Episode 50- PJ's Protocol: A Lifesaving Procedure Fueled by Love

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Mindy Henderson: Welcome to the Quest Podcast, proudly presented by the Muscular Dystrophy Association as part of the Quest family of content. I'm your host, Mindy Henderson. Together, we are here to bring thoughtful conversation to the neuromuscular disease community and beyond, about issues affecting those with neuromuscular disease and other disabilities and those who love them. We are here for you, to educate and inform, to demystify, to inspire, and to entertain. We are here shining a light on all that makes you, you. Whether you are one of us, love someone who is, or are on another journey altogether, thanks for joining. Now, let's get started.

For patients living with neuromuscular disease and their families, there are important tactics and strategies unique to these conditions that it's imperative to be aware of and prepared for in an emergency situation. Specifically for patients living with Duchenne muscular dystrophy, there's an important protocol we will be talking about today. March marks the 10th anniversary of what has become known as PJ's Protocol, and today, I have three incredibly impressive guests with me to discuss the protocol. Their credentials are far too many to mention, and you'll hear more about their accomplishments as the conversation unfolds. But I am truly grateful to each of them for taking the time to be here with me today.

We're going to jump right in and start with Brian Nicholoff. But first, I want to just thank all of you for taking the time out of your busy schedules to be here with me today. Brian, your passion and powerful advocacy led to the creation of

PJ's Protocol. I know that your son, PJ, passed in 2013 and the protocol in his name was established in March of 2015. It's now available in several languages and referred to by countless families and physicians in many parts of the world. Can you just share your son's story with us and how PJ's Protocol came to be?

Brian Nicholoff: Thank you, Mindy and Dr. Mendell and Amy for being here and sharing this special journey of love, labor of love, with PJ. He passed in 2013. I'm going to try to compress a little bit of this six days from hell. We were down in Florida, and we're getting ready to go to the pool. And I picked him up to transfer him to the wheelchair, and he kind of, didn't fall, he kind of sat. And he said, "Dad, I think I broke my hip," or, "my femur," whatever. He would know, because he's done it before on both sides. And I said, "Well, just relax for a minute." So I picked him up, and when I tried to pick him up, I shattered his humerus bone.

We called emergency, and then, they come by. And this was in Tampa, and they went to Tampa General. And naturally, he had broken his hip, up by the hip joint, or his femur up by the hip joint. And he's broken every bone in his right leg before, due to the fact of being on prednisone since 1987. Matter of fact, he was in the first prednisone trial at Ohio State with Dr. Mendell, which was 30-some years ago. And so, instead of having surgery there, we transferred him up here to Indianapolis, because we had a surgeon already set that worked with him before. And that was on a Tuesday. And we came up, and he didn't have surgery until a Thursday. Now, remember that, because that's about 48 to 50-some hours, where long bone fracture, that keeps on coming up, long bone fracture, which is the humerus and, naturally, the femur.

But he had his surgery, and everything seemed to go fine. And then, Friday, everything seemed to go fine. Saturday, things were different. His heart beat was different, his blood pressure was way low, a lot of symptoms were coming up. And those symptoms were, according to the physicians, it could have been ARDS, it could have been pneumonia, this, this, and that. Never did come up about steroid deficiency or stress dose or anything like that. Why? Because we brought PJ's Deflazacort to the hospital, gave it to the nurse for them to administer. Okay. They administered the other drugs, because they had them there. They didn't. So long story short, he had several different tests, and one thing led to another. And he did not survive. About three days later, on a Tuesday, he passed. He didn't pass in any pain. He was under at that time. He didn't talk to us for the last three days, because this was all part of, which we learned, part of the steroid adrenal crisis, whatever, all these factors, all these symptoms.

