

Living with PKU: Patient Approaches to the Management of PKU

Webcast

June 19, 2008

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Introduction

Andrew Schorr:

Hello and thank you for being with us once again for the third in our three part series, special edition series on PKU. I don't think there has ever been a series of programs like this. I'm delighted to play some small role thanks to the team that has made it possible. Remember back on May 8th, so this has all gone pretty fast, we did a program in understanding PKU and the new treatment options, and if you haven't heard that I urge you to read the transcript or hear the replay. That discussed the new world now that there's the first approved drug, Kuvan, added to the medical foods and the formula to help people better manage their Phe levels for PKU, and then it was just on May 27th we did a program on the assistance programs to help all the people who need it get assistance in having access to Kuvan, and also as you heard there are also some new assistance for the medical food as well. So we've covered that, and we urge you to listen to that.

Today we're going to talk about living with PKU and patient approaches to the management of PKU, and we have a wonderful array of guests. I want to introduce them to you. I'm really excited. So first of all let's meet somebody who is living with PKU and actually flew from his home in Toronto. He's just 21 years old, John Adams, Jr. flew down to make a presentation to the big biotechnology conference, an organization called BIO having their conference in San Diego, California, and John Adams spoke about how he has lived 21 years with PKU, and now with the help of biotechnology and the new drug Kuvan he is better than ever managing his Phe levels. John, thank you so much for being with us.

John Adams, Jr.:

Thank you so much for having me.

Andrew Schorr:

So you're up on the 10th floor of a hotel. Your dad came with you, and it is really very much a family story managing PKU. So John Sr. is with us, and he is way down in the lobby. John Sr., thank you so much for being with us too.

John Adams:

This is a treat, thank you Andrew.

Andrew Schorr:

We are going to talk about the family issues, and of course as you go on with PKU it is a long-term relationship with a metabolic disorder expert. One of the people who has done that now for over 12 years is up the road from where the Adams' gentlemen today in San Diego, up in UC Irvine, University of California Irvine, is Debra Hook who is a metabolic dietician. Debra it's a journey isn't it over many years as you work with little babies and families and teenagers when things can be difficult certainly with a special diet and then adults too. You work with all of them, right?

Ms. Hook:

I work with all ages. So I work with them once they are diagnosed as an infant, and I see them all the way through adulthood, and I like it because you get to see all the changes, and that they have different interests, different topics, different things that are important to them at various stages of their lives, and it makes it fun for me.

Andrew Schorr:

Well we are going to talk about that. So it is one big talk show in the PKU community worldwide today talking about living with PKU. Now someone who has been living for 37 years with it is Sarah Foster. Some people know Sarah Foster. She is very active on the Internet and in listservs, and she didn't join us live, but she did very recently on tape. We're going to hear from Sarah a couple of times along the way. She is very inspiring, and I'd like you to hear a little bit of her interview now.

Sarah's Story

Andrew Schorr:

I think it's really important to meet someone who has lived with PKU for many years, starting as a kid, and then has the adult wisdom, if you will, to reflect on this whole experience and give some advice and inspiration to others. Such a person is Sarah Foster who is 37-years-old and who is traveling for work today. She joins us today from Lexington, Kentucky. Sarah, thanks so much for joining us.

Sarah:

My pleasure, Thanks for having me.

Andrew Schorr:

So Sarah you're on the diet now and you are really good about it, but you weren't for about 16 years, but you were of course with your parents on top of you through age 10, and then resumed again at age 26. So you have been through the struggles of trying to manage your Phe levels with diet and formula, and I know with the website

you've put up www.PKUworldlink.org you really have reached out to a lot of people. So let's get some advice from Sarah.

So first of all, for kids and parents it's not easy is it, and there are a lot of struggles going back and forth. Any advice for them, and then when you reflect on yourself as a kid on how to really acknowledge you're dealing with PKU, it's a lifelong condition, but then go on with your life.

Sarah:

Sure, you're correct, it can be extremely difficult. I think it varies from person to person. You know we're not cookie cutters as I know you know, and some people are able to manage it just swell, and some people really struggle. So I think it's important for people to find a good place and be able to decide what's best for them and to deal with it. I think it's important just for people to deal with PKU because I spent a lot of time as a teenager being angry and not dealing with it. So I think it's important for them to deal with it.

Andrew Schorr:

Is counseling sometimes helpful or certainly support groups, and you link to a lot of people online, for parents and for kids? Do you recommend that that communication being so important?

Sarah:

Oh Absolutely. I think one of the reasons why I set up the website is because I felt so alone as a child. I mean the wonders of the Internet is I can talk to friends from around the world who have PKU, and anything that can keep you informed and educated about what's going on and to help you feel less alone whether it is going to a conference or meeting a friend for dinner. I mean I have really fantastic friends, and a number of them just happen to have PKU, and we've made really strong bonds. So I think any support that you can get does wonders. I have friends who are parents of people who are children who have PKU, and they're some of my best friends. So you can connect not only with people who have PKU but also the parents.

One of the weirdest things for me was one of the conferences I went to, and some friends and I were sitting around the table and I was about 26 at the time, just returning to diet, and there was a family sitting at the table with us, and the person we were talking to was younger than myself, and she was the parent of a new baby with PKU. So that was quite a generational change if you will. So it took a little bit of 'Oh, I'm old.'

