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Speaker 1: Funding for this podcast is made possible through the generous support of listeners like you and sponsorship grants from organizations like Genentech, Teva Pharmaceuticals, Neurocrine Biosciences, Vasonex, and Wave Life Sciences.

Kevin Gregory: Welcome to another episode of the HD Insights Podcast. I'm Kevin Gregory, Director of Education, Communications and Outreach with the Huntington Study Group. In this episode, we have the privilege of chatting with Dr. Martha Nance. Dr. Nance is a long time member of the Huntington Study Group. She's a neurologist and geneticists at the Hennepin County Medical Center and Struthers Parkinson's Center. She also is director of the HDSA Center of Excellence in Minneapolis.
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Kevin Gregory: We thought, based on some feedback questions about family planning, Huntington's disease in youth, we thought it would be a great opportunity to talk with Dr. Nance about her experiences. And while this episode has been our longest podcast of the series so far, I think you're really going to enjoy it. Without any further ado, let's get to that episode now. Here is our interview with Dr. Martha Nance.

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Kevin Gregory: Well, Dr. Nance, thank you for joining us today on the HD Insights podcast.

Martha Nance: Absolutely, my pleasure to chat.

Kevin Gregory: I think ultimately, it's going to be certainly, my pleasure and I think the listening audience's pleasure as well. Let's start out, before we dive into some of the topics that I know are of keen interest to folks. I want to talk with you about your background in Huntington's Disease, and how you got into this. What prompted you to pursue a career in neurology and genetics, and then what ultimately, led you into the HD field?
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Martha Nance: Well, you know there's things that you plan, and things that you don't plan, and things that just have to do with how you were brought up. My father is actually a geneticist. I have, what I refer to, as a warped childhood. The only summer job I ever had, when I was about 14 and beyond, was working in a cytogenetics lab, looking at chromosomes under the microscope. And so, I sort of grew up with an interest in genetics.
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Martha Nance: But then, when I went to college and went to medical school, I found that I was actually also interested in the brain, and neurology, and I was going through training. I finished college in 1980 and medical school in 1984. Believe it or not, this is just about the time that things like CAT Scans and MRI scans started to be done,
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[00:04:00] so the ability to actually look at the brain, which was something we really couldn't do before, in a living person, that was just happening. It was also really a time of an explosion of genetic techniques in the laboratory. I think I must have just known what would really interest me most was the interface between those two things, between neurology and genetics.

Martha Nance: I dutifully did my neurology residency out here in Minnesota, and then I decided to do a fellowship in clinical genetics, so I actually could be a card carrying, fully trained geneticist. Those are all the things that you plan.

[00:04:30] Martha Nance: What I didn't plan and just happened to be here in Minnesota, was there already was a Huntington's disease clinic at Hennepin County Medical Center. It was started in the late 1970s. While I was resident and then as a fellow, I would come to the Huntington's disease clinic and found it fascinating, difficult but really fascinating. When I finished my training, the person who had run that clinic left for a different job, and so there I was a newly minted geneticist neurologist in a Huntington's disease clinic that needed somebody to run it. That was a pretty easy choice, and I've never looked back since.

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[00:05:30] Martha Nance: It continues to be a difficult disease but one that I think is fascinating. I think the scientific progress that we're making has been slow for many years, but I think it's very exciting now to be doing trials of therapies to actually turn off the abnormal gene. Huntington's disease has been good to me, and I think that our clinic has made a huge different in the lives of our patients, even though I have yet to make Huntington's disease go away.

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Kevin Gregory: What were the early days like there? You said that clinic for Huntington's patients was started in the mid to late '70s.

Martha Nance: Mm-hmm (affirmative).

Kevin Gregory: That was well before some of the more recent discoveries in Venezuela and more of the identification of the gene. What was care and treatment like back then, compared to what you've seen it evolve into now?

[00:06:30] Martha Nance: Well one of the things that actually got me hooked, the Huntington's gene was localized in 1983, which basically means if you're looking for the needle in the haystack, that they actually figure out which haystack to look in for the needle. It took another 10 years, until 1993, before the gene was actually identified but one of the things that we did very early on, even before the gene was identified, was to offer predictive genetic testing, using markers that were known to be close to the gene. So we actually were involved in genetic testing of Huntington's disease back in the ... I think it was probably the late '80s, before the gene was actually identified. I feel like I was on the ground floor. It was one of the programs in the

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country that was actually offering this as a clinical service, so that was neat.