So we immediately came home after he passed and brought his clothes home, and Barb noticed that he didn't give him any of the pills. That blew my mind. Blew my mind. So we thought about it for a little bit, and we immediately called Dr. Mendell. I might not have this exactly, but we called him and he said, "Well, tell me again." So I told him what the blood pressures were and the other symptoms and this and that. He said, "Well, can't make a total definition of it or that, but I would think that that would have something to do with not having

steroids." Steroid deficiency, I believe it was. "Look into that." And I said, "Well, I don't know how to look into that." He said, "Well, call Pat Furlong and Kathi over at PPMD." That's why I made initial contact with them. And that led into a total connection that is indescribable, not only with PPMD, but MDA and other organizations, just DMD, just the neuromuscular world in general, the village, it's really something.

So one thing led to another, and I got the medical records. And I didn't get all of them. And according to my personal physician, he said, "There should be boxes of those." So I went back, and I got boxes of those. And my personal physician took about three months to verify that so-and-so was in there, that he signed. So he determined also that it was steroid deficiency per se. I met with the insurance risk people at the hospital, and they declined that. And so, I told them, point blank, I says, "You know, I've got the time and the resources to take this to the limit, but I don't want to do that. I want a win-win-win. I want PJ's legacy. I don't want this to happen to anybody again. I want your physicians, your staff, to be educated about DMD and neuromuscular disease and the symptoms of what happens in critical care situations, different things can arise and you've got to refer to these and so forth."

And so, that ended up being passed. Pat and Kathi came over here a couple of times, got it written up. Dr. Mendell was involved with that. Dr. Norris, Norris and several other physicians, and it created PJ's Protocol, a six-page protocol, which Dr. Mendell will describe more in those terms. And so, that was instituted on March 2nd of 2015, about a year and a half. And I remember when we took it into effect, we had a, not a function, but kind of a release of that. And I remember standing in front of the physicians, Dr. Norris read it and all that, that, "You know, I hope to God, someday, that this protocol will be acknowledged anywhere from Boston to Bakersfield." And I tell you what, it sure as hell have been a lot more than that. So that 10 years of that has been... I've stated this over thousands of times, I've got it memorized in my heart of what happened day by day and minute by minute.

Naturally, sometimes, the emotions take over and change that. But to put it in a nutshell, PJ passed, he was failing to a degree anyway. He was 31. His fraction was in the mid twenties. He was obese. His bones were like brittle. They were minus six or something like that. But what a joyful, joyful person. We would've never expected that, because we've been in and out of hospitals many a times with the surgeries. Everything went well, we came back home, we went through our rehab, we went through all that, and the times just kept on going on. This was different. This was life-altering by all means. But to have people like yourself join this and lead me through this, you're the wind beneath our wings, Barbara and I. It really is. It's undescribable of how I can sit here and be in front of you three and the other people and share this situation. And I'm encouraged that it'll continue on more and more. So PJ is right next to us all the time.

Mindy Henderson: Always. Absolutely. Thank you so much for sharing that, Brian. I know it's not an easy story to tell. And I have deep admiration for you and your passion that led

us to this point. I am going to move to Dr. Mendell now. Dr. Mendell, you have a background that is, Brian's already talked about it, it's so important to the neuromuscular community. And I want to share just a little bit about you, if you don't mind indulging me. You attended medical school at UT Southwestern Medical Center and completed your residency at Columbia University. You also completed a postgraduate fellowship at the NIH, and your first employment provided contact with the first patient with Duchenne, inspiring decades of work as a translational clinician and researcher. Over a 50-year career, you devoted efforts to understanding Duchenne muscular dystrophy and ways to intervene. You are the lead author introducing prednisone for DMD treatment, and the importance of that treatment is validated in over 30 publications.

It's the standard of care for DMD and led to FDA approval for, I hope I'm going to say these right, Deflazacort and Vamorolone. Your impacts to the DMD community are mind-blowing and your awards are too many to mention, so I'm going to stop there. But Dr. Mendell, for anyone listening, could you give us a little bit of an overview of DMD and how corticosteroids and steroids, I'm not sure if it's one or the other or both, but come into play with and use for treatment for that condition? What's accomplished with those drugs?

