Bone Marrow Transplantation For the Treatment of Patients With Nonmalignant Disorders
Webcast
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Laurie Burroughs, M.D.
Mary Pablo

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Introduction

Andrew Schorr:
Imagine your baby was one of the one out of every 50 or 100,000 newborns that was born without an intact immune system and what a risk that would be for them. Where would you go for care, and what is the latest care? We’ll tell you all about that with a mom who experienced that with her small child and a leading medical expert next on Patient Power.

Andrew Schorr:
Hello and welcome to Patient Power sponsored by the Seattle Cancer Care Alliance. I'm Andrew Schorr. Well, often we've talked about more common cancers and then some less common cancers, and then there are other conditions that affect the immune system, sometimes in adults and sometimes in newborn babies. And we're going to talk about one of those nonmalignant conditions in children now. It's called SCID, and basically it's when there's maybe a birth one in 50,000 or one in 100,000 when a child is born without a complete intact immune system, and therefore they're at risk of dying from an infection that maybe would be fairly harmless or not serious at all to someone else. Who do you do about it?

Well, here in Seattle, the Seattle Cancer Care Alliance and the Fred Hutchinson Cancer Research Center and doctors from Seattle Children's and the University of Washington all work together in really what's been groundbreaking work to restore the child to a healthy immune system and give them the greatest chance for a full life. So as I always like to do I'd like to introduce you to someone who has lived this, so I want to let you hear the story of Mary Pablo and her baby, Maddie or Madysen.

So, Mary, you and your husband, you were living back not too far from Oklahoma City on a military base, right? And you have two older children, two boys, Storm and Brock, right?

Maddie’s Story

Mary:
Yes, that's correct.
Andrew Schorr:
And then here comes Maddie, born a little early, like five weeks early, but still you had every reason to believe that she was healthy, right?

Mary:
Absolutely. We left the hospital. My husband worked at the hospital, and we went home, and she's supposed to be fine.

Andrew Schorr:
Right. And we should mention that your husband, does he go by Manny?

Mary:
Yes, he does.

Andrew Schorr:
So Manny is a nurse, an Army nurse, and so there you were in the Army hospital, everything is fine, start nursing. What sort of things started happening that made you rush to the emergency room?

Mary:
Well, I was just nursing her, and before I actually went into the ER where my husband worked, it happened the time before, Madysen turned blue, almost as if, very lifeless. And it alarmed me.

Andrew Schorr:
No kidding.

Mary:
Yeah. And then, you know, because I also had two other kids that were normal. And the second time I took her into the ER and my husband worked there, I think at the beginning they thought I was just kind of, maybe I had postpartum depression or something. But it wasn't the case. She had turned blue, and I would nurse her and her oxygen, they ended up Medevac'g her to Oklahoma City because it continued to keep happening.

Andrew Schorr:
Wow. Now, I'm going to tell a little bit of your story in the interest of time, but you were on a journey there in Oklahoma, trying to figure out what was going on. And in the end, finally after many months there was some concern about her bone marrow, right?

Mary:
Yes.
Andrew Schorr:
Okay. And then that triggered something for Manny. Manny gets on the internet, starts searching about bone marrow and realizes that back in Seattle is where they invented bone marrow transplant and where they had a lot of knowledge about it related to treatment.

Mary:
Yes. He researched a lot about it, especially with us being in the military, there was Madigan Army Medical Center there, and he made a couple of phone calls, and I believe we got onto a conference with an infectious disease doctor there, Dr. Fairchok, and my husband and the doctors and his command discussed what could be done for us as a family just because we were going to probably foresee some kind of bone marrow transplant and we were all new to it.

Andrew Schorr:
Right. So the Army helped out. You were transferred to the major Army base in the Seattle area, Tacoma area, Fort Lewis, another Army hospital, Madigan, your husband could work there. Doctor there, you were mentioning, and then with ties up the road to the Seattle Cancer Care Alliance where they among other things specialize in bone marrow transplant for this condition.