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Martha Nance: There really has been an explosion of medications to use to treat symptoms over the last 25 years. One could think of it as just being variation on the theme. I mean, there have been anti-depressants around for a long time, but there are 20 different anti-depressants to choose from. And although there have been medications to treat things like hallucinations, or irritability, or explosive behavior, we have an ever growing number of medications to choose from now, and so you can fine tune much better now, what medication you might use, given what side effect you either want or don't want.

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Martha Nance: For instance, some medications make a person sleepy. That would actually be a good thing, if part of the trouble is that the person isn't able to sleep. Other medications might tend to perk a person up, which might be more useful if somebody is a groggy/sluggish or has a lot of apathy.

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Martha Nance: The treatments, we have more choices. And, obviously, we participated in the research study that Tetrabenazine to the market. My patients were just thrilled to be in that study. And then three years later to be able to say, "The reason this drug is on the market, is because I participated in the study." So, that was fun.

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Martha Nance: We have always had, at my center, a multidisciplinary team. Again, this predated me, but it is a wonderful thing and it makes my care so much easier, when I have a physical therapist, occupational therapist, speech therapist, dietician, psychologist, neuropsychologist, genetic counselor, social worker, all these different people to help look at the different aspects of Huntington's disease. And so we've actually been doing that for decades. It's made my job easier and actually, more interesting.

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Kevin Gregory: That's a good touchpoint there that I wanted to chat with you about in some more detail. I know that multidisciplinary management is certainly one of your passions. But in general, people and even some clinicians maybe, that aren't familiar or experienced with Huntington's disease, may not really recognize the importance of that approach. Can you explain for listeners the value of having a care team across multiple types of discipline that you mentioned, particularly for HD patients?

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Martha Nance: I would love to talk about that. What doctors tend to do is hand out pills. The thing that the physician has power over, that no one else can do, is prescribing medications. And certainly, medications can be helpful for people with Huntington's disease but I always say that sickness or even death in Huntington's disease, is often related to trouble swallowing.

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Martha Nance: The doctor word for trouble swallowing is dysphagia. When you have dysphagia, sometimes the food goes down the wrong tube, and then you get pneumonia, or

[00:11:30] sometimes some people just sort of seem to not really want to eat anymore. I think some people may be actually sort of scared of eating. If you don't eat as much, then you lose weight. And then you could even have an abrupt episode of choking, that could lead to death if you have a piece of saith that gets lodged in the throat or something like that.

Martha Nance: Because morbidity and mortality in Huntington's disease are related to dysphagia, don't you think that maybe you should have a speech pathologist involved, even very early on to evaluate the swallowing, to see if there are troubles, to make sure there aren't any additional unrelated problems? People in their 40s or 50s can also have throat problems, a stricture in the throat or a little pouch off the throat, so make sure you're optimizing everything about the person's current ability to swallow.

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Martha Nance: Do a little teaching early on about the kinds of changes that occur in Huntington's disease. And then, just work with the patient intermittently throughout the course of the disease as things change, to help the person maybe change the textures of the foods that they eat or maybe change the approach to eating.

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Martha Nance: For instance, we're all used to thinking of eating as being a very social activity but for some patients, it may be a bad idea to try and eat and talk at the same time. Some of our patients are actually better off eating in a very quiet situation and if you're gong to do your talking, do that before or after dinner but not in the middle of dinner, because you're more likely to inhale and swallow at the same time, which could be bad.

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Martha Nance: And then we tag team at our center. I've always been very proud of the fact that we have a dietician also on our team as a really an integral part. The dietician and the speech pathologist often go in together to see the patient. The speech pathologist does what they call a bedside swallow or a clinical swallow evaluation, just in the exam room, and talks about textures of foods and so on that might be better or worse for a person. The dietician can then translate that into what specific food should you not be eating, or should you try to avoid, or what specific kinds of foods would be safer?

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Martha Nance: And so, they really work very closely together, to really educate people early on about the swallow changes that can occur. Many of our patients also lose weight and whether it's due to having too much chorea, or whether it's due to financial problems that they can't buy food, or whether they need somebody to prepare food for them that isn't there. The dietician will also identify, if somebody is losing weight, what's really going on, "How much are you eating? How many calories do you need?" And can recommend resources or strategies to improve the calorie intake.

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Martha Nance: We get those folks involved very early with essentially, all of our patients. The social worker, obviously, is a critical person, as things change and care needs increase. One of the challenges with Huntington's because of the young onset age,

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Martha Nance: And oh boy, aren't there some strategies or resources in the community to brighten up the life or make things easier for the family? Or if people need to be placed outside the home at some point, it just isn't working at home, how do you organize that? The social worker is obviously, an integral part of the team from beginning to end.