Dr. Jerry Mendell: Duchenne muscular dystrophy is what we call an X-linked recessive disease. It affects boys predominantly. The remarkable thing is it actually starts in utero before the boys are born. We know that, because they're born with an elevated muscle enzyme, which tells us that their muscles are already affected. It also manifests by not uncommonly delayed motor milestones, but it's ignored at that age until about three to four years old when parents want to have some answers why their kids are not playing up or not playing as well with the other kids on the block. And that leads to the diagnosis. So most kids are diagnosed by four and certainly by five years of age, and that's improved tremendously over the last 10 years, certainly since PJ was diagnosed. And once the disease is diagnosed, the kids now are put on steroids, and we'll come back to that point.

The steroids, Brian mentioned them all, prednisone, Deflazacort and Vamorolone, and we'll discuss those a bit more. But any one of those steroids are effective. They have different side effect profiles. And then, about early teenage years, well, actually, before steroids, it was before teenage years, but now that's been extended to teenage years, that wheelchair dependency comes into the picture. And then, after kids are in a wheelchair, they have a slowly progressive disease that leads with accompanying heart involvement. The heart also depends on dystrophin. Dystrophin is the protein that supports the muscle. It was known first to support skeletal muscle, and now, we know it supports heart muscle and actually even smooth muscle, that is the GI tract is also dependent on dystrophin. And so, with that dependency, the patients really suffer, progressively, their heart becomes more severely involved, develop heart failure, and then, the respiratory muscles are affected and they pass, as Brian described for PJ.

The story of steroids does involve me from very early on. In the late seventies, I was involved with a muscle study group. There were four of us who had been trained at the NIH, and we got together and said that, "There's no treatment for this disease. Steroids are debated as to whether they can be effective." And we developed a protocol for a clinical trial. It was a very large protocol for the time. This was 50 years ago basically. And we did a study of 103 actually boys with Duchenne dystrophy, and PJ was part of that clinical trial.

And we did a dose escalation study to establish clearly the dose of prednisone. We found that prednisone was effective. It was debated before that, but now, it showed efficacy, it increased strength, delayed the progression of the disease, and improved the quality of life. Of course, it came with many side effects, and the impact of prednisone was illustrated in many ways. First of all, it was published in the New England Journal of Medicine, which is the highest level of clinical publication recognition you can have. So that kind of spread the word that prednisone was effective.

And then, it led to further developments, and Brian mentioned one was Deflazacort. Deflazacort was introduced because it had fewer side effects and the same efficacy of prednisone. And more recently, Vamorolone, which I'll mention a little bit later in this discussion, Vamorolone has dissociated side effects or efficacy as we refer to it. Because it preserves adrenal function and doesn't allow what happened to PJ. So it has merit. So that's the story of where we are with PJ and how we got to prednisone and potential withdrawal, which we'll discuss more in a few minutes. So I'll leave it there.

Mindy Henderson: Perfect. Can you talk a little bit more about PJ's Protocol and how it's applied in a medical setting, what parents who may be listening should know about making sure that it's enacted?

Dr. Jerry Mendell: Well, we know PJ's story, so I don't need to reiterate that. But just to emphasize the salient points about PJ is everything was going well with the doctors, but they didn't have an appreciation for steroids and muscular dystrophy. And that's not uncommon. That's why Brian and Barb developed the PJ Protocol. They recognize that other doctors who don't have much familiarity with Duchenne muscular dystrophy, and remember it is a rare disease, although, over the last 50 years, we have more and more recognition for this, because it's the most common severe form of muscular dystrophy. But still go to the emergency room, the doctors, they may know a little bit about muscular dystrophy in a generic sense, but they don't know much about the importance of prednisone within the disease or the other steroids I mentioned, and that these steroids have to continue.

Once you've been on long-term steroids, your own source in the body of producing prednisone or its equivalent, which is hydrocortisone, is suppressed. It won't make it anymore. If it doesn't make it anymore, you have to take it by mouth or worse, by intravenous, but that's usually not the case. Usually, you can take it by mouth. And this occurs in any stressful situation, whether it's

emotional stress, surgical stress, dental stress, you can name it. But boys who are under stress need five to 10 times, depending on how much stress there is, five to 10 times the dose of prednisone that they ordinarily get. And in this situation that Brian described with PJ, he underwent major surgery, and we came out of major surgery and his body is expecting to have more steroids and not getting any. So the setting for crisis is set, and that's what happened.