Andrew Schorr:

Well you are not old, believe me. I know what old is like. Let me mention that of course there is that PKU teen-adult conference that is coming up I think August 15-16, this year 2008 in Chicago. So I want to urge people if they can to go to that.

Now how do you make it work over the years with friends, going out, etc. when you are trying to manage your Phe levels? How have you done that?

Sarah:

Well a variety of things. First I think it is always important to do your treatment whether it be formula or if you are taking Kuvan or whatever you are doing that's one thing. If you don't do it then your blood levels will not be good, but then as far as food goes sometimes I bring it with me or sometimes I just eat the food that is available to me naturally low in protein. It can be a challenge, and it takes a lot of planning sometimes.

I'm a big college hockey fan, and they don't exactly have low protein foods in the hockey rink. You know a lot of times where you are going to be able to find low protein things or where you are going to be finding high protein ones, and usually it's kind of a pain to go into a restaurant, but sometimes you'll be pleasantly surprised.

My friends and I went to the ESPN Zone before the game and found an appetizer that was low in protein that I certainly wasn't expecting. So sometimes you get pleasantly surprised. We can all have the unpleasant experiences at restaurants, so you just have to look at what's out there, and explore all your options.

Andrew Schorr:

Sarah you are 37. You are really a wonderful example of life with PKU. You have an advanced degree, a Masters degree, and you are having a successful career, but there are other adults who have been struggling in their life with PKU. Advice for them?

Sarah:

Well I think that if I could use, and I'm not going to say her name because I haven't asked for her permission, but I have a friend who has PKU, and I met her probably going back seven years ago now. She was a teenager then and was struggling really hard with it. Well she was doing the diet. She was kind of struggling, and I kind of became her friend and mentor, her older sister if you will, and one of the most touching things she did for me was she lives in a neighboring state to me, so she didn't live very close, and she called in to Casey Kasem and requested a long distance dedication on the American Top 40, and requested Mariah Carey's song she wrote, and she let me know it was going to be on the radio. She didn't tell me, she just said listen to the radio. Part of the line for the song says you know it goes on and says, 'A hero lies in you,' and I called her and I said you know, 'Thank you so much. That was really touching, but I don't consider myself to be a hero. I'm just me,' and I said, 'What you need to do is listen to the words 'that a hero lies in you' and kind of figure out what you want to do,' and hopefully I can encourage her to listen to this program because she has since gone off diet, and she is struggling. So I think you just need to kind of dig down in yourself and find that strength, and look to others around you, be it from your parents or professionals or whoever, and kind of just deal with it because

I spent, like I said, a long time not dealing with it, and it becomes a very liberating thing to be able to talk about it and just deal with it, whatever you choose to do.

Andrew Schorr:

We are back live. She is so eloquent. So John Adams, Jr., for you, you know that struggle, but then here comes a medicine Kuvan that has helped you a lot. Help us understand where for you that's enabled you, where the diet was difficult, and we heard it was very difficult for Sarah. How has it helped you, and how are you doing?

John Adams, Jr.:

I'm doing very well, very well with the new medication. Actually let me give you a little story of how we got the medication just to kind of give you an idea of how well it's working for me.

Andrew Schorr:

Sure.

John Adams, Jr.:

We went to a trip to a hospital in Chicago and picked up the medication from a doctor there, and I took the tablets. I had to take about 17 of them. I took them with food. We got back to Toronto the next day, and we did a blood test. We had actually done a blood test at the hospital in Chicago. So they measured my levels there and told me that they were 800 the next day, or actually just a little bit above 800. So they were very high. That was actually the highest it had ever been in my entire life. After I had taken the medication the following day we did a follow-up blood test. We got the results a couple of weeks later, and my levels had gone down to about 130. So my Phe levels went down 75 percent in about two days.

Andrew Schorr:

Tell us about since then.

John Adams, Jr.:

Since then it's been great. I've actually been able to have diet liberalization implemented into my life, and now I'm able to eat a multitude of foods that were not available to me prior to that. So obviously that would include high protein foods such as meats, eggs, dairy, nuts, legumes. So that's all open to me, and obviously I'm a super responder to this drug. Not every PKU patient is going to be able to have as much diet liberalization as I have, but I'm very fortunate to have that now because it has resulted in huge gains in terms of my physical stamina, my ability to concentrate in school, just my confidence level, self esteem and everything.

Andrew Schorr:

Well good for you. It's a wonderful example and you are at the Biotech conference of research really paying off here for a condition, a lifelong condition. I want to ask your dad, so John Adams, Sr., you hear this but also you are a part of a big group,

particularly in Canada, but I know you reach out worldwide. Here you have your son's story. Are we in a new world of living with PKU now where not just for your son but for many people it's a little bit easier for many people to manage those Phe levels because I imagine that number 800 is a scary number.

John Adams:

Yes, we have last seen the number anywhere close to that when John was about three weeks old, and just before we instituted the diet controls, and we are living in a new era, and for John, Jr. it has come along at a particularly telling time.

