Juvenile Huntington’s Disease

Health Radio
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Martha Nance, M.D.
Sandra Kostyk, M.D.

Introduction

Andrew Schorr:
Hello, and thank you for being with us again. Andrew Schorr broadcasting live from Seattle, and I love doing in-depth series’ on important medical topics where we can partner with a leading patient advocacy group, and everybody can hear about it, go to it live, ask questions, listen to the replay, read the transcript, and connect with world experts in a condition. Well, that’s what we’re doing for the next three days on Patient Power. We’re discussing once again Huntington’s disease, a very serious condition, and we are taking different bites out of the apple, if you will, as we go every day.

I want to describe what we’re doing, tell you how you can ask questions because we are live, and you can be connected with leading experts. Today were going to talk about juvenile Huntington’s disease. It’s very rare, but if you’re family is affected by it, it can be terrifying. You have many questions, many issues. We are going to introduce you to two experts. One of them is live with us, and the other is on tape but with some very important information, and then you’ll get to ask questions.

Tomorrow we are going to discuss Huntington’s disease in the prime of life, and we’ll be introducing you on that program to Dr. Karen Anderson from the University of Maryland, and then on Wednesday will be back with Huntington’s disease research. Everybody wants to know about that, and we’re going to have with us Dr. Joseph Jankovik who is from the Baylor College of medicine in Texas, and then also Dr. Jody Corey-Bloom from the University of California in San Diego, but today we’re going to tackle juvenile Huntington’s disease.

Now a reminder, we a few months ago we did a one-hour program with Dr. Jane Paulsen from the University of Iowa on sort of an overview of Huntington’s disease, and thanks for the help then and now from the Huntington Disease Society of America. That program is already on the Patient Power website, www.patientpower.info, but let’s go talk about juvenile Huntington’s disease.
I’ll mention who are guests are. In a minute, you will be hearing from Dr. Martha Nance, who has written extensively about it. She’s a neurologist and director of one of the 21, I think it is, Huntington’s Disease Society of America Center of Excellence. She’s at the Hennepin County Medical Center in Minneapolis, so you’ll hear more from her in a minute on tape, but now let me introduce you to Dr. Sandra Kostyk. Dr. Kostyk is the director of the Center of Excellence at Ohio State University Medical Center in Columbus, Ohio. Dr. Kostyk like Dr. Nance is a neurologist. She specializes in movement disorders and particularly in Huntington’s disease. People come from all over to see her and her team. Dr. Kostyk, thank you so much for being with us today.

Dr. Kostyk:
You’re welcome. It’s a pleasure to be here with you.

Andrew Schorr:
Dr. Kostyk, we’re talking about rare conditions; Huntington disease is rare to begin with and juvenile Huntington’s disease, rare. How much do we know about it now for families who are touched by it? Where did this come from? Maybe they were aware there was a gene in the family, and then they wonder well, what road lies ahead? How experienced are neurologists today with juvenile Huntington’s disease?

Dr. Kostyk:
Well, because the disease itself; Huntington’s disease in adults is rare, and the juvenile form is actually even much rarer; there are a lot of neurologists and primary care physicians who have never seen or followed a case themselves. It helps to have a center or someone who’s had at least some experience with this, but even a lot of the centers don’t see a lot of children with Huntington’s disease. The other thing that makes it a little bit hard is when you’re talking about children with Huntington’s disease, you’re talking about children with onset at age 3 or onset at age 20, for the juvenile form, and there is a very big range of variation in the way the disease presents even across those first 20 years of life.

Andrew Schorr:
So simply having the diagnosis doesn’t mean that that child’s case will be identical to the next child.

Dr. Kostyk:
No, I don’t think I’ve ever seen two cases of juvenile HD that were even close to each other.

Andrew Schorr:
Know that you’re working hard on research. There is a lot of genetic work going on trying to understand what’s going on. We don’t have a treatment yet, but can we help children and teenagers live better while they’re coping with Huntington’s disease?
Coping with HD in Youth

Dr. Kostyk:
I think we’re headed toward a point, or research is getting to the point where we have agents that may slow down the course of the disease, but a lot of what we’ve learned over time includes the good preparation, good planning, good support networks, certain nutritional interventions, and possibly even some of the supplements that we’ve begun to study in the adults may be applicable for use in the juvenile form.

