one of you to just keep that perspective in mind because even if one baby has experienced something, it does not mean that's going to be exactly the same for your baby. And part of that, a big part of that has to do with the genetic differences and how it affects every baby is going to be very different. But in general, I will still say that these topics, these big picture topics are going to be things that are going to be important that we consider in every single baby.

And we have to follow these these things throughout their lifetime so that we can make sure we're not missing important important points.

Thyroid function

I want to start with the thyroid. So, why do we check thyroid function in babies with Prader-Willi syndrome? So first of all, what is the thyroid? The thyroid, you hear about it a lot in the media. It's really important for growth, critically important for brain development and for our metabolism. And that second point in particular, brain development is a very key point. So unlike in adults, if you have a thyroid issue, you might just feel a little tired. There may be concerned that you might gain weight. In babies in particular, it's critical for their brain development.

And in fact, if we're low on thyroid hormone, it can cause impairment. So that's why it's really important that if we have a risk for thyroid issues that we're closely monitoring that. So because the hypothalamus normally sends signals to the thyroid, there is a risk that we might have thyroid issues. And in general, hypothyroidism is more common in infants and children with Prader-Willi syndrome than in the general population. So you might be aware that thyroid is tested on the newborn screen. Depending on the state that you're in, the way that they screen for thyroid issues may not pick up on the type of thyroid issue we're seeing here in Prader-Willi syndrome.

Because the classic thyroid issue is an actual thyroid issue. And in most of our babies with Prader-Willi syndrome, we're not actually concerned about a thyroid issue. We are actually concerned that the brain is not sending the right signal to the thyroid. So there's two labs here that are shown and you may be familiar with some of this, but I'm going to explain it in case you aren't familiar. There's one called TSH that's made in the brain and there's one called T4 that's made in the thyroid. And they're normally I think of them like a seesaw with one another.
If normally the brain will send a signal to the thyroid and if the thyroid levels are low, the brain is going to recognize that it's going to make more TSH. And you would see a high TSH. That is not going to always be the case in Prader-Willi syndrome because if the brain is not regulating properly even if the thyroid level is low that TSH may not go up the way it should. So the key point here is that in Prader-Willi syndrome in babies where there's in anybody where there's a possibility of a brain related issue the T4 is critical the free T4. So that's why you would see your endocrinologist will probably say that they're going to pay a lot of attention to your baby's free T4 more so than the TSH.
So the key point here is a normal TSH does not always mean normal function. The free T4 is critical. And I'll tell you just in general with the thyroid not all of our babies with Prader-Willi syndrome are going to need thyroid hormone. Some of the babies will okay and there are studies that have looked at this where sometimes a baby needs to be on it during that critical period in early infancy because babies need a lot of thyroid hormone relative to older children and adults. The amount for their weight that they need is a lot higher. And I do see this in my practice as well that sometimes babies need it for the initial period and then they actually kind of come off of it later on.
But we still have a risk for this. So, it is still an important thing that throughout life we have to keep an eye out on that. And your endocrinologist is going to be very careful about whether they allow your baby to be on thyroid hormone. And let me tell you why that is. Because if we give too much thyroid hormone to a baby or let's say a baby doesn't need it or they get too much, it could impair their weight gain. So we have to be very careful because too much in endocrine I see some of my endocrinologists in this room here. We're very careful about we got to be in that fine balance.
Too little is a problem. Too much is also a problem. So that hopefully that helps there. And again at the very end we will hopefully have time for questions. So please do save your your questions. And by the way, that little consensus statement is going to be one of the things that's that comes out, which is that in babies with persistent low free T4, levothyroxine treatment may be considered. But the key point following an individualized endocrine assessment.

Central adrenal insufficiency

I want to move on to central adrenal insufficiency. Okay, the adrenal glands are a very important part of the body that is also regulated by the brain. And what do the adrenal glands do? Okay, you might hear in common social media adrenal fatigue. A lot of adults adult groups might talk about that, but really in babies, what we're talking about is if a child is at risk for adrenal issues, the adrenal controls a lot of important things such as your blood pressure and your blood sugar. Everybody is supposed to make a little bit of what's called cortisol on a day-to-day, but when we're sick or let's say we go under anesthesia, we need a lot more.