There was a time in his middle teens when he, I'll tell tales out of school here John. First of all he got I will describe it as a bit sloppy with taking the special synthetic amino acid formula. Mom and Dad did our best, but we just became boring nags, but then John fell in love with his first girlfriend, and God bless her soul, she took a keen interest in John's health. She reprogrammed him. She would say, 'Have you had your medicine today?' We would call it formula; she would call it medicine, and John would turn off the TV, get off the couch and get into the kitchen and mix it up.

So that helped John through his middle teens to be steady and reliable with the use of the amino acid formula, but dad had a feeling as he was in University, that he starting, he was still good with the formula but he was getting a bit sloppy with the diet, and Dad's hunch and you could tell by mood swings and things like that.

Andrew Schorr:

And girlfriends change too. That happens.

John Adams, Jr.:

Well I'm single now and I'm doing okay.

John Adams:

Okay, I had the feeling that John was getting a bit sloppy with the diet, and I guess that hunch was confirmed with that baseline blood test that the clinic in Chicago did before he started the Kuvan therapy. This is a fairly typical pattern as PKU kids grow up, particularly in their teens and young adult years. It's I think the available information and data is that most lose diet control, and so it is absolutely wonderful. We're here in San Diego as Canadians celebrating a wonderful part of the US healthcare system, which is the Orphan Drug Act, which is now 25 years old, and Kuvan is one of 326 brand new products approved by the FDA for rare disorders and with a California biotech company. We're here as Canadians to say that this is really, really important, and the same US legislation, which creates incentives for drug developers to take on new challenges for small markets, for rare disorders, orphan products. The wonderful part of this is the same company that developed Kuvan has now just started the first human trial for a second and different therapy for PKU.

Treatment Options

Andrew Schorr:

Right, that was mentioned with a lot of excitement with our expert from Stanford in our first program folks I made. So be sure to listen to that and you'll hear more. I want to bring in Debra Hook, our metabolic dietician. So Debra you've seen the struggles people have had, and the story of teenagers that is a tough one. I know I have a teenager, she was on a special diet, not for PKU but another condition and we had, what would you say, we had quite the stormy fights along the way. So I know you've seen that. Are we in a little better place now as you have additional options like Kuvan?

Ms. Hook:

I think so. That's the one thing I've noticed in my clinic is I have one more item to offer, and we have started everyone who wants to start Kuvan, but what we've seen is that it gives them a sense of hope. I think it allows the teenager or the young adult to feel a sense of control. You know they didn't ask to have PKU, and many times they think they lost control of what they get to, how they treat themselves during their childhood. So having one more therapy and having the young adult or teenager to say, 'I would like to try that,' I think gives them a bit of control, and it gives them some other options, and for those who are responders it really opens up their diet. The diet, although not impossible, I think it is difficult in the teenage years especially when you don't want to be different.

Andrew Schorr:

Oh right, amen. I mean you experienced that didn't you John Jr. as just there you are with your running buddies and you've got to be so conscious? That must have been difficult.

John Adams, Jr.:

Yeah, well I mean in terms of dealing with my friend groups it was pretty easy for me because I had a really good support network around me, which included my family as well. My friends and their families were also pretty good about it. They recognized my diet, and I told them what foods I would need to eat. Obviously my childhood and the early teens were not as smooth sailing as I'm saying here right now, but there were times where I would go to birthday parties or events where there was no food that I could eat, and I would basically starve. Then I would come home and try to eat as much as I could, but for the most part my friends and their families were really good about it.

Andrew Schorr:

Let me ask your dad. So John Adams, Sr. so we have some young parents who are listening and they are worried about that situation, or living it with their four-year old, or five-year old, and the kid wants to eat birthday cake and all this. How did you handle that? How did you communicate with the families? How did you explain it to your little kid?

John Adams:

Well you've explained it a lot to your child by the time they are four or five and at different levels as best they can understand it at different age levels, but yes those first few invitations to a birthday party for a young child's peers, frankly you had to go into crisis mode. You had to get a hold of the parents, the hosts, or the first sleep-over, and they had never heard of PKU, and they are startled and shocked and fearful frankly. First you have to get through the . . . most of the peers had never heard of PKU, and their first fears are, 'Oh my God, is this something like a peanut anaphylactic, life threatening possible risk?' Right? And you just had to develop those relationships and the information sharing and the confidence building, that it wasn't one of those kinds of situations. It was a manageable situation, but it did require constant care because it's someone you love dearly and brought into this world and you are responsible for every bite of food could potentially be a problem for protecting the development of their brain and their central nervous system. But you rose to the challenge. You did what you had to do.

Andrew Schorr:

Well we are going to talk more about that as we go on. Mary, a teenager from Pennsylvania has sent in a question to our e-mail address, PKU@patientpower.info. Mary wanted to know if John, Jr. is classical PKU how many exchanges did he take before Kuvan and now after Kuvan, any side effects from Kuvan? So John Jr. do you want to comment on that about exchanges before and after?

John Adams, Jr.:

Sure, actually I'll start out by saying in am not classical PKU. I have a mild version of PKU.

Andrew Schorr:

Okay.

John Adams, Jr.:

So my diet isn't even before I took Kuvan wasn't as restrictive as that of a classical patient, but Kuvan was actually was the first trial I went on. So I really hadn't tried anything else before that, and I'm hoping that I won't have to try anything after either because it's working so well.