So we owe a lot to Barb and Brian to develop this PJ Protocol and the people they worked with, he mentioned the private foundations that supported that, PPMD and MDA, and then, other doctors who pitched in to develop the protocol. The protocol is really basically an emergency manual that tells people stepwise how to use prednisone or how to use steroids. It is a six-page document and goes through every phase of what should be done, when it's needed, and why it's needed. It's needed because the body will not produce its own hydrocortisone anymore. It's needed because if you don't give it, you go through what's called a withdrawal or adrenal crisis, and this makes it impossible for these children to survive. So then, it will tell how, the protocol actually is very detailed, it tells how this happens, explains that. It explains what the stress doses of prednisone are.

It explains when it's needed. It explains how to administer it. Some boys who are under such stress, for example, coming out of surgery, a major surgery, they can't take oral medication, the doses that are equivalent to what they were taking. It describes the symptoms of withdrawal. And even for doctors who are taking care of patients with muscular dystrophy, it provides test protocols for how to test if a boy is susceptible to developing or susceptible to being dependent on steroids and what doses would make that acceptable in any crisis situation. And then, of course, it describes the adrenal crisis and the symptoms that Brian alluded to, heart rate going up, fever developing, blood pressure dropping, weakness developing, faintness and really essentially passing out. So it describes everything. It's a critical protocol that PCPs or primary care physicians, especially emergency room physicians and doctors that encounter the boys during their medical care for any, whether it's mild, moderate, or severe. So it's critical. And I don't know if you want to talk about how parents should handle this, but I'll stop there and give you the lead for the...

Mindy Henderson: Absolutely. Thank you. That was such a great overview. And clearly, this is an incredibly powerful tool for parents to have in their toolbox for their children. If you can believe it, I have one more equally impressive guest that I want to introduce now, Amy Aikins. Amy, you are a board certified patient advocate and were led to your current career from your experience as a mother to a young man also with Duchenne muscular dystrophy. You have unselfishly utilized your personal experience, navigating systems and overcoming barriers for your son in order to assist others. I understand that you're currently the Director of Patient Access at the Little Hercules Foundation, where you tackle treatment access concerns, through extensive engagement with a variety of stakeholders and involvement in various groups and projects focused on access issues within the

rare disease community. Would you please just start by telling us a little bit about your son?

Amy Aikins: Sure, Mindy, and thank you for that introduction. So my son, Elijah, he is currently 20 years old. He's a high school graduate. And while he does live with Duchenne or DMD, he definitely doesn't let that stop him from having a full life and doing the things that he loves to do. He's a rock music lover, just like his mama. He's a movie buff, a baseball fan, and he enjoys museums and all sorts of community activities in our city. And as a family, we go to many concerts, movies, and baseball games, so we live a pretty full life here.

Mindy Henderson: Fantastic. And Amy, how did you first hear about PJ's Protocol? And what role has it played in your son's treatment journey and medical safety?

Amy Aikins: Well, it played a very important role, and I'll talk about that in a little bit. I first learned of the protocol from Parent Project Muscular Dystrophy or PPMD, that's now been mentioned a couple times by both Brian and Dr. Mendell. And on one December day, when Elijah was 13 years old, we actually ended up having to use it. So at 13, he fell walking down our hallway, tripped over carpet. And his falls are a common occurrence in Duchenne, especially when our boys are slowing down. Initially, it didn't seem like a big deal. We thought, "We'll just rest. He'll be fine and bouncing back by tomorrow." Well, that's not what happened. Several hours following the fall, he was still in a lot of pain and he wasn't able to bear any weight at all. He felt hot to the touch, and he started to stop making sense when he was speaking, appeared to be passing out mid-sentence. Sound very familiar, just like Dr. Mendell was just mentioning.