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Martha Nance: And so on, the physical therapist, occupational therapist. OT talks about safety in the home and how you get activities of daily living accomplished.

Martha Nance: Physical therapy typically talks about the mechanics of walking, and balance, and falling, and exercises that can help to maintain function as long as possible.

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Martha Nance: Psychologists can be useful, not only for the patient but for the other people in the family. How do you adapt to having this progressive neurodegenerative disease at your age? How about your marital relationship? How about relationships in the family? The psychologist can be very important.

Martha Nance: The neuropsychologist helps us with assessing cognition, both in terms of helping the patient and family to understand if there are problems that maybe one or another doesn't realize or doesn't acknowledge. Formal cognitive testing can also be very helpful in supporting a disability claim.

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Martha Nance: And so on, I can go on for hours about the roles of each of these people. It makes my life much easier to have all these people be part of my team. I think it's better for the patient.

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Martha Nance: I had a slide I made once, of the HD molecule, which had the patient in the middle, surrounded by a shell of their immediate family. That was also surrounded by a slightly more distant shell of the more distant family members, aunts, cousins, or people that might help out. And then the medical care team is another shell that's there to help support the patient. And then finally, there's training of people in the community, so that they understand a little bit more about Huntington's disease, so you don't feel quite so lost or alone when you go to the grocery store or go a movie when you have Huntington's disease.

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Martha Nance: We really try to build this strong base of support, that really surrounds the person with Huntington's disease.

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Kevin Gregory: As I understand it, from things I've learned over time here or heard about, your

approach to patient care also gets you out and about in the community a lot, particularly in terms of not just newer patients or earlier stages of diagnosis, but people that are in long-term care facilities or hospice.

Martha Nance: Yeah.

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Kevin Gregory: Can you talk a little bit about ... Give us a day or a week in the life for Dr. Nance, in terms of getting out of the clinic into the community with patients?

Martha Nance: Yeah. Well, so one of my mantras has always been ... Well, there's several of them but there's never nothing you can do for somebody with Huntington's disease. You can flip that around and say, there's always something you can do for somebody with Huntington's disease, up to and including on the day they die.

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Martha Nance: I think doctors sometimes take a narrow view of what doctors do. We have dominion over the prescription pad, and so we see patients, and we refill their medication, and we're very sorry to hear that things are worse, but here's your refill and we'll see you next year. I think quite often, the patient and the neurologist lose touch with each other, quite often in the late stages of the disease. Why is that? Well, the neurologist will say, "Well, the patient moves to the nursing home, and becomes immobile, and then they quit coming back to clinic."

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Martha Nance: I think that's probably true, so what you might have to do then instead, is go to where the patient is. I think, unfortunately for the doctor, it's not very efficient if you have 20 patients, and they're in 20 different assisted living facilities or long term care facilities. It's very inefficient for the doctor to drive to 20 different places.

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Martha Nance: But I have, again, this marvelous resource here in Minneapolis of a long term care facility, that has a 32 bed unit for people with Huntington's disease. It's actually much more efficient to go to the nursing home and see 10 people in an afternoon, than for those 10 people to get loaded up into vans, and come to see me in the clinic.

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Martha Nance: I do physically go to the nursing home and see patients. And then I can also support the staff at the nursing home. If you think about the work that direct care staff do for people living in nursing homes, it's a lot of work. I think maybe we don't give enough thanks to the people that do that work. For my folks on the HD unit to understand what a unique service they're providing, that there aren't that many nursing homes for people with Huntington's disease.

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Martha Nance: A few years ago there was an aide that worked at the nursing home, whose side gig was she liked to do nails. And she, one day, decided to bring her nail polishing kit into the nursing home with her and she did everybody's nails. That's been a thing ever since then, that my patients get their nails done. It really is a remarkable thing. I can tell you that ladies in the late stage of Huntington's disease are happier when

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they have their nails done, than if they don't.

Martha Nance: It that kind of little thing. I don't do these things myself, but I think that we've created an environment where people are able to go that one step further. We're not just doing custodial care of some guy with late stage Huntington's disease, we're going to make sure that person actually looks good and feels good. That's one of the things I do, is go out to the long term care facility, and actually see the patients there, and I think support the staff who are providing the care.