Andrew Schorr:
It would seem that it’s very important for a family touched by this, and I would say the adult form as well as juvenile, that they connect with a center such as yours for two reasons. One is to have people have providers who are experienced with it in all aspects, the family issues as well as the individual patient’s issues, and second of all you all are actively doing research, and it seems so important that families partner with you to try to move that research ahead for their benefit and everyone else’s.

Dr. Kostyk:
One of the things I tell families who come here or whether it’s with the adult form or the juvenile form is that actually we’ve made a lot of progress in understanding the basic science underlying Huntington’s disease in the last 10 years, and there are lot of things in this disorder, unlike a lot of other diseases, where we are actually ready to go from the laboratory to the clinical trials, and because this is a rare disease were almost at the point where we have more agents that are worth starting clinical trials than we have people with the disorder to answer all the questions and to do the good studies to help the advancement of our ability to treat Huntington’s disease. The more people who can participate in clinical trials the better because were at that phase where we have a lot of different clinical trials, and we’re asking a lot of questions about the different agents; which is the best, which will help the most, which will help in Huntington’s disease.

Andrew Schorr:
I blogged about some of these issues that may be coming up on commercial television on my blog on www.patientpower.info. I noticed in the final episode of “House, M.D.,” and I don’t know if many of the listeners watch the show, I’m kind of a fanatic about it, Dr. Thirteen, who’s one of the younger doctors who works with Gregory House, that kind of curmudgeonly star, she I guess has Huntington’s running in her family, and Dr. House kept saying. You should be courageous, be tested. So one night in the lab all by herself, and I know doctors don’t normally get to do that to go in the lab by themselves, she draws blood, she runs it through the machine, and part of the season finale it’s positive for Huntington’s disease. So I know some of the questions we will get to later is if there is that in the family or if there is already one child with Huntington’s, how do you plan for that child, and what about considering other children. What do you do? So lots of genetic questions too.
Let’s go ahead and listen to our interview with Dr. Martha Nance, and then will be back but be sure to just listen carefully to this folks. There is very important information. Dr. Nance has written one of the very few juvenile Huntington’s books, and she’s highly regarded around the world as it is Dr. Kostyk, and then you’ll have a chance to ask Dr. Kostyk questions. You can e-mail your questions to questions@patientpower.info.

Here’s Dr. Nance.

Andrew Schorr:
Joining us now on tape is Dr. Martha Nance. For 20 years, Dr. Nance has been working on Huntington’s disease. She’s a neurologist, and she’s Director of the Huntington’s disease Society of America Center of Excellence at Hennepin County medical Center in Minneapolis. Dr. Nance thanks for joining us.

Dr. Nance:
It’s my pleasure.

Andrew Schorr:
Dr. Nance, let’s talk about Huntington’s disease, and then understand from your point of view where juvenile Huntington’s disease fits in.

Dr. Nance:
While Huntington’s disease affects somewhere between five and ten per hundred thousand people. Juvenile Huntington’s disease is said to be about 10% of the total people with Huntington’s disease.

Andrew Schorr:
Okay, so pretty rare. Now I understand that it is onset is usually when, around age to or when would it show up in a child?

Dr. Nance:
Well, that’s really all over the map. In adults, we can say that the average onset age is somewhere between 35 and 45, but clearly we see people on either end of that range including people whose onset isn’t until they’re 60 or 65. I have seen children as young as two whether symptoms began, but it’s probably more common I guess I would say in the teenage years.

Signs and Symptoms of Juvenile Huntington’s Disease

Andrew Schorr:
When someone is diagnosed as a juvenile, how does it show up? What does it look like?
Dr. Nance:
The main symptoms in Huntington’s disease are difficulty with movements, and the difficulty with movements often has two parts. One is difficulty doing movements that you want to do, in other words, poor coordination. In a child that might take the form of losing motor skills that the child had previously attained such as riding a bicycle or difficulty walking or balancing.