At that point, I feared a fracture, as well as FES or a fat embolism syndrome, which can be a life-threatening emergency as well, that results from a fracture. So at that point, we headed straight to the emergency department. We lived in a rural area, so we self-transported. Because we knew we'd have to wait a decent amount of time for an ambulance and we didn't feel we had that kind of time. Upon arrival at our local emergency department, in that rural community, he had a very high temperature. His breathing was very fast and his heart rate was extremely high. His pulse ox, which monitors blood oxygen levels, was in the mid to upper seventies, and for reference, the normal range is 95 to a hundred. So obviously, that is significantly decreased. He was immediately put on oxygen, blood work was completed, and X-rays were taken. And after the x-rays were read, just as I expected, they informed us that he did have some femur fractures. We later learned that he had three fractures in his femur. At that point, I requested an emergency steroid dose multiple times. The doctor at the rural hospital refused.

At that point, he admitted a transfer to the hospital where our clinic was. And I also put a call into Kathi at PPMD, who really helped me along this process, because this was rather sudden. During the two hour ambulance ride down to our clinic hospital, I was able to obtain a digital copy of the protocol. Upon arrival to the emergency department, I told them that he needed the stress

dose of steroid and that the rural hospital had refused to do so. I also told them of my fears of a fat embolism syndrome, also known as FES. I provided them the information on PJ's Protocol, and they administered the stress dose.

I will say, following that administration, it was pretty much like night and day. His stats and appearance vastly changed for the better, very, very quickly. And that FES, just as a side note, was diagnosed on day two of the admission. Over the course of that hospitalization, I shared the protocol with as many physicians as I could. Some of them had even taken down the link, so they could review it again for themselves and they would have the protocol if they would ever need it again. I will say, had it not been for PJ's Protocol, I really feel like our outcome could have been substantially different. So I'm very thankful.

Mindy Henderson: Amazing. Thank you so much for sharing that story, Amy. And as a mother, my heart goes out to you and having to live through that, but I think your son is incredibly fortunate that you were his mother and advocated for him in the way that you did. Dr. Mendell, clearly, parents of children with DMD need to have access to the protocol to provide to doctors, but is there any other advice that you can offer parents about preparedness, whether your child with DMD experiences a medical emergency, the type of information you should have prepared and ready or if you're working even with first responders or paramedics?

Dr. Jerry Mendell: So it's a tough issue, because you can't tell people what to do. And it's frustrating to do that. But I will give advice for this circumstance. And the advice is that the parents of muscular dystrophy, of Duchenne patients, should have the protocol handy. They should have a copy of the protocol. That means that they'll have the protocol in their home, and I would advise that it could be put in a folder and put in their car, put in the glove compartment of their car. So that there are circumstances that both Amy and Brian described where the necessity for encountering doctors, emergency doctors, providers, who aren't familiar with the disease and have no idea the importance of long-term steroids, of long-term care when there's withdrawal at risk, so I would also recommend that they carry a card with them that shows the internet link. So that it could be like a credit card in their wallet, if they go to the emergency room.

And under both the circumstances that were described by [inaudible 00:30:23] and Amy, they could show the first encounter doctors that are not familiar with that this is a necessity, the stress causes the adrenal insufficiency, and we need to have replaced the steroids. So these are issues that I think can best be handled by parents' awareness. And I don't want to put more stress on the parents, because this is the very difficult disease to deal with on a day-to-day level.

Mindy Henderson: Brian, turning tragedy into advocacy and creating a safer, better world for people with DMD has impacted so many families, like Amy's. It's kind of beautiful that PJ's legacy lives on in that way. Can you share why you feel it's important for people to use their stories to create progress and better care?

Brian Nicholoff: We've had webinars and conferences in various countries, in various organizations. As Dr. Mendell alluded to, the parents are the biggest advocate, there's no doubt. Parent's the one who has to tell the doctor this and that, who has to insist on it. And if things don't go right, then they need to get ahold of those that can get it done. Some doctors will say, "I don't need to know that," but then, they do. That's when you kind of put the pedal to the metal. You really have to, and Amy did that. Otherwise, those things could have been different. And I've got emails and texts and Facebook posts in numerous ways, unbelievable ways, that were very similar to Amy. And they said, "I showed them this. I gave them this. I gave every nurse this. They have them with the teachers at the school. He's got a hanging card on his wheelchair. We've got them in our wallets, we've got them on the website."