And so there this is one of the areas that I will tell you all is of limited evidence and mixed evidence in the literature meaning that there are some studies that show that there is a really high rate of adrenal problems. And there are other studies that don't show so much. I will tell you in general in discussions with other pediatric groups and from what I've seen in the very limited literature on this, it appears to be relatively uncommon in babies. However, it is something that your endocrinologist will probably be keeping in mind and periodically assessing, especially if your baby happens to go for a procedure under anesthesia because like I mentioned when we're going under any medical stress, that's when the risks are a lot higher. So, that is something that they'll probably be keeping in mind. And let's say your baby's going for a procedure under anesthesia. That might be a question you ask your endocrinologist is, is my baby's cortisol okay? And if a baby is not making cortisol, I want to give you some reassurance. And this is one of the things I love about endocrine, which is that if you're missing a hormone, we can almost always replace it. And so that is something to keep in mind is if your baby is not making cortisol, there is a treatment which is hydrocortisone steroids.
Again, in endocrine, that same philosophy, too much is a problem, too little is a problem, right? So your endocrinologist can help you figure that out. Okay. And that's what is listed here in that Delphi consensus that's going to come out. Which is the physicians are going to tell your community doctors that, you know, not every baby necessarily has adrenal issues, but we don't want to miss it. So there are going to be certain circumstances where your doctors are going to be more likely to want to test for this.

Mini-puberty and undescended testicles

I want to talk about puberty. Did you know that babies undergo a puberty? Yes, they do. It's such an interesting phenomenon. It's called mini puberty. And during the first few months of life, it is actually normal for babies to have a small surge in puberty hormones. Girls might have little breasts that appear. Sometimes a little bit of vaginal discharge. Now in babies with Prader-Willi syndrome sometimes this response is altered again a little suppressed because we may not be sending that full signal to the gonads. And so where does that come to play in our little girls not so much early on in life but for our little boys undescended testicles are common.
Sometimes one, sometimes both and sometimes, and I have seen this, sometimes those testicles do actually descend. They come pretty close to descending. So, if you have a baby with unescended testicles, you know, that doesn't mean right away we jump to surgery. Your urologist and your endocrinologist will probably tell you, you know, let's watch and wait and see. And a lot of times they will come down quite a bit, but there are some times when it doesn’t really come down. And so in general this recommendation that is coming from our board group is in line with what the general urologic urology recommendations are for babies which is that if it's persistently not descending there comes a point in time where a baby may need surgery to bring those testicles down.
There is a treatment that you may have heard of called hCG and I will tell you this is this that's the common hormone that rises in women during pregnancy but there are some very very small reports that have looked at using hCG to bring those testicles down. I will tell you that is something that would be considered on an individual case-by-case basis. In general, the small

case reports that exist show that if it's in the abdomen, it's unlikely that the hCG therapy is going to bring those testicles down. But let's say your baby's testicles are maybe just a little bit high. It just needs a little bit more in those small small reports. Sometimes that's enough to bring it down. It's not a super simple treatment. So, it's always one that I always tell my families, listen, there's very limited data on this. Some of our families sometimes opt to try this and see if it will bring the testicles down. But again, this is going to be a case by case discussion with your doctors.

Growth hormone

Growth hormone. This is a critical medicine that I want to spend a few minutes on. Growth hormone is so much more than just height. In Prader-Willi syndrome, it is so critical for so many other functions. For our muscle mass, for our body composition, physical functioning. This is one that's a little bit harder in terms of measurement, but we do have data on its improvements on body composition. I would say just talking to our families, a lot of our families do seem to feel and I do see differences when my babies start growth hormone, we with that increased muscle mass that we get, you know, we have more vitality that happens as well.
And again in Prader-Willi syndrome what we are doing is we are replacing what our babies are not making. Okay, every baby is different how much growth hormone they need. So it's it's very easy to look at your friend's baby who is on a totally different dose than your baby and that is going to happen very likely because what your baby makes and needs replaced is going to be different than what another baby makes. Okay, these are some really nice pictures. They're old. They're from 2000 from I think it's PWSA took this booklet out. But you can see the difference in this baby on the top before starting growth hormone and then after.
You can see that vitality. And then on those bottom pictures, the left two are before starting growth hormone. This is an older child who started growth hormone. And then look on the right. What a difference you can see in this child's height and in his body composition. So, it's really nice to know now that growth hormone and we're able to get growth hormone a lot younger in our babies.