Mary:
Yes. They Medevac'd Madysen and I straight to Children's Hospital, and then we went up to Madigan and got into contact with SCCA after they looked up everything, all the things that were happening with her and so forth.

Andrew Schorr:
Right. So was your thinking that you heard this term "SCID," and we're going to get your doctor to tell us more about it in a second, but basically your child was lacking a healthy immune system. You became aware that a bone marrow transplant at the Seattle Cancer Care Alliance was her best hope?

Mary:
Yes, that is correct.

Andrew Schorr:
Well, let's meet one of the doctors who was involved, and that's Dr. Laurie Burroughs. Dr. Burroughs is a pediatric hematologist oncologist. She's an acting assistant professor of pediatrics at the University of Washington. She's of course at the Seattle Cancer Care Alliance, Seattle Children's. She does research at Fred Hutchinson Cancer Research Center. Dr. Burroughs, what is SCID?

Understanding SCIDs

Dr. Burroughs:
Hello, thank you. Severe combined immunodeficiency or SCID is a disease where a patient or a person is born without a healthy immune system, and because of that they're unable to fight infections, normal infections that you and I would encounter
in the environment just in normal daily living. Our own immune systems, our healthy immune systems can fight those off. However, in a patient who is born without a healthy immune system they're unfortunately not able to fight those off, and it can be quite dangerous to them. In fact in severe combined immunodeficiency most babies do not survive beyond a year of life without a transplant.

**Andrew Schorr:**
Now, some of us remember the story of the boy in the bubble or the Bubble Boy and the idea that he didn't have an immune system and literally had to be isolated. Is that the kind of thing we're talking about?

**Dr. Burroughs:**
Correct. That is.

**Andrew Schorr:**
Wow. Okay. That's quite an image. So you want to give the child a new immune system, a healthy immune system, and so when it comes to transplant we know that you look for matches. So, Mary, help us understand. Your whole family was typed to see was there a match of bone marrow that could benefit your new baby.

**Mary:**
Yes. The two boys, myself and my husband, Manny, were all typed, and the day before my husband got sent to Iraq they found out from SCCA and Children's that Storm was a match, our oldest son.

**Andrew Schorr:**
Wow. Well, we'll tell the story of Manny. I should just say that Army nurse sent to Iraq, as many people in the military have been or are even there now. But he wasn't there long, right? The Army said your daughter is really sick. We're going to bring you back.

**Mary:**
Yeah, well, the bone marrow transplant was planned right after he had left, and we had our doctors call him home just because it was more of, you know, there's a lot of things that happen with bone marrow and it's not something that, you know, if you don't have to do it alone you shouldn't.

**Andrew Schorr:**
Right. Well, it sounds like the Army did a lot of good things for you, and finally it was figured out what it was. Let's talk about your older child, Storm. He's 17 now, but we're going to flashback two years. So Maddie is about one, he's 15, so you find out he's a perfect match. So, Dr. Burroughs, help us understand. You're withdrawing some bone marrow cells or cells from the bone marrow of Storm and then you're giving them so they'll take, if you will, and become the new immune system for Maddie. Did I get it right?
Dr. Burroughs:
That is correct. Yes. The cell, so a lot of your immune cells, the ones that are not working correctly in severe combined immunodeficiency or SCID, those cells come from the bone marrow, and so that's why you can do a bone marrow transplant for severe combined immunodeficiency. So you take those bone marrow cells from the donor and you literally give them to the patient, Maddie in this case, like a blood transfusion. And they are fairly smart, and they find their way back to Maddie's bone marrow and set up shop there and therefore produce her immune system.

Andrew Schorr:
Now, what is the expertise here in Seattle for doing this? Because we're talking about an extremely rare condition.