Everything. They've got it covered. They've got it covered, because that's what can happen in that critical situation. And then, your mind's not thinking right anyway, you need to relay that information as soon as possible. So for that to happen in this protocol, that's why we advocate as much as we do. That's why I've been so involved with this over the years, because I know that it can save people's lives. And it has. And it has, and Amy's a perfect example of that with Elijah. I've got them all over the country. Matter of fact, in a couple of weeks, they're going to have every attendee, and they've done this at various conferences, every attendee, scientific physicians, researchers, et cetera, get the protocol in their attendee bag or whatever.

Mindy Henderson: Amazing.

Brian Nicholoff: It's been numerous, numerous times it's been flooded with information about this. So hopefully they use it. Again, it goes back to the parents. Number one advocacy is the family.

Mindy Henderson: Absolutely. And I think Brian, what you're alluding to is MDA has its clinical and scientific conference coming up in the middle of March. There are going to be almost 2000 researchers, clinicians, scientists, people from pharmaceutical companies. And like you said, PJ's Protocol is going to be included in their gift bags. The other thing I want to mention is that, in the show notes for this episode, we're going to make sure to put a link to PJ's Protocol. And I believe, like Dr. Mendell was saying, we've got, on the MDA website, another sort of emergency information card that you can download. So we'll put links to all of that for people listening to go on and grab. Amy, is there anything else that you would like to add about, you're such clearly an amazing advocate for Elijah, anything you want to add about the importance of being an advocate for your child?

Amy Aikins: Sure. So I believe that we, as parents, we have to be ready to advocate in multiple areas of our child's life. With Duchenne being a rare condition, as Dr. Mendell mentioned, it's not well known by the general public. And this advocacy, I think it really can take forms in multiple things. Everything from educating medical staff who aren't familiar with Duchenne, like I attempted to

do during the emergency department visit and getting down to our clinic hospital, to engaging with school staff, when maybe an approved IEP isn't being followed, to even an accessibility concern in your community, maybe there's no ramp to get into a restaurant. And I really think we can't be afraid to keep just pressing the issue when we know something's needed or change is necessary.

And then, I also believe that, by advocating and modeling effective ways to advocate, we're also teaching your children effective ways to advocate for themselves. Because let's be honest here, Duchenne is a condition that's going to require a lifetime of advocacy. And as a parent, you're not always going to be there a hundred percent of the time to advocate for them. So I think it's really important that children, even from a young age, learn to self-advocate as well for themselves, and then, as they age, increase the responsibility of self advocating.

Mindy Henderson: Absolutely. You nailed it. So Dr. Mendell, can you tell us a little bit about some of the newer treatments that are available to people living with DMD?

Dr. Jerry Mendell: Well, things are advancing very rapidly in Duchenne muscular dystrophy field, and we are very grateful for that. We're talking about steroids, so let's open up that discussion first. There's a new drug called Vamorolone. Vamorolone is what we call a dissociative steroid. That means that we separate now the anti-inflammatory component of the steroid, the prednisone, from the adrenal function, so that adrenal function is now preserved. And we can achieve the same benefit of prednisone, but not have the adrenal crisis, because the body doesn't shut off hydrocortisone or the steroid from it. And at the same time, it can do everything that Brian alluded to. It can preserve bone health. Bone health is very important, so we don't have kids who have multiple, multiple fractures. So that is one important development. Number two, we have another drug called givinistat. Givinistat is a drug that promotes muscle regeneration, and it prevents one of the complications of steroids and of the disease in general that we fear very greatly, and that is fat infiltration, fat replacing muscle and scar tissue replacing muscle. So givinistat has that potential benefit.