Sleep and breathing

One quick pause I want to take here is talking about sleep and breathing Prader-Willi syndrome and I'm going to explain why this is important with regard to okay so first let me define this term sleep-disordered breathing that is kind of a big term that is capturing the various type of issues that can occur during breathing and sleep okay there's one called obstructive sleep apnea where you've got the drive to breathe okay the brain has sent the signal, but the airway is blocked for one reason or another. Maybe the tonsils are enlarged, which is more likely in an older child.
Adenoids can develop in infancy and not necessarily in the first 6 months of life. There are some studies that do show it as young as 3 months. However, usually it's after 6 months of life that we might start to have some adenoids again that can block that airflow. Then you've got something called central sleep apnea where the airway is open and there's no blockage here but we are

missing the drive from the brain to breathe which is again that hypothalamus is a big part of this right and central sleep apnea is often treated with oxygen especially in infants.
Okay, one important point that I want to mention is that growth hormone in the studies that we have so far seems to improve central sleep apnea. But if the baby has a blockage that is present, depending on the reason for the blockage, that's where growth hormone can be a bit more challenging and it's something that needs to be monitored a lot more carefully because growth hormone can make lymphoid tissue grow. So if we had adenoids or tonsils that were very enlarged, for example, your doctor might tell your family, we got to pause on the growth hormone right now until we get this under control and then we're going to get back onto that.
So depending on the type of obstruction, that's where this is important. So I want to briefly share some data that our team has worked with four other large PWS centers and looked at babies who had a baseline sleep study. A polysomnogram is a sleep study. And what we learned is it's actually really common. This is actually surprisingly different from some of the other data that we've seen. Sleep apnea on our study, nine out of 10 babies did have sleep apnea on their baseline sleep study. However, what's really interesting is that three in four of those babies had no new intervention that was done after that baseline sleep study.
And when a treatment was recommended, it was usually oxygen. It was very rarely something more serious such as surgery or airway pressure. And under six months of age in this study of 88 babies when there was an intervention it was oxygen. So all that to say when growth hormone was started young we did not see an overall worsening of sleep-disordered breathing and oxygen measures actually improved after we started growth hormone. So I will say this is an area that we are still learning a lot about. We need more studies in this area. There's a lot of interest among clinicians in general to really look at this because our goal is to figure out this point right here.
We want to start growth hormones. The key point is as early as possible but also as safely as possible.

Before and after starting growth hormone

This is a general pathway that I like to share with you all which is before growth hormone there's some important baseline safety assessments that we need to do which is to kind of check baseline thyroid studies for example so you got to check potentially adrenal function and the endocrine systems it's very interesting that sometimes when you start one hormone it kind of uncovers another hormone issue that's present. So that's why these baseline assessments are something that your endocrinologist will likely recommend. From a sleep and breathing standpoint, it's important before we start growth hormone that we have somebody with that specialty expertise, a pulmonary doctor, a sleep medicine doctor.
So, your endocrinologist is very likely going to want somebody with that particular expertise to also weigh in and say what their thoughts are on your baby's breathing status. There are a few different ways that you can assess your baby's breathing status. Polysomnography, the sleep study is the classic method that's been done, you know, all these years. We are slowly looking into better understanding when it is acceptable to do something different such as oximetry which is basically measuring your oxygenation levels and as part of our guidelines or our pathways

that we'll recommend the consensus pathways we are going to provide more guidance [unclear] on how you kind of navigate that.
When do you wait on growth hormone? This is the key point is when there is acute medical instability. So if if a baby for whatever reason is acutely really having issues, breathing issues, remember that your doctor will be working very closely with you to get that under control. And I know it can feel like a lifetime when your baby is not on growth hormone but remember that growth hormone in and of itself is not enough. We've got to balance everything, right? We have to make sure that the breathing part is balanced. Once that is resolved or whatever acute issues are going on, then the goal is get back on that growth hormone as soon as you can.
And after you start growth hormone, it's really important that you reassess how things are going. So, generally speaking, around 6 to 12 weeks. It takes a few months for that growth hormone to really work in the system. After about that time, your doctor's probably going to want to reassess these different hormones and potentially see if we need to do any kind of things. I just realized I have no idea what time.