Andrew Schorr:

Side effects?

John Adams, Jr.:

Side effects, there are none as long as I take it with food. The only side effect that I've ever had with it was one day I took it without food, and I had stomach cramps for about two or three hours. Other than that I have had absolutely no side effects. All the side effects have been positive for me.

Andrew Schorr:

Now let me ask Debra a question. Debra, so classical PKU, that's not what John, Jr. has. We are sort of in this early phase with Kuvan added to the available treatments to see who responds and how well, right?

Ms. Hook:

Yes we still are. It was thought that Kuvan, those who would respond to it would be more the mild, sometimes we use the term hyper-Phe, but we are finding that there are some patients with classical PKU that are responders. Those with classical PKU usually it brings down their levels, and we might slightly liberalize their diet, but they never completely go off medical foods and formula, but it does allow them a broader range of foods.

Andrew Schorr:

Right, which can open up a whole new world. Now also I think I want to make it clear that with John Jr. for instance with a Phe level of 800, and I don't know what Sarah's was when she went off the diet for so long, but that's what we worry about right? We worry about the toxic effects of it. So it's great, I mean liberalizing a diet or even John with a very liberal diet now, that's great, but we are really trying to limit the toxicity. That's what it's all about isn't it Debra?

Ms. Hook:

The main goal is to get your blood Phe levels, or phenylalanine levels between, I use milligrams per deciliter, two to four, you know within what we call the acceptable range, and that's our main goal. However you get there it's fine, and that differs between patients, but the main goal is to get rid of that toxic byproduct.

Andrew Schorr:

Now, John Adams, Sr., so you lead groups in Canada and communicate with people around the world. So we have new parents as I mentioned and some that are struggling, maybe struggling with their teenager. Nobody is walking down this path alone, and nobody is doing it for the first time. I mean you preceded them, and we have been dealing with PKU for many years. How do you help people calm down a little and know that they have support? Also we had a question from one of our listeners that said, 'Well I've looked in books and one thing says one thing and one thing says another, and I'm confused.' How can they get clarity, if you will?

John Adams:

We have a really important challenge here in beginning a strategy to address the somewhat different tactics and goals in different PKU clinics around the world. In Europe the different countries actually have different levels for treatment goals for children and for adults, substantially different from what is typically the case in Canada or the United States, but even within Canada and the United States there are differences in treatment goals from clinic to clinic. So we actually need to exercise the muscles of Patient Power to invite, I'll use polite language, our clinicians to have a structured conversation so that we actually can come up with much more crisp guidelines for clinical treatment goals.

I will use a specific example. We had exclusive interaction with one clinic in Toronto for 20 years, and our goal was, the equivalent there was, to use the more common American metrics somewhere between two and ten. We went down to Chicago to get early access to this new drug before it was approved by the FDA. It was John Jr.'s idea, and the first conversation we had my jaw dropped when the doctor in charge there said, 'No our goal here is between two and six. Well I had been told that it was between two and ten. Frankly as a parent, that bothers me that there can be that much difference in accepted clinical practice.

We've just heard the example of two to four as a goal and that's right, but two to ten, and two to six, or two to four? They can't all be best practice. So I'm actually throwing out on this Internet program that we have a challenge. I had an opportunity to have a conversation with the doctor, who is chair of the scientific committee for the European society for PKU, and he had completed a survey of the variances, trying to identify what difference there was in clinical practice across Europe, and there is a huge set of differences. Not just in the typical . . .

Andrew Schorr:

Okay, I can see our next series coming on Patient Power and trying to get everybody on the same Patient Power page. John we are going to take a . . .

John Adams:

I'll just wrap up very briefly. The US National Institutes of Health in the year 2000 had a professional consensus develop in conference, and out of that the fabulous recommendation came "diet for life" because even that used to be a variable practice from clinic to clinic. We need, now that we have a real new tool in the tool kit for dealing with PKU, and we have the prospect of another drug coming along in human trials, we need to revisit the professional consensus. That's my parental opinion.

Andrew Schorr:

Okay Dad, I agree with you, and as I've been learning about PKU I think it's about everybody feeling part of a community. We are going to get some answers. I know we have a question from Susan that came in wanting to know more about struggling, and what does that mean, and how do you get through that struggle. So we are going to

talk all about this. We have a brief break. We'll be back in just a second as we tackle the questions and try to help everybody be smarter patients and parents. We'll be right back.

Listener Questions

Andrew Schorr:

Here I am back with our live webcast, a third in our special edition series on PKU. Thanks to BioMarin for an educational grant so we can do this and really help people understand how to support each other as you deal for yourself or a family member with PKU. Here's a question we got in from Susan. Susan said when adults mention struggling with being off the diet like Sarah did earlier, what does this struggling entail? 'I'm a new mom to a five-year-old son and am trying to look forward to his liberal future as a hyper-Phe.' Debra could you comment on that? What does struggling look like and hopefully that won't be the struggle ahead for Susan's son.

Ms. Hook:

Well I think traditionally, and Sarah probably had the struggle that we used to keep kids on diet up to about age 10, and we took them off. So then they ate meat and eggs and milk and ice cream and cheese and they learned to like those items, and then about probably somewhere eight years down the road we realized that it should have been diet for life.