The second feature of Huntington’s disease in terms of the movement disorder is abnormalities of muscle tone or even the presence of involuntary movements. In adults with Huntington’s disease, one of the most common symptoms is chorea, the presence of extra wiggly, fidgety types of movements. Those movements may be present in a child, but the younger a child is when the symptoms began, the less likely the child is to have chorea, those fidgety wiggly movements, and the child is much more likely instead to have stiffness or rigidity or spasticity. So those are the motor symptoms, but the other two areas where you can see trouble in Huntington’s disease are one is thinking and memory or what in adults we tend to call dementia, and again in a child who gets tested frequently at school what you would see as a fall off in school performance from whatever level they used to be at, and then the third area that is quite commonly impacted in Huntington’s disease is mood. So depression or anxiety or irritable behavior or obsessive-compulsive types of behavior. All of those things are quite common in people with Huntington’s disease and may be quite common in children as well.

Andrew Schorr:
Is that the same illness as it is in adults?

Dr. Nance:
Well that’s a very good question, and it’s kind of a difficult one for us to answer. We know that everybody who has Huntington’s disease whether they’re an adult or child has the same root cause of the disease, which is a mutation in a very specific part of a specific gene. How big the mutation is, what tends to happen is that there is a little section of the gene that gets copied over and over too many times, and that section in the gene is called a CAG repeat. Normally the gene carries something between 10 and about 35 CAG repeats, but once that number of CAG repeats within the gene gets to be over 35, that’s the gene change that we associate with the development of Huntington’s disease. The bigger the gene change and the bigger the mutation is and the more CAG repeats a person has the younger the symptoms tend to start.

Diagnosing Huntington’s Disease

Andrew Schorr:
Now as you describe the symptoms, since most schools and most pediatricians would not at all be familiar with it personally, how does someone get to a diagnosis? At what point do you do a test when it could be as it starts out so many other things?
Dr. Nance:
Again, that’s difficult I think even for seasoned doctors to answer that question, and sometimes there’s disagreement among different specialists as to when do you rush in with the syringe and take a blood sample to do the gene tests. Most commonly of course Huntington’s disease runs in families, and in order to get the very large gene mutation that we associate with juvenile onset Huntington’s disease it’s quite likely that the person has an affected parent. Quite often for children with Huntington’s disease, it turns out that the affected parent is the father. So I think one thing that would really lead a doctor to suspect Huntington’s disease would be knowing that there is an effective parent, and then you look for this constellation of symptoms; the motor symptoms and the cognitive symptoms and the behavioral symptoms. I think if you see those things, if you see a child having a fall off and their ability to function in the classroom, after previously functioning at a certain level, I think you can be suspicious that it might be Huntington’s disease. Most of us again don’t want to be wrong making a diagnosis of Huntington’s disease, and doing the gene test by itself doesn’t prove that the person’s symptoms are caused by Huntington’s disease. So we tend to be cautious or conservative about ordering the blood tests.

Andrew Schorr:
With parents listening who may have heard or worry about this, how do they navigate the process to either find out that is the diagnosis, which would be scary, or hopefully rule it out? What process would you recommend for people?

Dr. Nance:
I think that’s the most important thing, well, there are several things that are important. One is to make sure that the child is otherwise healthy. So I think most parents with children who are having trouble start with their pediatrician and just have a general checkup and make sure that there is not an easily solvable problem. The type of specialist who typically sees children with these types of symptoms would be a pediatric neurologist. Many pediatric neurologists don’t have much experience with juvenile onset Huntington’s disease, so quite often families or even the doctor might seek out some help or a second opinion from a clinic or a specialist who really has an interest in Huntington’s disease or other movement disorders. The Huntington’s Disease Society of America has established a number of what it calls Centers of Excellence around the US, which serves as areas where both the clinical work and research in Huntington’s disease are going on. Typically they’re at university medical centers. It is not exactly one in every state, but I think that they have Centers of Excellence scattered throughout the country, and that I think can be a referral site for both families and also for the local pediatric neurologist who needs somebody else to take a look.

