Spotlight on the Individual, Achondroplasia
December 2022

Sarah: Hello, I'm Sarah Chart. On behalf of Optum Health Education, I would like to thank you for listening. Living with a rare disease impacts individuals and their family and caregivers, both physically and mentally. Today we welcome Deborah Hecht, an individual with achondroplasia, to discuss her insights and perspectives and provide strategies that the interprofessional health care team can utilize to better support those that are impacted by rare diseases such as achondroplasia.

As a reminder, this podcast is accredited and worth 0.50 credits. More information can be found on optumhealtheducation.com.

I'd like to invite the listeners, if they haven't already, to participate in the webcast on achondroplasia and other causes of disproportionate short stature, featuring Dr. Gary Bellus from Geisinger Health System in Danville, PA. Dr. Bellus discussed a great deal about achondroplasia, and other causes of disproportionate short stature including diagnosis, current and emerging therapies, and how to encourage individual and caregiver engagement in shared decision-making. If you haven't had a chance to access it, it is available on demand at optumhealtheducation.com.

Deb, thank you for joining us. To get us started, I thought we could discuss a little about your childhood. You are the youngest of five children and the only one with dwarfism, can you tell us what it was like growing up as the only member of your family with dwarfism?

Deb: Thank you, Sarah. I was raised as a very strong independent woman from my parents. They made sure that they wanted to treat me just like everyone else in the family, and my siblings did. The siblings did the same thing.

one of the biggest independences that they provided me was being able to drive.

Driving gave us freedom to be able to do the things that other people do and in life I always said to myself, I've always wanted to go the extra mile to prove myself, and that usually works and really helps me in my everyday life.

Sarah: It sounds as though your parents really did you a great service by encouraging you to be really independent. And would you like to discuss a little bit about how you decided to start your own family and how the Little People of America, how are also known as the LPA adoption network, played a part in your decision?

Deb: Yes, I joined LPA as a teenager after high school and I wanted to see what it would do for me.

UM, since I had lived in an average sized world for about 19 years and I did join and I was a member for several years, I had not married or my lifestyle had not changed, and so I checked into when one wanting a child I we looked into adopting and we had an adoption network, and we still do. And I got to adopt my daughter, who was nine years old at the time. Also, with the contemplation dwarfism from Moscow, Russia.

Through the Little People of America, my daughter is now 36 and has two new babies. I've become a grandmother. I have a granddaughter and grandson.

And they're both as a babies are dwarfs too. So I really have enjoyed being a single mom and helping out my daughter and her two little ones.

Sarah: well, congratulations on being a grandmother now. That must have been quite an experience adopting 9-year-old Maria from Russia. How did her early medical care in the orphanage impact her as an adolescent? And then as an adult with achondroplasia?

Deb: Well, when I found out when I went to Russia to adopter, I found out that they do not keep medical records and the medical itself on being done with her.

Umm. Doctors don't see the children in the orphanage often, so the lack of treatment as a child did lead to her lifelong chronic ear infections that she has.

Sarah: Well, that's really unfortunate. It sounds as though if Maria had received more care, more thorough care as an infant, she may not have had to have dealt with those lifelong chronic ear infections as a new grandmother of two little ones with achondroplasia. Are there any other medical considerations or insights that you could share with us?

Deb: Umm. Well, first of all, with Lorraine was born. She's now 10 months old. She was born with breathing issues.

And then Roland has some, um, issues in the hospital now with breathing and oxygen breathing treatments for now, but possible shunt that they put in the back of the head if it doesn't resolve as they grow. I am with a conservative doctor right now that is watching the process on Lorraine.

On the size of the head in relation to the body, UM is important in a counter pleasure, dwarfism and of course the advancements of prenatal care.

when I was born, the only thing that I can remember hearing from my mother and sister were that I was turning blue until 9 to 12 months, and I was having breathing issues.

UM. Anesthesia considerations? we have to be careful with, you know, with the NSC geologists that they know how to do that. If we have any surgeries.

And then I also with me, my legs go numb. If I either walk too long or I sit without putting my feet on the floor, not being able to walk very far. I have to sit and rest.

UM at 25 years old, I collapsed in the mall when I was meeting my sister and I had what was called the lower lumbar Laminectomy and I was able to walk.

UM, very, very nicely. Once that surgery was done. Also, we also have leg straightening my daughter Maria. She had leg straightening. The legs are bowed, usually in a counter, plays the dwarfism and one of her legs was extremely bowed and we had that done when she was about 17 years old. But we do have back problems and leg problems and sometimes some arm problems too in regard to pain.

Sarah: Well, thank you for sharing that with us. I’d like to transition a bit and talk about shared decision-making. For those of you who may not be familiar with shared decision-making, it promotes the collaboration between the individual (or family of the individual) and the health care team, with the ultimate goal of improved health and satisfaction.

Individuals (or families of individuals) with disabilities and complex medical conditions, face many decisions about medical treatment. These choices may be added stressors for the individual, the family, and the health care team involved. The nature and complexity of decisions, of course, are highly variable and may involve diagnosis, evaluation, treatment, care management, and support services.

When conducted well, shared decision-making affords an appropriate balance, incorporating voices of all stakeholders, ultimately supporting the individual, family, and the health care team.