So I think the really big struggle, or the biggest struggle I see, are those individuals that are in their 30s that were our first cohort of individuals with PKU that were taken off diet from probably about age 10 to about 18. Nowadays what I see as a struggle is you know those teen years, the rebellion, and depending on how the kids rebel and how the parents allow rebellion because rebellion is healthy. So I always tell parents instead of making the diet the place where the child rebels, allow them to do something else, you know dye their hair, paint their room black. Do something where the child can rebel and be a normal child and not make it about the diet.

I've seen a lot of success with that, whether it is PKU of another condition, but I think what's hard and is a real gray area, and I can feel for John Sr. is that you get different messages from different clinics and depending on some clinics still challenge, they'll keep pushing more and more Phe to see what the individual does. Our clinic tends to be more conservative because we are always worried that someone will taste a food, like it, and then not be able to give it up.

A personal example of mine is I went for a limited time a vegetarian and I missed bacon. It was bacon that brought me back to eating meat, and you know what I worry about with my patients is as we introduce new foods is making sure that it's not a food that I introduce and then I have to take it away. I think that's the biggest struggle.

Andrew Schorr:

So I have a question for John, Jr. up on the 10th floor of that hotel in San Diego. It's beautiful and sunny out there. John, how did you rebel?

John Adams, Jr.:

Well I think my father mentioned it earlier. I started to veer off diet and there were times when I'd get tired of seeing my friends go out and eat certain foods, and I'd have to go look at the vegetable menu or something like that, or look at other foods that I could have. So I kind of started to I guess experiment. Not being on medication, that was not a good idea, and it resulted in higher Phe levels. So I mean in retrospect I wouldn't have done it. With hindsight I wouldn't do it again, but I mean that's how I rebelled was going off diet.

Andrew Schorr:

You were a teenager. I've got a couple of them. I know what it's like. So now we had a question that came in from someone from Australia, let's see Debby from Sydney. Australia, and she plays softball, and she is trying to understand what's the best way to incorporate the appropriate diet living with PKU and her needs, her nutritional needs as she is playing vigorous softball. Now you are an athlete too I know John Jr. So how do you do that?

John Adams, Jr.:

Well I mean thanks to Kuvan it's made my athletic life a lot more easy because I have a lot more stamina, I'm a lot stronger than I was before, and I'm just allowed to do more things. I can take in more calories since I can have foods with higher sources of protein. They also have a higher amount of calories. So I'm a pretty large guy, I'm 6'3" and almost 200 pounds so I require a lot of calories in order to get through the day because I remain pretty active. So I mean the best way to do it is make sure you get enough calories. What I did was research the foods, look at the nutritional information, find foods that you can eat and you can digest properly that have a good source of calories so that you have the energy to go out and do these activities that you like to do like softball or whatever it happens to be.

Andrew Schorr:

And you lift weights, right?

John Adams, Jr.:

I'm sorry?

Andrew Schorr:

You're a weight lifter.

John Adams, Jr.:

I do that. I try to do that four days a week, and I try to play sports at least twice a week.

Andrew Schorr:

We're going to get you a girlfriend out of this. That old girlfriend left.

John Adams, Jr.:

I don't have time for that right now with all the stuff I'm doing.

Andrew Schorr:

What did we say 6'3", 200 pounds, working out with weights and probably headed for the beach there in San Diego. A walking Canadian Adonis. Okay we are going to come back to you. We'll get your phone number out there.

Okay, well let me ask Debra something. Debra, so these kids as they grow up and I'm sure become handsome like John, they are on a journey and their needs for calories change over time or the sports they are doing or their growth. So that's really a lifelong relationship with someone like you isn't it?

Ms. Hook:

It is. Usually they know their dietician very, very well. I always tell people if you don't feel your dietician is listening to you, or any of your healthcare team, that you need to move on and find someone else because usually out of the healthcare team you know the physician, but for those with PKU they know the dietician very, very well.

Andrew Schorr:

Alright. Now we got a question in from Barbara from Medina, Ohio and she says, 'Where is a good place to find the phenylalanine content for common foods? I'm wondering how much is in things like Goldfish, Honey Maid graham crackers, Nutri-Grain Bars. Is it clear on the label, or how do you know?'

Ms. Hook:

There are a couple of tools. It's not clear on the label. The US labeling law only dictates that protein is listed and if it is half a gram or less they can list it as zero grams. So you can have something that says zero grams of protein but it actually maybe has a third of a gram and it does have phenylalanine in it. It depends on kind of where the family is in their life. If it is a young child I refer them to the low protein list by Virginia Schuett, and they can get that on the web, www.pkunews.org. If they want like a computer program there is a free one that's www.nutriciandata.com, and it ties into the US database, the USDA database and so it has very accurate information. You can put in banana and it will tell you everything that is in banana, and that's the best one.

For the phenylalanine level you are better off going to www.nutriciandata.com. If you are counting grams of protein then the easiest one is www.mypyramid.com, which again is done by the USDA.

Andrew Schorr:

We have a worldwide audience. This question came in from Mandy in London, England, and she wonders are children with PKU more prone to febrile seizures? That's part one, and the other one is how quickly do the Phe levels rise and fall after inadvertent ingestion of something high in protein?