Dr. Burroughs:
Right. I think the expertise is actually multifocal. So to begin with we have a world-renowned immunology team at Seattle Children's that has an enormous depth of experience with patients with primary immunodeficiency disorders and severe combined immunodeficiency is the most commonly transplanted immunodeficiency. And Dr. Torgerson and Dr. Ochs and Dr. Rawlings and Dr. Scharenberg and Dr. Skoda-Smith are all excellent, excellent physicians who have enormous expertise in diagnosing, because that's one of the biggest more important things for these patients who have an immunodeficiency is to get their diagnosis quickly, and to try and sort out what's going on with these patients and then to figure out what do they need.

And particularly with SCID we know that the sooner we can get them to transplant the better their outcome is, the more likely they're going to survive the transplant. And the reason for that is that you can't live very long without an immune system, one particularly that's not working as much as SCID, so the quicker you can get those kids to a transplant the better. And so that's why having immunologists who are experts in this and know exactly when they start to see some of the red flags going off, so to speak, or the bells and whistles chiming, saying, hey, this kid doesn't look like they have a normal immune system, then Dr. Torgerson and his group really work hard to try and diagnose it and figure out where, what happened in the genetics there that led to this problem.

So she was quickly then referred from the immunologist to the transplant service. We did the typing of the family, found that Storm was the donor, which was excellent, because if you can transplant with a matched sibling the outcome is way better than it is if you have to go to an unrelated donor or someone who isn't matched with the patient. So we were very lucky. There's only a 25 percent chance that the sibling will match, so we were quite happy that Storm was a match.

Andrew Schorr:
All right. Now, I have to get back to Mary for a second because I think people understood that there was some time going on in Oklahoma, and when the
transplant finally happened in Seattle and you'd moved and everything Maddie was a year old. So it wasn't so clear to the doctors back in Oklahoma, was it, Mary? It took quite a while.

Mary:
It wasn't clear at all actually, and it actually took a lot of probing as parents and a lot of going back and forth and being told that it's just pneumonia. And I think like she said, it needs to be caught immediately and you have, by the time we left there Maddie was six months old, and she was only 12 pounds.

Andrew Schorr:
Wow. Now, Dr. Burroughs, why is it so tough? So we said it's really rare. So would it be that your pediatrician has never seen this in their career, may never see it again, and it's probably just a cold that kids get?

Rarity of SCIDs Among Healthcare Professionals

Dr. Burroughs:
So it is very rare, and as a result many physicians have never seen it. So most children are healthy, and as a result many pediatricians do not see what's called severe combined immunodeficiency, and many children are coming in with colds and there's lots of other reasons that could account for why they're having the weight loss or different things that are occurring to the baby. And so unfortunately many of these kids do not get discovered until they have that life-threatening infection. And in fact as a result of this we're really trying to push newborn screening because it is a disease that can be found in the newborn screen. So when the baby is first born you test them for it, and it is a disease that we'd like to try and get nationally in the newborn screening because it would significantly, many of our kids do not come in until they are several months old and already having a significant infection and failure to thrive, which is what Maddie had.

Andrew Schorr:
Who would the screening involve? Because kids are screened for a lots of things, those little heel pricks.

Dr. Burroughs:
It would be exactly like that, with the heel stick. So the technology is coming down the pike to be able to put this into a newborn screen, similar to how we screen for different, sickle cell disease or thalassemia or some of the other metabolic diseases. We're hoping that one day we can have this in newborn screening because it would greatly improve the outlook for patients.

Andrew Schorr:
Well, happily things so far are working out well for Maddie. So now she's three years old, and while you have to be really protective of her, obviously in that interim before the transplant, Mary, and you've been, you know, incrementally
maybe less productive post transplant, still three years old, she's still playing at home. I understand she loves her dolls. She loves to dress up. Can we say hi to Maddie for just a second?

Mary:
Yes. Hang on one second.

Madysen:
Hello.

Andrew Schorr:
Hello. That is Madysen.

Dr. Burroughs:
Hi, Maddie.

Andrew Schorr:
Hi, Maddie. There's your doctor, Maddie. So are you dressed up as a ballerina today.

Madysen:
I'm Madysen.

Andrew Schorr:
Oh, okay. And what's the name of your doll?

Madysen:
Madeline.