Andrew Schorr:
Dr. Martha Nance, all of us listening thank you for your devotion to families affected by Huntington’s disease and all your work in juvenile Huntington’s disease. I know you’ve written a book, “The Juvenile HD Handbook; A Guide for Physicians, Neurologists and
Other Professionals.” Thank you so much for being with us. We wish you all the best with your research. Dr. Martha Nance, who is a neurologist and Director of the Huntington’s Disease Society of America Center of Excellence at the Hennepin County Medical Center in Minneapolis, thank you ma’am.

**Dr. Nance:**
Well thank you so much. It’s been my pleasure.

**Andrew Schorr:**
We are back live now as we connect you with another leading expert in Huntington’s disease and another Director of a Center for Excellence, this one at Ohio State in Columbus, Ohio, and that’s Dr. Sandra Kostyk. Dr. Kostyk, the first thing is, from what you’ve heard Dr. Nance say that you all are trying to figure this out, not everybody agrees. Are there any other perspectives you’d want to share on some of those issues we discussed?

**Dr. Kostyk:**
I think Dr. Nance made most of the points of what our concerns are is one doesn’t like to jump to genetic testing or a miss another diagnosis. There are other things that might be mistaken for Huntington’s disease, and we usually recommend that people do start with their pediatrician if there’s a problem. Something simple like there is a disorder of thyroid disease that might present and have some similar things, which is more easily treatable.

My experience is that we tend to because again as she had said a lot of even pediatricians or pediatric neurologists have not had much experience tend to work with community pediatricians and pediatric neurologists to coordinate care in making these decisions for a family who is concerned about HD, and we do try and make sure that everyone is very well-informed about the pros and cons of getting genetic testing and have them meet with a genetic counselor to understand what it means to have the diagnosis.

**Andrew Schorr:**
Right, in that example that I made from the TV show “House,” I can imagine in next year’s plot line that young doctor has a lot of decisions to make on whether she would have children, what does it mean for her, as well as a patient should she become one. So it’s a very murky area for families. We have lots of questions we want to ask you, but I’ll just start with this one. It’s rare, and I know that typically people live maybe 15 to 20 years I’ve heard with Huntington’s disease; how do you predict how long the life of a child might have, and do you think as you have researched, I know there are no promises, that there could be something that would alter someone’s future in a positive way based on where research could head? How should someone view the future?

**Dr. Kostyk:**
I always encourage people to view it as optimistically as possible because you can’t predict when a cure may come or the data suggests that this is going to slow the course
of the disease. I think we have a lot of agents right now that have great potential for slowing the course of the disease, so that may change with the natural history of what Huntington’s disease is for each individual in the next five to 10 years. So trying to predict, well 15 years ago we had just found the gene, and now open where finding things we think may change the course of the disease, so it’s probably not going to be five to ten years ago the same disease it was before as we have more early interventions, as we come up with new treatment plants, and even new medicines to treat the symptoms; those have changed over the years. So I tend to encourage people to try and lead as normal of a life as long as possible and to be as optimistic as possible.

**Andrew Schorr:**
Right. We’re going to take a short break. When we come back we’re going to continue our discussion with Dr. Sandra Kostyk, an expert in Huntington’s, from Ohio State University Medical Center. We’ll be right back with much more of Patient Power.

Welcome back to our live juvenile Huntington’s disease webcast. Remember were doing a whole series. And thank you to the Huntington’s Disease Society of America website putting is front and center in there. So it’s today, it’s tomorrow and we’re going to deal with adults from Huntington’s, and then we’re going to talk about research in particular on Wednesday and then there will be a replay. After we finish each program usually several hours later or certainly by the next day, if you go to [www.patientpower.info](http://www.patientpower.info) there will be the replay. Remember we are earlier did a program with Dr. Jane Paulsen a couple of months ago, and that’s there as well as we have transcripts once we can get them typed up. Were visiting with Dr. Sandra Kostyk for the next half hour or so, and she’s at Ohio State University Medical Center. She’s Director of the Huntington’s Disease Center of Excellence of their, and people come from all over and of all ages to see her. So during the break, Dr. Kostyk was mentioning something that she gets asked all the time, and that is Dr. Kostyk, if there’s a family that has one child with a positive diagnosis the question is what, should we test the other children, and what’s your answer?