Ms. Hook:

Children with PKU are not more prone to febrile convulsions, and then the second part of the question which is how high and how soon does the phenylalanine rise and then come down, and we don't exactly know that. You know you would have to . . . there haven't been really good experiments where we are doing continuous monitoring. When you eat something, a food load, it takes about a half an hour, it gets down to your small intestine and you would have an increase in phenylalanine. We tend to have patients measure their Phe levels about three hours after ingesting food because then we can see what we call the low point, or the trough. You know we talk about peaks and troughs, how high it goes and how low.

Let's say your child inadvertently went to a party and ate a hamburger or ate quite a bit of a hamburger; a huge phenylalanine load. Probably that would take a day to clear out. You know the phenylalanine would peak and then it would spill over and you would urinate it out, in I would say a good 24 hours. What you will notice if it's young child is they will be cranky; they might have temper tantrums. If it is an older child what older individuals tell me is they don't feel well, or they feel like they have a headache.

Andrew Schorr:

John Sr., you know people listen to this now. Fortunately, hopefully everybody has or can have a relationship with a metabolic dietician like Debra who is an expert at this and has been for over a decade, but it still can be crazy making with an emotional overlay for a parent. You know trying to get everything just right, going by the numbers and then this behavior issue that she just mentioned. When is it a kid being a kid or their personality, and when is it Phe levels out of whack? So how do you counsel people on that John?

John Adams:

Well patience. I guess we have in our family, my wife and I we had a slight advantage in that John was our youngest child. We at least had lived the experience of raising his older sister through those phases. I think this would . . . my heart goes out to a parent if their PKU child is their first child, because then it becomes even more of a challenge.

You need to develop your social network of support, and some people need to do that face to face. Other people can do it perfectly adequately through Internet connections. There is quite a wonderful PKU support listserv, that there is no charge.

All kinds of range of people are on it. All kinds of issues are discussed. All kinds of needs for advice and opportunities for offering advice and experience are there.

Andrew Schorr:

That conference coming up sounds great too, the conference in Chicago.

John Adams:

And the conference. The face to face to meet others who have walked this path or are in the same point in the life path, this is, have to use the tools that are available to us to make sure it's not a lonely voyage.

Andrew Schorr:

Very good point. Now here's a question that we got in. Paula is a mother of a teen with PKU. She is in Pennsylvania, and she wants to know, and maybe this is best for Debra, 'Do you see blood testing for people with PKU different in the coming years where you can know the level within minutes compared to mailing in the test?' That's confounding for parents and then not knowing for maybe a week. Are we going to get better or some kind of home testing for that Debra?

Ms. Hook:

I think somewhere within the next five to ten years we'll have home testing. There is, I think it is in Japan, they've done testing of something that would be like a glucometer, and I think now it has too much air in it, and they are cleaning that up, but the future definitely will be home monitoring which I think will, because you will have a closer relationship between "I ate this" and then "this is my corresponding phenylalanine level," and patients will start to understand that connection and feel that there is a connection.

I think that will lead to more, I hate to use the word "compliance," but it will lead more towards people saying, 'Aha, when I eat this that's what it does to my body,' and if they don't like that then they can manipulate that meal. So that's our hope is that we will have home monitoring, and I know that there are a couple of companies working on that.

Andrew Schorr:

Now John, Jr. so you know from the readings that Kuvan has been controlling your Phe levels better than you were ever able to before. Can you tell how you feel?

John Adams, Jr.:

Well I could tell the following day after coming back from Chicago and first taking the drug. I told my father, because he asked me how I was feeling the next day, and I said that I felt more sharp and more clear headed. I noticed actually in the coming weeks that I had a lot more physical stamina; I had a lot more ability to focus and concentrate in school, and actually my marks improved. We found at the end of the school year that my marks had actually gone up as well, and so it just made me feel

better in every part of my life pretty much. It gave me a lot more confidence in my physical activities, in my social interaction with people in school. So I just felt sharper, and I felt a lot better and more confident.

Andrew Schorr:

Okay, so now we know you are handsome and you are smart. We're going to get your phone number out there.

Alright here is a question; I'm going to address this to your dad. John, Sr. so Stanley from Pleasant Lake, Indiana wrote in. 'How can we as grandparents of a PKU grandchild feel that we can safely care for our granddaughter if and when we are asked to baby-sit? We can only see her a few times a year because of the long distance of travel. What are a few basic tips you can give us to ensure her safety?' It sounds like they are pretty worried, walking on eggshells a little bit. Any advice for them John?

John Adams:

Well there are lots of wonderful resources thanks to the Internet, and you do, grandparents as well as the parents and extended family, need to say, 'Okay, I want to understand this enough not become a scientist or researcher or a doctor, but just enough so that I actually know with some confidence that I am able to cope.'

You need to educate yourself on what foods and drinks are high or low in protein and therefore high or low in the phenylalanine. You need to understand that it's a liver enzyme defect, and you can't push that child's body beyond what its ability to process that particular amino acid. It's not the sort of thing that if you can't recover from a mistake at one particular meal or snack. You don't have to beat yourself up and feel guilty about that at all, but you do need to learn from that, and hey, it's actually quite exciting to say that I want to understand this so I can manage it intelligently.