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Martha Nance: We also have some amazing families in Minnesota, at least three different families, who have ... Actually, there's four or five now, that we've groomed over the years, of group homes, that provide care for people with Huntington's disease in a home environment. You have a house that has four or five bedrooms, and you have four or five people with Huntington's disease living in that, truly a home-like environment.

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Martha Nance: Again, providing support for the folks who created these group homes, and empowering them, helping them to understand that they are also doing something unique, and special, and that you that for 10 years, and they too are experts.

Martha Nance: Those are some of the ... And along the way, we try not to abandoned our patients in the late stage of their disease.

Kevin Gregory: Right.

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Martha Nance: We try to maintain a connection up to and through including the day they die. You're better off dying with somebody holding your hand, than you would be dying alone.

Kevin Gregory: Absolutely.

Martha Nance: You know? There may not be a pill to give you at that point, but how about a hug?

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Kevin Gregory: Absolutely. You mentioned the facilities and the families that have put together locations and places for Huntington's patients, a long term care facility that focuses in on that, and these group homes. That's been one of the challenges in this field, is finding that type of setting in states across the country, that are willing to take in HD patients, just because of the challenges.

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Kevin Gregory: Just from your experience in interacting with these groups and these organizations, what are the types of things you would recommend to people in other areas of the country looking to start up these types of programs? What were the challenges or what are the recommendations you would suggest to try and make these things a

success and a part of the community for future care?

Martha Nance: I would deflect that to the people at the facilities who are the real experts. I mean, these things came to be, quite honestly, without me doing anything. Now one can argue, would there be an HD nursing home if there wasn't an HD clinic? Probably not. Would people have developed HD? Would a family have had the idea or the courage to develop an HD group home if their own loved one with Huntington's disease wasn't receiving adequate care? Probably not.

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Martha Nance: As to the details of getting through the legal stuff, and how do you make it work financially, and how do you keep it going on an ongoing basis, I just think there should be ... I mean, you could probably create a task force, or working group, or whatever you want to call it about late stage care and it shouldn't be me. It should be these other folks that run these programs that are in that group.

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Martha Nance: There are going to be different challenges state by state, both in terms of regulations and financial support available for late stage care. But there's also this, I think great misunderstanding, that I think it probably should be me or us, the researchers, that should try to fix. I think there are many care facilities that are sort of allergic to the word Huntington's disease. You just say, "I have a patient with Huntington's disease," and they hang the phone up. That I think is based on a mid-20th century misunderstanding of what Huntington's disease is or requires, in terms of care. Not everybody with Huntington's disease ...

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Martha Nance: I think what they're worried about at care facilities, is that the patients are going to have behavior problems. And yes, there are occasional patients with Huntington's disease, for whom behavioral management in the late stages is challenging, but there's an awful lot of people with Huntington's disease in the mid to late stages of disease who don't have challenging behavior to manage. And by the way, with the explosion of people with Alzheimer's disease, people with Alzheimer's disease have challenging behaviors to manage too.

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Martha Nance: First of all, there should be more knowledge, just in general, about how to manage patients with dementia, perhaps than there used to be. And number two, not everybody with Huntington's disease has challenging behaviors. I think we, as a HD research community or a professional community, could probably do more to try to fix these misunderstandings in the long term care community.

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Speaker 1: We'll return to the interview on the HD Insights podcast in a moment. We hope that you're enjoying this episode. As a nonprofit organization, the Huntington Study Group relies on the generous support from the community and listeners like you, to continue bringing you in depth content on HD, like this podcast series. If you like what you're hearing and are interested in supporting HD Insights through a grant or

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donation, please contact us through our email address info@hsglimited.org or by calling toll free at 1-800-487-7671. We greatly appreciate your support. And now, back to our episode.

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Kevin Gregory: I'd like to talk with you Dr. Nance about the other end of the spectrum now, and that's youth and the impact of HD on youth. Starting with juvenile onset HD, some people might just naturally assume that the juvenile onset version is the same as adult onset, it's just that it happens sooner in kids. That's not entirely accurate.

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Kevin Gregory: What are the main differences really between the juvenile onset symptoms and those you would find in an adult?

Martha Nance: Yeah, so the symptoms, there is some overlap but they certainly can be different and sometimes quite different. I think the closer to age 21 a child is when they

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Martha Nance: The ones where it really looks quite different from adult HD, are the younger children. In a younger child, there's quite likely to be little or no chorea, none of those wiggly extra movements. There's actually much more likely to be rigidity, stiffness, truncal stiffness, which if you have stiffness as a mid-line, the trunk, then these are often kids who are toe walkers. They're up on their toes because their legs and trunk are stiff. They may have ... Again, another mid-line structure is your throat, so children may have stiffness in a sense of the throat. They'll have drooling or trouble with articulation.