Swallowing and aspiration

Endocrine care and how it connects with nutrition, sleep, growth, and development. So I want to do a quick run through of some other issues because you know your baby is a whole baby. Endocrine is one important part but there are all these other things that our families are thinking about and how does this all connect together. So dysphagia is a term I want to define. What that is swallow dysfunction. This is common in our babies with PWS and I want to reassure you it will get better. So if your baby is in that season right now, hang on. It will get better even if it feels like a lifetime right now.
There have been a few studies, Dr. [name unclear]'s group and our group. We looked specifically at swallow in babies with PWS and our study was a study of 18 of our babies. What we found is that all of our babies had [unclear] at different rates. All of them. One out of every three of our babies did have some aspiration. Aspiration is when food rather than going into the gut is going into the airways. So, one in our three babies had aspiration. But the tricky part is that 100% of the time it was silent, meaning that we couldn't hear it. Couldn't hear a cough or a gag to be able to indicate that.
So what that means is sometimes a baby looks like he or she's feeding safely but may still be aspirating. So early on in life is when this seems to be more of an issue. So that is important point to be thinking about for our neonatologists our community doctors. One of the things that we are going to be recommending is that even if a baby doesn't seem to have symptoms of that, we want them to not miss the possibility of aspiration. Now, I do want to give one disclaimer here, which is we don't actually know yet what that means. In other words, if the baby is aspirating, what does that mean in terms of long-term risk?
Okay, nobody really has that data yet, and I'm sure we will get that. It's going to be important for us to look at that, you know, and actually figure out does that lead to long-term breathing issues. So, we don't actually know that yet. But it is important still as of now, we are still recommending that we have a low threshold to evaluate this.

Temporary feeding support

Because of the feeding issues being so common, a lot of our babies do need some temporary help with feeding. This has been a really hot topic in the PWS community over the last several years. Really thinking about, you know, what type of feeding tool is right for your baby. I do want to first emphasize again that every baby's situation is very different. So sometimes a baby does we need a G-tube but most of the times what we are actually finding is that a baby is fine just with a nasogastric tube if a tube is needed I will say very very rarely a baby doesn't need it most of the times our babies do need it most of the times the nasogastric tube is sufficient and there is limited data here but some emerging data that is showing some evidence that there's less risk with the nasogastric tube.

So this is why as a clinician group we are working to spread this information to our neonatologists that most of the time an NG tube is sufficient. The caregiver perspective here is also really important. We cannot just go based on the evidence and the data sometimes we also have to pay very close attention to what the needs the caregivers are. And I have had occasional circumstances where after months and months of needing an NG tube, a family just could not do it anymore and they were really struggling. They opted for a G-tube. So that is your family's choice and your decision.

Just just hold on to what is important to your family. We are still learning what's what's real in terms of the evidence and gathering that information. So again the goal is for now what I would say the goal is NG tubes are the preferred [unclear] feeding method when there is insufficient intake. All right. Building the ah I want to go back. I'm sorry. There's one more point I wanted to say here. Going back to the feeding issues, there’s one important point I want to make. In the study that we did, this was about 350 babies from the Global PWS Registry.

We actually found that by 6 months of age, the NG tube was out in most babies. So, if you are in the season of feeding challenges, again, want to reemphasize that point that it will get better. A lot of our babies actually between two and four and six months of age it's kind of coming out so a lot quicker than some other populations.

Building healthy eating habits

I did want to take a second to talk about this. So once we are out of the season of the feeding tube and dealing with those type of feeds and we're actually taking those complementary foods, what do you do? That's a totally different season. No matter what type of baby you have, whether we have Prader-Willi syndrome or any other issue that we are dealing with, infancy is a very critical time for adequate nutrition and healthy growth. It is not a time to be focused and absolutely driving yourself crazy [unclear]. I do want to make sure people know that Dr. Miller has done some incredible work in this field really categorizing the various nutritional stages of PWS that we are still very much close attention to today.