Then we have another group of drugs called exon skipping, and the concept of exon skipping is relatively simple in one sense. Of course, technically, it's complicated, but it means, if you have a mutation, these drugs can skip over the mutation. If they skip over the mutation, then dystrophin, the vital protein for muscle, can still be produced. And in the past, we have had three acceptable exon skipping drugs. Now, we have a new one called Viltepso, that skips exon 53. So the mutation is in exon 53. It'll skip over that. The problem with exon skipping is it can't be applied to all patients. It can only be applied to about 30% of the Duchenne population. Then we move on to gene therapy, which is something I've worked on for the last 25 years. And we now have a drug that's successful called Elevidys, which is successful in the treatment.

And it is beneficial in the sense that it can slow the progression of the disease. It doesn't cure the disease, but it definitely slows the progression, improves the

quality of life, and it's the best drug that we have for the treatment. Now, in that same sense of gene therapy, there are two more developments that could be very important that are improving on the gene therapy, that potentially improve on Elevidys. Elevidys is approved by the FDA and available to all patients who have Duchenne dystrophy. But these drugs are in clinical trial. One is produced by REGENXBIO, and it is one that allows for a slightly different form of the gene that's put in. The gene that's put in by REGENX includes the final component of the gene. The final component is called the C-Terminal. And the C-Terminal potentially improves gene expression and improves the quality of the dystrophin that's produced.

We'll have to see. It's in clinical trial. The second one is also in clinical trial, and it's produced by Solid Biosciences. And this is an important one, because it improves the delivery of the gene. And so, it changes the capsid of the virus, which is where you put the gene into the virus, and it can hold a different transgene. It can allow for greater transgene expression, in other words, dystrophin expression. So we have many advances now, and we're really waiting for these new drugs to be fully tested in clinical trial. And we'll continue to improve the picture. So things are looking very good, and as I like to say, this is helpful, if you've been around long enough, you can appreciate this statement, this helps Jerry's kids, if you understand what that means.

Mindy Henderson: Yeah.

Brian Nicholoff: Yeah.

Dr. Jerry Mendell: All right.

Mindy Henderson: Well, it's funny that you bring that up, because of course, MDA is celebrating our 75th year this year. And so, I think there are a lot of people out there who know that reference. Boy, I wish I could live inside your brain for a day, Dr. Mendell. It's absolutely fascinating, the things that you talk about. One question, obviously, each patient needs to be evaluated independently, but theoretically, some of the drugs that you mentioned or therapies that are available today, can they, again, theoretically, be taken simultaneously and used to address different pieces of the condition?

Dr. Jerry Mendell: Different pieces of the...

Mindy Henderson: Well, can they be taken simultaneously, I guess, it's really the question.

Dr. Jerry Mendell: Well, I've been in this healthcare provider business for more than 50 years, as we've stated a number of times. And I saw my first patient with Duchenne muscular dystrophy in 1969, when I was at NIH in a postdoctoral fellow. And at that time, there were no treatments. And I made a commitment to myself that I would do everything I could over my lifetime to make a difference. And in that way, I was involved and instrumental in bringing prednisone on the clinical

market, bringing exon skipping on, and bringing gene therapy on. And all these improvements have merit, but they fail in one regard, they don't cure the disease. We have to have treatments that cure the disease, and we're working on that. And those advancements are also taking place. So we'll never be completely satisfied until we have a cure for Duchenne muscular dystrophy, and we're on the way to achieve that. So I think that's where I can end my contribution to this. But I appreciate being asked to participate in this podcast.

Mindy Henderson: Absolutely. Thank you. And Amy, for parents of newly diagnosed children who may be listening, what advice can you share, anything that's helped you and your family along your journey?

Amy Aikins: I think the best piece of advice that I could share would be to find your tribe, so to speak, parents who are walking the same path. I actually have an amazing group of Duchenne parents that I interact with on an almost daily basis. And sometimes, I don't know what I'd do without them. We're spread through the country, but the distance doesn't matter. We lean on each other and learn about navigating the condition itself by sharing our stories and learning from each other.