**Dr. Kostyk:**
It’s a very personal decision, but we try and respect both the family and the child’s rights in giving advice on this, and we usually encourage our families to talk in depth with the genetic counselors. In general, we discourage testing before there are any symptoms in an individual who is younger than 18 because the diagnosis can affect their future. It can affect individual self-image and self-esteem, which is particularly important I think and difficult for a teenager. It can affect, although we’re hoping that legislation will change this, but it might affect employability, it might affect insurance. It may cause someone to make a decision in their life to limit their academic career or social goals when even though they may be positive, they may not have a repeat size that is as large as their sibling who had early onset. Maybe they’ll have onset when they’re 40 or 50 and maybe by the time that child is 40 or 50 were hoping HD won’t even be an important factor in decisions in their life. Maybe we’ll have treatments by then, so it’s a very touchy issue that needs to be addressed on an individual and on a family basis, but in general because
we want to make sure that everyone has a good sense of things we discourage testing in an individual before they are 18 or 21 years old.

**Listener Questions**

**Andrew Schorr:**
Okay, thank you for that. Now as you can imagine people are listening today and many of them are touched in a big way. I guess there’s no small way when it comes to Huntington’s. Here’s an e-mail that got from Elysia in Mexico, and she gives a little background and then with the question. ‘I’m an American nurse who married a doctor from Mexico. We live in Mexico and have seven children. My husband had severe personality changes as he grew older having a late-onset HD around age 50. We finally got the diagnosis when my husband was 61 while in Houston where we met with Dr. Joseph Jankovik,’ who I believe we will have on the program too. ‘He died when he was age 72. Four of our children have the HD gene. One of our boys had an early onset when he was 30 and died at age 48. My two daughters have various degrees of advanced HD, and my other son who lives with us is age 48, and his HD seems to be advancing rather rapidly.’ So not good. Here’s the big question though, ‘What’s the chance of my other three children having a child with the HD gene?’ Can the gene skip a generation?’

So she has kids where it hasn’t shown up, and can it skip a generation?

**Dr. Kostyk:**
The easy question to answer “can it get the generation” is no. If an individual does not carry the HD gene, their children will not get Huntington’s disease and should not get Huntington’s disease. In terms of the other children who as yet do not show symptoms, and I presume they have not been tested, each individual child has the same chance of getting the disorder is any other. So basically each person whose father has Huntington’s disease has a 50% chance of getting it. So the odds do not change. If you had eight children, and you had four children who definitely had it, it does not mean that the four who don’t seem to have symptoms yet are clear free. It doesn’t mean that they have it. It means that each individual of the remaining children still has a 50-50 chance of having the disorder.

**Andrew Schorr:**
Now here’s a question a lot of people wonder about. Harold from Newport Vermont says, ‘Has there ever been a juvenile HD child who has grown into adulthood?’ I would think just from listening it depends on when the diagnosis is, it’s all over the map, but what about longevity?

**Dr. Kostyk:**
We don’t have a lot of data, but most of the data suggests that the children similar to adults live around 20 years after the time of the initial diagnosis. So if a child is diagnosed at three, again you might expect that they would live until 20 or in their 20s. It’s more common to have a child diagnosed between the ages of 10 and 20, and those children do
therefore progress into adulthood. So yes you can live into adulthood, but there is some suggestion that the younger the onset that there may be a little bit more rapid decline in the state and the stage of the disease.

**Treatment Options for HD**

**Andrew Schorr:**
Let’s talk about treatment either in trials or in standard practice now. So I know there’s no cure and as best we can tell we cannot modify the course of the disease. What about just assisting with some of the symptoms?