Everything is a little bit different as to when those nutritional changes happen, how they occur. I will reassure you that it is not a light switch. So please please do not drive yourself crazy looking at every little thing that your child is doing and thinking is my child in this next stage. As long as you are closely following with a dietitian and with your endocrinologist they’re going to be able to

[unclear]. It is really important that from the beginning you set good habits early. And so what I mean by that is variety is important. We want to have a whole variety of different textures and flavors as appropriate as developmentally appropriate for your baby.
We want to focus on nutrientdense foods. We want to try to set good habits and avoid the high concentrated sweets. I will say there is a lot even beyond PWS. There is a lot of research about sugar in general. Okay, the inflammatory properties of sugar and just in general you think about yourself. I think about, you know, just think about ourselves and the foods we eat. I would say most people would often say the more sugar we consume, what happens? We crave it. I would say the more we consume sugar, we often tend to crave it more. And I would also say that the sweet taste is something that's very important to just be mindful of as a family.
And I would say try to set those good habits early because if we get that sweet taste and that sweet craving at a young age it's going to be a lot harder when we get into those older stages and we are dealing with those more significant nutritional and hunger related challenges.

Procedures and anesthesia

So this is an area that's also of great passion to us in the clinical community. I think what's really challenging is for any child any child when you go under anesthesia that is a stressful time as a parent about thinking about that procedure. And I will say for families of a child with any kind of condition there is an added stress because you're thinking to yourself what does my doctor need to know about PWS? And I will say there are some good resources that are out there. I think PWSA actually has a good resource on anesthesia and Prader-Willi syndrome and this is an important thing to talk about with your doctor before your child goes under anesthesia.
And you know just to find out you would ask your endocrinologist for example are there any risk that we need to think about before going under anesthesia such as the adrenal issue that we talked about earlier. Is my child safe from a breathing standpoint to go under anesthesia? That's another good question to ask your doctor. And you want to make sure and let your anesthesia team know that your child has Prader-Willi syndrome. So, one of the things that I think as a clinical group we are working on is how do we get more resources out there for our families so that you're not having to do all this heavy lifting yourself?
Because that is another added stress for you. Will your child go home that same day? Every child is going to be different. Every anesthetic drug is going to be different. So, a lot of it is going to depend on your baby's specific circumstances. Is their sleep apnea controlled? If they have sleep apnea and it's not controlled, there's a very good chance your baby probably needs to stay the night and be monitored, especially in a baby under two years. We can't communicate in particular, right? So, but if we don't have sleep apnea or it's controlled then then you know in some cases we might have just a longer observation period.
These are all things that it's important to plan before the procedure.

Hip dysplasia

Hip dysplasia is more common in Prader-Willi syndrome and that's basically where those hip joints are a little bit more loose. And so babies again because of our low muscle tone they may

look fine and not have symptoms. We may have some laxity or looseness of those hips. So that is another thing to be keeping in mind. In general between 6 to 8 weeks of age or so is a good time for baby to think about whether or not a hip ultrasound should be done to see if there's any looseness. The goal is if there is an issue, we want to address it as soon as possible. It is better to get on it on the front end than be correcting it on the back end.

Development and early therapies

this is so important. And this is basically getting into working on motor skills and our communication skills, our feeding skills. And I'll tell you all that just you know experientially this does get better with time. I do believe [unclear] very likely helps as well. Starting those therapies early is a big benefit to your to your baby. That is a very formative period. Those early years there's a lot of room for improvement during those years. [Unclear.] And remember your baby will get stronger. It's it's just going to be he or she will follow their own trajectory, but keep working on it.

Don't get frustrated by that slow progress because you'll get there, but you just have to push through it. And then the needs will change over time. You know, your child may not need every single specialist, every single year and moment of their life. And so just be keeping that perspective in mind as to what is the important need right here, right now.

Your baby’s care team

Caring for a baby with PWS takes a team. This looks like a very intimidating list, I will say. Not every single specialist is needed all the time, but there are likely going to be key moments where your child does need to see these different specialists. Endocrinology is going to be one that you're probably going to get very close to for a long time, particularly during those, you know, developmental years through age 18, although there are benefits beyond that as well, too. Your sleep and pulmonary doctor, particularly early on, we need to make sure that things are safe.