For local resources, I'd suggest finding a group that supports the special needs folks in your community. And it doesn't necessarily have to be Duchenne specific to find community resources. These folks will help you learn about what services and supports are available in your area that you live in, such as Medicaid or Medicaid waivers. And if you can't find one or one just doesn't exist, there are a few Duchenne organizations who also assist with resource navigation. I guess, as a final, I'd also like to add to not get tied up on social media, comparing what other individuals and families are doing. A friend just recently reminded me that comparison is the thief of joy, and really just to keep in mind that what's right and what's good for somebody else might not be right for you and your family. And your family and your child will forge their own path, so just to keep that in mind.

Mindy Henderson: That's great advice. And Brian, I'd like to give you the last word. What would you like to leave everyone with today?

Brian Nicholoff: Well, first of all, Amy, we've met each other on a few occasions. It's always been with an organization and such, and I consider you a good friend. I know you're one heck of an advocate for the disease. You live it every day. You stated something very important as find your team, find your village, reach out, communicate. Conceiving plus believing equals achieving, and if good is enough, better is possible. And that's what Dr. Mendell says all the time. I want to reach out to you, one thing, Dr. Mendell, I met you in June of '87 at OSU. PJ was on the first prednisone trial. He had a second biopsy done through you, okay?

Dr. Jerry Mendell: Wow.

Brian Nicholoff: He started out at, oh, I don't know, eight, 10 milligrams of prednisone, ended up at 51 Deflazacort. We'd come there every quarter. It was 181 miles one way, with Wendy and with Linda. And you would say, and you didn't have anything but, "Keep it going, this and that," there was nothing to really hold your hat on, except the prednisone. And look where we are now. And that's what I tell people who just recently found out that their son's been diagnosed or whatever. "What do I do?" Et cetera. Find a group, find an organization. And like Amy said, because it works for you doesn't mean it works for them, whatever. You will get the feeling of that. Dr. Mendell, I've got to say that at '19 or 2016, I came by and saw you at Nationwide, and you said, "Come here." I went into your office, and you said, "Let me show you this picture." And it was of a young girl, about two years old, maybe less. And he says, "See this? Her head's floppy." And he says, "Now look it. It's not."

He says, "That's SMA." And he says, "I know," he looked at me in the eyes, he says, "I know that Duchenne can have a cure too. I know we can do this for Duchenne." So you're one of the most highly respected, knowledgeable individuals, humans that pioneer with neuromuscular disease that anyone could imagine. And I am more than, more than grateful and indebted you forever for what you've done for the community. Mindy, I've only known you a couple hours. What you do for Quest Magazine and what the advocate you are for disabilities, you're talking earlier about airlines, disabilities, and so forth, you're a total inspiration. You're a total inspiration. And I'm so grateful to each one of you for this opportunity. That's all I can say. I'm very grateful and very hopeful for the future, very hopeful for the future, not PJ's Protocol, because that's going to go on and on and on. That's going to keep on building. It's what we start now that builds up. Like Dr. Mendell referred to, there's a lot of things in the oven, that's going to come out baking, there's a lot of things happening. That's it.

Mindy Henderson: Well, thank you for that. That was a beautiful tribute to each of the guests that we have here today. And I would like to just reiterate my gratitude for all of you being here today, and I echo your sentiments, Brian, there is no community like the neuromuscular disease community. It's like nothing I've ever been a part of before in my life. And that all of the scientists, clinicians, medical professionals, I include in that community. It's, I think, one of the true gifts that comes from all of this. And so, thank you all for being here. I know that there are people out there listening who needed this information. And like I said, we're going to put all of the key links and things, resources into the show notes, so people can grab all the information they need. Thank you so much.

Brian Nicholoff: Thank you, Mindy, for everything you do. Appreciate it.

Mindy Henderson: Thank you for listening. For more information about the guests you heard from today, go check them out at MDA.org/podcast. And to learn more about the Muscular Dystrophy Association, the services we provide, how you can get involved, and to subscribe to Quest Magazine or to Quest Newsletter, please go to MDA.org/Quest. If you enjoyed this episode, we'd be grateful if you'd leave a

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