**Dr. Kostyk:**
That I think one of the keys here is because the disease can vary so much, and I think that you can sort of get a sense from the Huntington’s Disease Society of America website, where very much into the team approach. Because we have no one cure, we treat all the symptoms, and there can be multiple symptoms and multiple issues. We recommend that everyone has a primary care doctor. Dental issues can be a problem where people have difficulty with dental care, so sometimes you want a specialist who can deal with children particularly with disabilities who may not be able to cooperate with regular dental care. The teenagers in particular in my experience and even the younger children have had a lot of behavioral and psychiatric issues that the child psychiatrists may not be used to seeing so again we’ll work together with a child’s psychiatrist to choose medicines that are appropriate, and early intervention with physical therapy, occupational therapy, speech therapy to head off things before they can become problems.

With respect to school, each child needs individual assessment and plans, and often it helps to educate the school and as a center if there’s a child here and there’s a school nearby who is willing to participate, we’re willing to go out and talk to them about Huntington’s disease. There’s actually a resource available through the HDSA on school and educational problems with juvenile HD that includes a CD. Talking to the students classmates can be helpful so that they’re not scared of the individual with Huntington’s disease. They don’t isolate them and we can keep them active in social situations as long as possible. A nutritionist, and with regard to school, one of the problems is people with disabilities often have a static disease. They’ll have cerebral palsy, and they have a deficit, but it doesn’t get worse over time. One of the problems with Huntington’s disease and dealing with it especially as a child in school is that the disease may change over the course of the year, and within six months or nine months the child may have different needs than it had at the beginning of the school year when the child was assessed. So, frequent reassessments are necessary.

One of the things you have said about can we change the course of the disease. We’re actually doing research and clinical trials with a number of agents in adults at this point, and we haven’t really moved to doing them with children yet. On several supplements and
agents like coenzyme Q10 and creatine and actually even agents in fish oils that may, we have evidence at least in the animal models and in the basic science models that it may slow the course of the disease. So we have a lot of hope that we will be able to modify this disease in the next few years.

Andrew Schorr:
I sure hope so. I always like to give my leukemia example. So when I was diagnosed with my leukemia I started connecting in the early days of the Internet with other people and when we opened a book in the library it said “An always fatal condition.” It talked about the normal life expectancy was. But then came clinical trials. I participated in one, a phase-II trial, and now that’s the treatment that most people get, and knock on wood I’ve been in a deep remission in the eight years since having that treatment and no medicine. Now that illness is a whole different illness, but I think it’s an example of what we proved them wrong with the patients working with the researchers, so I wish that for Huntington’s.

Here’s another question we just got in. This is from Robert in Madison, Wisconsin, and it brings up the whole issues of genetic testing for people worried about this so here it goes.’ Before we got married there were certain tests that my wife and I had, but this webcast raises the question about Huntington’s disease tests prior to marriage. I know the onset is usually mid 30s, but are their advanced tests to see if there is a chance for the onset of this disease?’ So what about that in pre-marriage testing? I know I’ll just mention as an aside, my family is Jewish, and my wife and I had testing for the genetic condition Tay-Sachs disease, which certainly would be a death sentence for child in their early years, and we have a friend where that happened so we were tested. We were both negative, but what about for Huntington’s disease?

Dr. Kostyk:
I’m not sure I quite understand the question. This is I presume an individual who is at risk for Huntington’s disease?

Andrew Schorr:
He’s listening, and I don’t know if he’s just worried or it is in the family, so I guess we have the question of if it is in your family and your going to get married should you have a premarital test, but let’s also talk about it more generally. Should it just be part of tests that people have before they get married generally?

Dr. Kostyk:
That’s a very personal decision about what one should get tested for especially when there is a disease for which there is no cure and when you don’t know exactly what the age of onset of the disorder will be. As you recall when Dr. Nance was talking about the variation in the age of onset, although she and I have both seen children under five with the disorder, we’ve both also seen people who have not even started showing symptoms until they were 65, and I have someone who probably didn’t start showing symptoms until
he was 70. So do you want to change your life plans based on a disease which although there is a test for it, getting the test and saying okay you have a CAG repeat size of this number does not tell you exactly when you’re going to have the disease. So is that going to be really beneficial for you, or is it really going to be more detrimental to you? Again as we talked about it’s affecting your relationships with other people, your self image, the way people view you, employability. It’s a decision that requires, generally what we recommend is that the individual and their fiancé come in and spend an hour or two or three I have to admit on occasion in our center talking with our social worker, the genetics counselor, with myself about the pros and cons of genetic testing. It’s not as straightforward yes or no issue, it’s not everybody should be tested, and it’s not everybody should not be tested. The decision has to be made on an individual basis.