Andrew Schorr:

Right, very good advice. A couple of questions for you Debra; so quick ones. So first of all there is Debby where she is from. She says, 'I'm 32 and have PKU and trying to start a family with my husband. I was seeing a doctor and then he transferred and I feel abandoned. I'm worried about keeping my Phe levels low for conception. Is Kuvan a safe positive way to do this or is it harmful for the baby?'

Ms. Hook:

That's not known yet. There have been a couple of women who were followed by a Dr. Koch, who is in southern California, and they were pregnant, and he gave them very low doses of Kuvan. BioMarin has a registry for those who . . . because we know there is going to be females on Kuvan who will get pregnant, and they will spend maybe the first month at least on Kuvan, you know the first month of pregnancy while on Kuvan, and I foresee in the future that there will be a study where someone will take on

looking at how safe Kuvan is during pregnancy. I think and this was said in an earlier show is that the most toxic thing is the high phenylalanine levels. That is the worst thing to have for a fetus.

Occasionally I'll have women who have not been followed for one reason or another and they don't have a doctor, a geneticist, they don't have a dietician and you know just getting on the internet, getting on a listserv and saying to people, 'I live here. Where is the nearest clinic?' I think getting in touch with the nearest clinic, and if you are pregnant telling them you're pregnant so you can be seen in the next couple of days; if you are thinking about a pregnancy making sure that you can be seen within the next month.

Ideally, although most of us don't plan our pregnancies, we get pregnant accidentally, is ideally it would be nice to be controlled, have your phenylalanine levels within acceptable range for about three to six months before you get pregnant. Most of the women I work with that's not how it happens. They end up getting pregnant and then trying to put the diet back together and getting back on to diet. It's doable I think just making sure that you put the word out there, calling people or getting on a PKU listserv and asking people where a clinic is so you can transfer your care.

Andrew Schorr:

Okay, good advice. Here's a quick question that came in from Heather in Pueblo, Colorado. She says, 'I have recently been told that having PKU is also having a type of developmental disability. Is this true?' I know people are throwing things at their computer. Debra, you're the clinician. Any comment on that?

Ms. Hook:

Well PKU is not. I think where that comes from is prior to newborn screening, when you had untreated PKU you got mental retardation, so you would become developmentally disabled, but now individuals with PKU have normal lives, normal intelligence, and so the connection between developmental disability and PKU has been broken for the most part. I do have patients with PKU who are in special reading, special math, but it's hard to say were they going to be there with or without PKU.

What I find though with a lot of patients is that you get on the internet and unfortunately there is a lot, and it's hard to tell whether it's new information or what is old information, but in California we don't consider PKU a developmental disability. It's an inborn error of metabolism so it's a problem with a pathway, but I don't consider it a disability.

Andrew Schorr:

Now we have a wider array of tools. So I want to find out from John Jr., how do you look at the future? So you have a medicine that is working for you. You and your dad seem to be getting along great. You have a good future ahead of you. How do you view the future living with PKU?

John Adams, Jr.:

It's more bright to me than it was six months ago or seven months ago because I wasn't on this drug, and I could sort of see myself veering off, and especially in retrospect now, I could see myself veering off of the diet as I got older. Now that I have this drug it's really allowing me to achieve my goals in terms of what I want to do, and how I feel at school, my performance there; my performance physically, how I feel like in the gym and all that. So I mean it is a bright future ahead for me now. It's great to be able to eat certain foods that I wouldn't have been able to eat and I'm hoping that I'll be able to stay on this drug for a long time.

Andrew Schorr:

Right, and there is more promising research coming, and how about from dad's point of view. Here is your third child, right? What's your view of a long, happy successful life for him now with this additional support he has in PKU therapy?

John Adams:

Well this particular new therapy has come along at absolutely the moment when it has made a real difference in John's life, and hopefully for others. We haven't mentioned this, but John was being tracked for bone density loss, and we think that was attributable to the PKU and the limitations of the diet, and hopefully that's not going to, with the results of the much better Phe control, and on top of that liberalization of diet in his case. Then I am looking forward to positive results at his next bone density assessment. So I'm looking forward to that, but previously that was a cloud on the horizon as well.

Family Relationships

Andrew Schorr:

I want to roll in another comment we got from Sarah Foster. Many people in the PKU community know her, and I was asking her about her relationship with her parents having been on this journey longer than John, Jr. She is 37 now. So let's listen to that because you know as you grow older we see things differently, and I think that is expressed beautifully in what she says:

Sarah Foster in one word when you look back on the struggles you had with your parents, do the diet, do the diet, their worry and concern and trying to be empowering. What do you say to mom and dad now after all these years, after you have sort of gone full circle, but now you seem to be in a very positive place?

Sarah:

I say I love you. I can't even imagine the struggles that they went through, but they were my . . . I put them through heck I'm sure, but they were a rock and when I was giving them trouble, you know I can use an example, well it's not a bad example, it's just an example of when one time I pretended I was sick and stayed home from school, and while I was home watching TV and stuff I was eating things I shouldn't, and my dad came home at lunchtime to check on me, and one of the things that I made that I wasn't allowed to have was I made toast and it burnt, and he could smell it.