Andrew Schorr:
Madeline, okay. Well, we hope you have a very nice day, and we're delighted to talk to you, okay? Can we talk to your mom again? Thank you, sweetie.

Mary:
Hello.

Andrew Schorr:
Okay. Well, I think for our audience, the fact that here's a three-year-old child who has survived this really life-threatening situation, first of all, Dr. Burroughs, that must do your heart good.

Dr. Burroughs:
Oh, for sure. And seeing her, I've been lucky enough to run into her, either, the clinic visits just seem to coordinate, but watching her grow up is really neat.

Andrew Schorr:
I'm sure. And, Mary, so let's talk for other parents. So there are people listening to this program somewhere in the world, and a condition, this condition or a related
Mary:
Yeah, and it was really hard for me in the beginning just because my husband worked in the hospital. And you couldn't escape that, what is going on in the hospital, you know. Your faith is there, and we think you should have faith in your doctors and your nurses. But I do think as a parent if you think that you need to question, that's why they're there. And that's one of the things with Seattle Cancer Care Alliance and Children's Hospital, that I had a lot of faith in what we were doing, even when I signed the papers. We did a clinical trial with Madysen, and one of my biggest fears was what would the outcome be. And we were doing a clinical trial, and how often had this been done before and so forth, and part of it was they've done this before and they've done it many times. And my thing was take the chance. Because if you take the chance and you go ahead and go on with the bone marrow as they suggested or what the doctor is suggesting depending upon how you go with it, you're paving the way for somebody else, and you're paving the way for somebody else's child.

Andrew Schorr:
That's a great message, but also though you had to connect your family and your daughter with the right people for her condition, and that took a lot of proactive work on your part. I mean, let's face it, there was nobody really in Oklahoma who was kind of pushing you to go to Seattle or saying, you know there's this center. You had to discover that yourself.

Connecting with the SCCA from Afar

Mary:
Yeah. You have to. And, you know, when your kids are sick and you don't know what's wrong or you don't think that there's, you need to. You need to get on the internet. And I'm not going to say that I didn't spend many nights on the internet, my husband and I, and, you know, I have to say I don't believe everything I read on there. I actually stopped reading it for a while. Because every individual situation is an individual situation, and you have to have faith in what you're doing. And what matters when your kids are sick is your kids. Anything else, whatever is going on outside, it's your kids and your family.

Andrew Schorr:
Right. Well, you know, this individualized situation is one that's important.

You know, as far as SCID goes, Dr. Burroughs, one child may exemplify it in one way, but it doesn't mean that the next child still diagnosed with SCID would be exactly the same, right?
Dr. Burroughs:
That's correct. There's a wide range of SCID, and it depends on how severe the
defect is, but the majority all present with a significant life-threatening infection.

Andrew Schorr:
But then you're looking right at the individual characteristics of their blood. Let me
broaden this discussion just a little, and that is we think of bone marrow
transplantation for malignancies, but here's an example for nonmalignant condition.
So there's a significant amount of that that goes on, not just in SCID but there are
other disorders too, right?

Dr. Burroughs:
Correct. There are lots of different immunodeficiencies or other nonmalignant
diseases, so patients are born with other parts of their immune system that don't
function correctly that come from the bone marrow where if you can replace that
bone marrow you can correct that underlying problem. Similarly we transplant
patients who have different marrow failure syndromes, meaning that for some
reason their marrow just shuts down, it doesn't work properly, and it doesn't
produce the blood cells that you need for life. So it doesn't necessarily produce
your red cells or your platelets or your white cells. So we also can successfully
transplant patients with those different disorders as well or those different diseases.

Andrew Schorr:
What are the names of some of those other conditions?

Other Diseases Treated with BMTs

Dr. Burroughs:
Shwachman-Diamond would be one, Diamond Blackfan anemia, Kostmann's
syndrome. I'm trying to think of, Fanconi anemia. We also do transplants for
patients who have what's called a hemoglobinopathy, meaning they're born with
something similar, such as sickle cell disease would be an example or thalassemia.