Andrew Schorr:
Okay, I can hear, and maybe the producers from “House” are listening about what the plot line should be for Dr. Thirteen as she has a positive result.

Dr. Kostyk:
She should have had genetic counseling.

Andrew Schorr:
Yes, right. Maybe there’s a new climate. We don’t know, and we should mention, and it’s on the Huntington’s Disease Society website too. President Bush just I think like a week ago signed a new Genetic Information Nondiscrimination Act. So our hope is that should someone be tested and it says positive that there won’t be discrimination against them, but there are a lot of issues around that. That brings me to family issues, and this is a question we got from Lynn in Topeka Kansas. She says, ‘What planning should family members consider with a juvenile HD child?’ So what should they be doing for themselves and the family and also you talked about schooling but other issues may be about insurance as well?

Dr. Kostyk:
That is an issue because what we’re talking about is something that often even mature adults don’t plan for. You have to with juvenile onset HD eventually you do have to face a very hard decision about end-of-life planning when the disease is progressing and being prepared for progressive disease, which is going to get worse over time. So again it’s very much dependent upon, it’s different if the child is three when symptoms come on or a child who is 17 and almost into adulthood who may have a very different course of the disease. So you have to look at each case, but planning for things like there is cognitive decline and decreased ability to make decisions and impulsive behavior control that you have to worry about with a teenager and being prepared to deal with that versus someone who’s getting to the later stages of life and not able to swallow any further.
Families need to sit down and even for a child make an advance directive. Get together if the child is able to participate or with the other family members so everybody feels comfortable with the decisions. Do we want to do a feeding tube of it comes to that? Do we not want to do a feeding tube? How much support will the child need? Social services; can the family take care of them? For a younger child that may be easier to take care of bathing and dressing. For an older teenager as things go further down you may need more support in terms of visiting nurses or community support. Volunteer agencies have been very helpful with some of our individuals and aid agencies like that. You have to be planning to think about unfortunately the worst-case scenario in addition to hoping for the best in the future.

Andrew Schorr:
Right, now, here’s a question that relates to that because certainly a serious diagnosis like this in a child is a heavy load for the family, so Sydney wrote in from Grand Rapids Michigan, and she says, My granddaughter has HD, and I am not sure how to accept it. How can the entire family get help?’ And then she says, ‘The main problem is she assumes she does not have long to live. How do you explain this to a 14-year-old?’ So, all sorts of family and family communications issues. Can a Center of Excellence help, or how do you get that specialized help for the family?

Specialized Help for Families with HD

Dr. Kostyk:
A center can help or someone who has some sense of the reality of the diagnosis. For instance, if a child has just been diagnosed at age 14, that concept of I don’t have long to live may be unrealistic. It may be that that child has 20 years to live, and one of the disadvantages of getting tested sometimes too early before there are symptoms is that you don’t want people giving up on life when there’s so much life left to live yet. So again it’s a support network. It sounds like if the child is basically, almost it sounds like very commonly after hearing the diagnosis that you have a disease like Huntington’s disease, it’s not uncommon for people to start showing signs of depression, and that itself can be one of the behavioral symptoms of Huntington’s disease. So getting psychiatric support from a child psychiatrist may be the avenue here and starting on there are a number of antidepressants that can be very helpful. There’s even some suggestion that some of the antidepressants that we are using today may have some ability to slow or affect the course of the disease. So I wouldn’t be hesitant to bring in other specialists and work again with a team approach to problems like this because it is a family disease.