But over time, that is probably going to be a specialist that your endocrinologist is likely going to want you to periodically check in with dependent as well on what symptoms we're experiencing. Nutrition is critical throughout and that is really because we do not want our parents to have to be figuring that out. It is so hard when it is your own child to look and figure out what exactly does my child need today. That's where the dietitian nutritionist comes into getting that big picture, take a bird's eye view in and help you guys navigate how do we transition those different nutritional stages.

Genetics is particularly important early on and for a lot of our families who want to grow their family. That's another important thing as well too to be able to meet better understanding you know what do other risks look like. And I do not want to miss primary care. I definitely want to emphasize the importance of a good primary care pediatrician. Your endocrinologist would love to help with those things, but I will tell you, I do not want to manage ear infections. I would be the wrong person to manage that. So, I would absolutely recommend that you find a good

pediatrician that your family feels comfortable with, that you feel like you can ask questions to cuz we want to partner with them.
And we want to make sure that between your pediatrician and between our specialty team, we can address all these different issues.

Questions to ask your care team

What kinds of things can you ask? And you know please feel free to take a picture of this if you'd like. You know growth hormone. How is my baby doing on growth hormone? What monitoring is needed while on growth hormone? Do we need to check levels? These are some good questions to ask while on growth hormone. I'll go into the sleep. You know, just and these are not necessarily every single visit you ask this, but you kind of try to figure out when you need to ask these questions. Does my baby need an overnight oxygen study? Does my baby need a sleep study?
Especially if we've got, you know, if we notice symptoms like snoring or, you know, issues with sleep. You want to be able to feel comfortable to ask those questions. Eating and swallowing. Is feeding safe for my baby? Do we need to consider a swallow study? Okay, if we've never had one, do we need to get one done? If a tube feeding is needed, and you're in that situation trying to figure out what to do, what is the least invasive option? What are all my options that are out there? When can we try more oral feeds? In some of our families who babies have had a G-tube, When do we take that G-tube out?
I will tell you, sometimes that G-tube just stays in quite a bit of time. And you know we don't honestly know the caveat is we don't really know what happens if you keep it in too long but if it doesn't need to be there could we get it out you know and avoid potentially infection risk etc.
Developmentally is my baby making the expected developmental progress that's where your pediatrician comes in right and remember your baby's trajectory is not going to look like maybe a baby's trajectory who doesn't have syndrome but the most important thing is movement we want to see that we're moving in the right direction.
Should we get PT, OT, speech, feeding therapy? What should we do at home to support development? That's a great question to ask your therapist because the therapist is going to do what they can right there. A big part of that development is going to happen at home. By the way, how many of your children have siblings? Can you raise your hand? That is a very key role. And I want to emphasize that to you guys. Use those siblings. Big brothers, big sisters, they are one of your best developmental tools. Encourage them to stimulate your child, read to your child, play, do silly stuff.
All of those things are going to help your baby's development. Especially when my babies are very, very early on in the NICU and maybe they're just not as responsive. That's a really hard time. It's very hard. I've been there, done that. As a mom, it's hard when they're not communicating with you and maybe engaging and showing that kind of reciprocity, that affection back to you, but that's an important time that we want to try to stimulate them and continue to connect with them. Procedures and screening. If anesthesia is needed, does my team know my child has PWS?

Does my baby need steroids? And then looking ahead, what specialist do we need now? Is there anybody we don't need to see as well? Don't hesitate to ask. You are a very critical part. You are, I would say, the most important part of your care team.

Taking care of yourself

I want to end with this part. I think this is so important. You have to take care of yourself. So important. And I would say this is probably the hardest part is seeing the struggles of our families when they feel like they're alone. I love that you guys are all here together so you can connect with each other. I would say form connections, exchange contacts. If you are in a dual partner household, try to give each other time to have for yourself. And you know just know that you are not on your own with this. You know that's where you want your team to be a part of it with you.

If you don't feel like you can bond with your baby right then, that's okay. Give it time. It will happen just with time as their personality comes out. Okay, I think that's everything I have. I have a lot of members of my team here that I want to introduce as well. We've got Allison is our nurse coordinator for my clinic. Nathan is with my research team. Stephanie is our endocrine nurse practitioner. Mark is my sleep nurse practitioner. Stephanie is also with my research team. So anyway, I want to say, you know, you can't do this on your own. This is why I have a team. We need a team of people to effectively care for our families and our…

[The source transcript ends here.]