Andrew Schorr:
Now, is the hope then that the donated marrow will do the whole job, and you can,
or is it that there may be other therapies that are indicated as well?

Dr. Burroughs:
It depends on the disease that patients have. So the goal is of course to have it
correct the entire underlying problem, but some of these diseases have other
associated problems with them. So for example some of patients who have marrow
failure syndrome also have problems for example with their kidneys or their heart,
and unfortunately the transplant can't correct that. It can only correct the
underlying bone marrow problem. So a lot of times the patients still have other
problems that are associated with their disease that they came with before the
transplant.
Andrew Schorr:
Let me understand the team that comes into play when someone comes to Seattle. So you are on the transplant side.

Dr. Burroughs:
Correct.

Andrew Schorr:
And we talked about all these immunology people in the lab and all the scientists who are looking very much at the actual characteristics of that patient. Now, what are some of the other team members that come into play? And then we’re going to talk to Mary too about just living in Seattle as your child goes through this. But what are some, when we talk about a multidisciplinary team related to these conditions, who are the players?

Importance of a Multi-Disciplinary Team

Dr. Burroughs:
Right. That’s an excellent question, and because of the complexity of these patients they actually do need a multidisciplinary team, one who is familiar with all aspects of their disease. And because these kids often come in with such significant, profound problems, so for example in Maddie’s case, Maddie not only had the immunodeficiency so the immunologists were critical, she needed transplant, so the transplant docs were critical. But she had significant infection, so we had infectious disease there as well. We also had nutrition involved because we required means of trying to put weight on her and helping her gain her weight since she was so severely malnourished from the disease itself.

We also have child life experts who really help provide, in Maddie’s case she was an infant, but it’s really been helpful for the little bit older kids, the toddlers and that, to help them deal with the emotional stress of going through a transplant and being sick and trying to get down to their level and help them cope with it. So we also have child social workers and child psychiatrists and psychologists to try and help patients who have significant life-threatening illnesses and help them cope with that.

In terms of the transplant team, we have an enormous, over ten transplanters here at the Seattle Cancer Care Alliance, all who specialize in pediatric transplantation. I run the nonmalignant program, but all of us are familiar with nonmalignant diseases as well as the oncologic diseases, and all work to try and improve the field.

Andrew Schorr:
Wow. Well, you must be very proud of what you have at the Seattle Cancer Care Alliance related to these conditions.
Dr. Burroughs:
The most exciting thing is to see Maddie, you know, to see a child who has gone through what she has and is smiling at you, a year or two years, three years down the line, and you know that you've made a difference.

Andrew Schorr:
Right. Right. Mary, let's just talk about the logistics. So again somebody may be listening to this and they still live in Oklahoma or New York or who knows where, and they say, well, I hear that there's this team that Dr. Burroughs just described. That's what my child needs. And you had or have two other children, and they're in school and all that. You moved the whole family here, but then you needed to be up in Seattle for the resources that she needed both when she was in the hospital or to be close to the hospital. So did the kids actually go to a special school? And where did you live?

Mary:
We actually decided as a family that it was important to keep our family together, and we decided to go ahead and opt to stay at the Ronald McDonald's House right across from Children's Hospital. So we went ahead, and we stayed there. I did take the two boys out of school, which was a big decision. My mom and stepdad actually flew out. We're from Guam, and they flew out here and stayed with us, an option that maybe some people don't have, but I was able to, they were here for a couple of weeks in the beginning to get us started out right. But my kids had a lot of options when they went down to SCCA and Children's Hospital. I took them out, and they went to the Hutch school.

Andrew Schorr:
Right. The Fred Hutchinson school.

Mary:
Yes, which is amazing. It's geared towards kids who are going through similar things like bone marrow transplant and so forth. And it kind of, when I think about what we had to do down there in Seattle I think of it as being, we basically stayed in a triangle, and it was probably the best triangle you'll ever have to stay in, but probably, you're going through a lot at the time but it's very geared toward, my philosophy of it was that it was very geared towards treating the family.