Everybody needs to understand it; the other siblings, the parents, the grandmother, and having the diagnosis doesn’t immediately turn you from the granddaughter you were on Tuesday to getting the diagnosis on Thursday. You’re not a different child by the end of the week. It’s a very slowly progressive disease.
Andrew Schorr:
Good point. Thank you for that. I was just going to mention, this applies to every group we talked to. It’s certainly apply to my family with leukemia etc., and that is that the family members in a way, maybe it’s too strong of a term, but in a way you’re patients too, and so sometimes you might need help with counseling and maybe even help for depression. You need people to talk to. Now there are support groups, but it’s not unlikely given that we have millions of people affected by depression for example that this could trigger that for you too, and you deserve help. So you don’t have to stuff it. You have to connect with other people and providers.

That leads us to a question we just got in from Dania from Redondo Beach California. She asks, ‘How often should a juvenile HD patient be monitored by different members of the treatment team?’ And she lists neurologists, psychologists, psychiatrists, occupational therapists, and nutritionists. So she wonders how often should the follow-up be? What’s your guide there at Ohio State?

Dr. Kostyk:
It depends again, and I don’t mean to not be able to give you a definite answer to any question, but we to gear all of our care to each individual, and at certain times I will see an individual every week if we need to. Every three months would not be uncommon for somebody who’s more stable as a child. Someone who’s is doing very well and is established and sort of has not had problems for a period of time, occasionally I’ll see them every six months, but rarely do I see them, especially for a young juvenile form, do I see them less often than not.

Andrew Schorr:
Okay there was another question that Dania had too. She said, ‘Is there any specific medication that has better benefit for juvenile HD symptoms? She’s heard that Haldol is the most effective medication for managing the chorea movements.

Dr. Kostyk:
It’s the one that has the longest track record for the chorea movements that have been used in adults. I don’t think we have any documented evidence that there is one particular medicine that’s better for children with HD versus adults, and the decision of which medicine to use again in that class of medicines; chorea itself does not always need to be treated. It’s not always the problem. If it’s not interfering, sometimes very often actually we don’t treat the chorea. More of the problems that we have are the behavioral problems or the other motor problems; trouble swallowing, trouble walking. We probably do more treatments for balance and gait and swallowing problems in terms of the motor difficulties than we do for chorea. Sometimes the chorea is a problem, and haloperidol is a medicine we may use, but there’s a spectrum of medicines, and I don’t think there is one that’s specifically better for children versus adults.
Andrew Schorr:
So as you look forward, Dr. Kostyk, what would you want to say to this community that have children and they’re very concerned about them, concerned about other children, or even having other children, which were going to discuss in some of our later broadcasts as we look at more about the future of genetic testing and where that fits in as well. But if you want to summarize where we are today and where we hope to be in the near term and the long term, where are we now, and also maybe with a recommendation for people connecting with a Center for Excellence?

Dr. Kostyk:
When you say where we are now, do you mean the kinds of research we’re doing now and where is the disease going now?

Andrew Schorr:
Yes.

Dr. Kostyk:
One of the things about the center is we do in almost all of the Centers for Excellence and in a number of the academic centers there is a group called the Huntington’s Study Group, which is a much larger group of academic centers interested in getting things from the laboratory and translating them into clinical practical use. So we are doing a large number of clinical trials, and as I’ve watched things over the last eight to nine years, the number of opportunities we have for participating in clinical trials and the amount of information we have has been growing exponentially, and we are also participating in trials not only to identify things right now that might be helpful, but we’re doing a number of trials where were trying to learn other things about the disease which may help us identify new avenues of research. So since this is such a rare disease again in terms of making further advances, connecting with centers where people are doing some of the research and looking at changes for the future, I think it’s very important.

Andrew Schorr:
With gun ago unfortunately I’ve got the music playing, so I want to thank you Dr. Sandra Kostyk who is the assistant professor in the Department of Neurology at Ohio State, and she is also the Director of the Huntington’s Center of excellence there. So we’ll have you back, okay?

Dr. Kostyk:
All right, thank you very much.

Andrew Schorr:
Thank you very much for being with us today. I really appreciate it. Remember though folks we’ve got a whole series. So tomorrow we’ve got more as we discuss more about
Huntington’s in adults and more about research the following day on Wednesday. All the replays will be posted on www.patientpower.info. This is what we do. Knowledge can be the best medicine of all. I’m Andrew Schorr. Thank you for joining us, and have a great day.

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