Deb, have you felt you and your family have had the opportunity to participate in shared decision-making?

Deb: Yes, very much so. When we meet with doctors. I make it a point to ask a lot of questions. we get second and third opinions because we have so many doctors that offer their advice, particularly the orthopedic doctors and the neurosurgeon doctors and now we have several breathing doctors involved in our lifetime in our lifestyle so and I like the conservative approach that they don't. For example, they don't say that they're going to take do surgery immediately. Right now, we're going through a long process on deciding whether there's going to be surgery on.

Breathing and there's I feel there's a lot to look at before they decide that we say yes to actually going in and doing something about it.

Sarah: It sounds as though you're really taking the time, and you've really found the right physicians that here you and what is right for you and your family for in terms of your medical approaches to?

And Deb, do you have any other advice for listeners on how to overcome the stigmas that you and your family experience on a daily basis? You know, how can we and those listening today, how can we make the change that we want to see?

Deb: This is a really interesting topic because you learn a lot being out, seeing the seeing the public and how they react shifts over the year to today. Adults more are a little more rude to us when they see us on out and about.

Children have been very understanding because they think they are around other people that have disabilities such as blindness. Being in a wheelchair.

We advise for people that come in contact with us to ask questions, make the change, don't start, you know, making fun of and of course TV shows out there, such as the learning channel and representation, and I go to grade schools to help kids understand I let them know when they see me. You know, I'm just like everybody else. I do everything like everyone else. So it's just more exposure and letting people know that we're just like everyone else.

Sarah: Yeah, that's great that you do that, that you go and speak with children at school.

And to give the learners a bit of perspective on what it's like to be a little person, can you speak to items and to things that make your life a little bit easier? So for example, if a health care professional wanted to make their office more accessible, what items should they consider incorporating that would make things easier for you?

Deb: OK, the one number one thing that we use that we use a lot, in fact, I have one in every single room of my home is stools.

And many little people can accommodate in their home to bring things down lower. Many have their kitchen cabinets lowered so that they can reach everything. I did not do that. I use a stool at the sink. I have a stool in the living room.

Still not bedroom cannot access. Getting in a high bed, so stools are very, very important. And then of course back to when we talked about driving. We have to have pedal extensions or for some of them that are not as.

Even tall as me at 4 feet, some have to have hand controls and it turns the wheel either left or right the way they need them.

But driving pedal extensions is very important and getting ones that are very safe and secure.

Sarah: Thank you. It's helpful to hear that information for us.

And we know that that you're very active with the LPA and you spoke to that a little bit when we first started speaking. Can you talk about how they have helped you?

Deb: Yes, Little People of America has been a just a wonderful thing to be a part of. I joined again at 19 after I was out of high school. My parents didn't think I needed it while I was a child. They had known about it, but they just didn't. So, I went ahead and researched it briefly and pursued to go attend my first convention in Detroit. A long time ago and.

Now they try to reach out at birth, to get the parents to let them know in many, many every size parents join. Now we have a large organization we have volunteering.

Except excluding the director, everyone volunteers to be a part of it, whether they're an officer or a regional director or a chapter president. My life totally changed by having LPA my life, I wouldn't have had adopted Maria if it wasn't for the LPA network. We network with a lot of hospitals and orphanages and other places where dwarf children have been placed because the parents couldn't accept the differences of that child. So, it was a bonus that if Maria didn't have her biological parents, I was able to be her parent and that was the best thing that ever happened.

Sarah: That's really wonderful, really amazing. And it sounds like a really terrific organization and the one that's clearly changed, it's been a life changer for you and I know you give a lot to that organization.

I want to remind the listeners that building awareness of rare diseases is important as many individuals go undiagnosed and, therefore, untreated. More research is needed for identification, causes, and treatments.

By advocating for earlier identification, research into causes and treatments, and increasing resources in our communities, we can help the millions of people that are affected by rare diseases and their families to live healthier lives.

Deb, thank you so much for being our guest today on our Podcast Series: The Diagnostic Odyssey for Individuals With Rare Diseases.

Before we close, do you have any closing thoughts that you would like to share?

Deb: I would just like to share with everyone that if when you see someone with a difference don't hesitate to ask questions. We are always open to ask because we know that those people sometime are hesitant about what to say, what to do.

Most all of us are open to.

Being able to have them feel more comfortable being around us and that's going to make everyone happy. So, it really does help the situation, but thank you.

Sarah: Well, thank you. And I can't think of a more perfect way to close out this podcast today. I know as well. Debbie, you had shared with us the little people of America motto. I wonder if you just want to want to close out with that as well.

Deb: OK, our motto and we say it very often is that little people like, me and my family, we can do anything that an average sized person can do. We just may have to do it differently. And that is really the truth. Our everyday motto.

Sarah: Thank you, Deb. I'm really appreciate your insights and your time, and I hope you hope you have a great rest of the day. Thank you for sharing with us today and to our learners.

On behalf of Optum Health Education, we would like to thank Deborah Hecht for providing insight on this very important topic. We would also like to thank the Family Engagement Center / Special Needs Initiative / Complex Care Concierge, and the (C3) Advocacy, UnitedHealthcare for their support.

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If you do have any questions, please reach out to moreinfo@optumhealtheducation.com.

This concludes our podcast. Thank you, and I hope you have a good day.