And when we got down there, you know, they set a date for transplant, and they said, this is the date we'll start and so forth which means that, you know, that's when you'll be inpatient and they'll go through and they'll do their realms of chemo or whatever have you. And it's a daily thing, and like I said we stayed there in the hospital for about 45 days and had to be 30 minutes away.

The thing that I most liked about it is that they do really have a great team. From the child life specialists to nutrition, the PAs and nurses, they have it geared where in the Children's Hospital where the child will be staying for the most part, well, in my case, they had an SCCA floor there, and all those nurses in there are geared to
take care of bone marrow patients. So you want to be somewhere, it's very special if you can be somewhere where they know what to do and they've done it before. That is, you know. Because they know, it's not just something that they're just seeing.

**Andrew Schorr:**
Right. So, Mary, let me give you an opportunity to say something publicly. So Dr. Laurie Burroughs is part of that team, and Maddie is here probably because of that and of course because of Storm's donation as well. What do you want to say to Dr. Burroughs publicly now?

**Mary:**
I want to thank her. Oh, God. You know it's, I'm a parent and I'm just very grateful. I'm very grateful that I'm able to speak today and that, they've really helped us. They really got us to a point where we had faith in the system and what was going on and what was going to happen to our daughter. And it takes a lot, and they saved her life. I think as a parent you need to fight for your kids, and when you work with great people, I mean they have wonderful people that work there, and it shows. They are a world-renowned team. It's almost like a song, but I mean they know each note and they know what they're doing. And that's the best thing possibly that I could say is just thank you. They are a great team.

**Dr. Burroughs:**
You are incredible.

**Andrew Schorr:**
Kind of chokes us all up. Well, this is what we wanted to bring to people around the world, to understand that when you're a parent who knows what lies ahead and you can be totally blindsided by a condition you've never heard of, Mary, right?

**Mary:**
Oh, yeah.

**Andrew Schorr:**
And maybe even the first doctors you see have never seen, and then you have to advocate for your child as you did, and you get your own Army medal for that, you and Manny. And thank goodness then the resources of the Army were brought to bear too to help, and I know you're grateful for that, and then connecting with the right medical team. Now, I understand there's still challenges along the way for Maddie as her immune system is kind of tweaked and measured and she goes on, but you must look at her in her ballerina outfit and just your heart must open up for that.

**Mary:**
Yeah. We're just happy that she's able to dance, I mean in everything.

**Dr. Burroughs:**
You should see her, she's absolutely adorable.
Andrew Schorr:
Well, how about this: Let's all make a plan to dance at Maddie's wedding.

Dr. Burroughs:
There you go.

Mary:
Oh, gosh.

Andrew Schorr:
Dance with her, and you better start, start saving for that and start saving for college. Believe me, I've got older kids. So, Mary Pablo, I want to thank you, well, first of you all, I want to thank you for what you've done for your kid.

Mary:
Thanks.

Andrew Schorr:
And I want to thank you for what you're doing for our listeners today, and I want to wish you, Maddie, Manny, Brock, Storm, the whole crew, if you have cats and dogs, all of you, to have a lot of happiness. Thank you so much for being with us.

Mary:
Thank you.

Andrew Schorr:
And Dr. Laurie Burroughs from the Seattle Cancer Care Alliance, thank you for what you and your team do and for helping us better understand bone marrow transplant as part of the treatment for patients with nonmalignant disorders.

Dr. Burroughs:
Oh, thank you. And it's patients like Maddie why we're here.

Andrew Schorr:
Right, no, absolutely. Well, this is what we do time after time on Patient Power. I'm so delighted we can do this with experts and families, touch, related to the Seattle Cancer Care Alliance. And remember Seattle Cancer Care Alliance is a wonderful organization combining the expertise of the University of Washington, UW Medicine, Seattle Children's, and also the renowned Fred Hutchinson Cancer Research Center. I'm Andrew Schorr. Thank you for joining us. Remember, knowledge can be the best medicine of all.

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