They could see that I was struggling and you know my dad was like 'Sarah you have to play the cards that you're dealt,' and I was going through a really hard time and he used to play poker and I said, 'Yeah, but dad you play poker and you are able to give back some of the cards you're dealt, and life is not a game.' You just have to play the hand you're dealt and make the most of it. I think it's really important especially for young parents out there, you know everything that's been happening with PKU you don't know the past and nor do I expect you to, but I think having an appreciation of the past is also important. You know one of the favorite quotes that I have of Dr. Fullings is that "knowledge leads to humility," and I think it's important to have perspective that my grandmother, my paternal grandmother Elaine is 93, and if you can just think back to 2009 is the 75th anniversary of PKU so within my grandmother's lifetime PKU has been discovered, a treatment has been found, and it is not perfect but it mitigates the effects of PKU for the most part, and she has a granddaughter that hopefully she considers successful. You know I don't know of anybody with any disorder, regardless of what it is, who would trade that. I mean I think some people would be envious. So I would also say to my parents and professionals, I'm so grateful because I've met a lot of PKU adults who were not diagnosed, and it's very humbling.

Andrew Schorr:

Sarah you are so eloquent. I wish you just a great future...

Sarah:

Oh, thank you.

Andrew Schorr:

...in your job, with PKU and I hope you stay on sort of a soap box, maybe not one you've chosen but believe me one you have in inspiring others. Thank you so much for joining us.

Sarah:

You're welcome, thank you.

Looking Ahead

Andrew Schorr:

I love that lady, and she offers a great deal of hope. Debra Hook, are we in a new era of the management of PKU with the addition of Kuvan and other promising research?

Ms. Hook:

I think so because I think it has given us an option, and I think it's pushed and made insurance companies and others aware that there is not going to be one treatment, that you will be on multiple treatments or what we call adjunct treatments. For some people they are able to go off diet and just be on Kuvan like John. There are others who will have to do multiple therapies, and formula has really changed. We really haven't talked about that but there are different forms of it; there's bars, there's pills, there's low volume, there's modules and I think really we are getting to the point where we are telling insurance companies and those who are on public forms of health insurance that this is the norm. This is where we want to be, and you get more compliance, people have richer and fuller lives by having multiple therapies, and I think that the thought of something else on the horizon I think really is going to change how we view and how we treat PKU in general.

Andrew Schorr:

So John you've been living it, John Jr. John Sr. has too in living with it, but John Jr. so what would you say if we have teenagers listening, and we know Mary is with us from Pennsylvania and I'm sure others and parents, to just look forward to a full life in that while you were a very full responder to Kuvan, somebody else may try it, and it may be different for them, or it may be a mix of diet, formula or whatever comes down the pike as far as feeling that they can have a full life and to keep pushing in that direction?

John Adams, Jr.:

Well I think for any PKU patient whether you are on a special access drug or not you ought to put the impetus on yourself to monitor your diet. It's really important; especially important PKU patients to know what they are putting in their body and to take responsibility for that. You know you are going to go through your trials, and you may veer off diet, but as you get older you start to figure out that it's very vital that you really pay attention to your nutrition and that's what I would say. Do your research, educate yourself as much as you can, know what you are putting in your body, and your life will be better for it.

Andrew Schorr:

Amen, and let's talk to your dad. Dad you've worked hard to let people know they are not alone and so questions are going to come up. There are many that we couldn't answer today, but they still have a place to turn with the listservs and other resources. BioMarin is providing resources as well, and great providers like Debra, right?

John Adams:

Absolutely, and one of the great things is that there is a new actually Genetic Metabolic Dieticians, International's professional association for example. They have a website. There is information available there. We have a Canadian PKU and Allied Disorders Association. We're new but we're growing rapidly, and I am very proud of our website at www.canpku.org.

Andrew Schorr:

Okay, John Adams, Sr. downstairs in the lobby of the hotel in San Diego escorting your son, thank you so much and thanks for the work you do with rare disorders and in support in North America with PKU. John Jr. up there in the hotel probably lying on the bed, ready to go to the beach, thank you for sharing your story with us too John.

John Adams, Jr.:

Thank you for having me.

Andrew Schorr:

Okay, and Debra Hook from University of California Irvine, and I know you go around in the area and help people with these metabolic disorders, thank you for your dedication to doing it too. It sounds like we've got a great story coming, developing further and certainly for people who learn about a new drug like Kuvan, wonder if they can talk to you about it, they can find out about the other options, and as we mentioned in our second program there is support. There is financial support both in the drug company, the medical food people through the National Association of Rare Disorders. They've been helping too, so places to turn. Debra thank you for what you do.

Ms. Hook:

Thank you so much.

Andrew Schorr:

Just to wrap up then, remember we have done two previous programs; the first one heavy duty on treatment. That was our May 8th program. The replay and transcript is there for you on www.patientpower.info in the special edition section. Then we talked about access to care, all the insurance and reimbursement issues. That's there and now this one. We'll add the replay for this program probably tomorrow and then add the transcript as quick as we can. So it's all there for you. Tell a friend, and I hope we'll get to do more, but I want to thank you for participating in our PKU series. I like to say knowledge can be the best medicine of all. I'm Andrew Schorr wishing all our friends in the PKU community all the best.

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