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Martha Nance: The motor symptoms look very different than the motor symptoms in an adult. The other thing is how you become aware of it is a little bit different. In an adult, adults already know how to ride a bike and drive a car, but they lose those skills. What you might see in a very young child, a five-year-old, or a two-year-old, or an eight-year-old is that they can't quite ever learn how to ride a bike, or hit a baseball. They may, at some point, fail to acquire skills or never really master certain skills that

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[00:32:00] you should master by a certain age. There may be a slowness or you hit a plateau and you've failed to acquire for their motor skills.

Martha Nance: Second thing is although you and I are probably losing brain cells, a few a day, me more quickly than you because I'm older, you don't really notice it. You and I aren't really having a quiz every week but children are when they're in school. They have tests every week and they have to learn a new subject every month or every semester. And so, it's likely to be much more obvious, more quickly, if a child is failing to gain intellectual skills. Or if a kid used to be getting B's in school and now they're just not performing very well, they're not learning very well, that sort of becomes obvious because they're tested all the time. Cognitive changes certainly can occur, but it's going to present as poor performance in school or a declining performance in school, compared to however it was they used to do.

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Martha Nance: And then the third thing is for some children, and this is where doctors and parents struggle with each other, for some children the most prominent early feature is behavioral. In my experience, when HD presents in a child with bad behavior, it is not subtle bad behavior. It's really bad, bad behavior. I mean, it's an eight-year-old strangling the two-year-old with a light cord. Or it's major drug abuse at age 12, or it's burning down the school building. It's not being sassy to your mother when you're a teenager.

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Martha Nance: But of course, every parent of a pre-teen or teenager whose spouse has Huntington's disease, and the kid gets a lousy grade on a test, or mouths off to mother, or won't do what they tell them to do, or heaven forbid has a tantrum and throws something, or is smoking behind the school building, every mother is going to be in a panic that, that child is starting to develop the bad behavior that might be a symptom of HD.

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Martha Nance: And that's where it's a challenge. I think doctors, neurologists, pediatricians are really hesitant to leap to a diagnosis of Huntington's disease, just based on mild disruptive behavior. Anybody who has ever had a teenager has experienced disruptive behavior.

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Kevin Gregory: Oh, yeah.

Martha Nance: We struggle with that and the parents know darn well that this isn't normal or this isn't the way the kid used to be, and the doctors are slow to get there. We look for something we can sink our teeth into a little bit more firmly, like a change in academic performance at school, or a change in motor performance, or some kind of finding on neurologic exam.

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Martha Nance: And then the last thing that's much more common in kids than adults would be seizures. Epileptic seizures are not present in all children with HD, but they're not unexpected. They really range from being a minimal, it's not a big deal sort of thing, to being a pretty dramatic problem that's difficult to manage. The seizures are different.

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Martha Nance: People argue, does juvenile HD progress more quickly than adult onset HD? Maybe. If you get symptoms when ... My little guy who had symptoms start when he was two, lived to be 18, so that's actually a 16 year course. It seemed like it went more quickly. But actually, if you added up the total number of years, it was 16, which isn't that unusual for a total course of HD.

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Martha Nance: I'm not sure that it really progresses more quickly. There are some papers saying that it does. But I think the difference between a normal child who is acquiring skills, and gaining strength, and energy, and knowledge, and beauty, and a kid with Huntington's who is declining, I think you see a bigger more rapid disparity between the normal kid and the person with Huntington's than maybe an adult.

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[00:37:30] Most adults are static in their performance during your middle aged years, and you compare that to somebody who has got a decline in HD. So maybe the differences between normal kids and kids with HD are more prominent, but I'm not sure that the course is really that much quicker.

Kevin Gregory: The other area of youth and impact with Huntington's disease, and I think this ties into what you spoke about earlier, having this larger care team to help, is the impact on an unaffected sibling of someone with juvenile onset, or the child of a parent with HD. What types of things have you seen or are you on the lookout for, in terms of, those younger family members that are part of an HD family, the risks or the things that you're looking to make sure that they're doing all right as well?

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Martha Nance: Yeah. Huntington's is a family disease, in every meaning of the word. It's a dominantly inherited genetic disease. If you've got it, then your children have a 50/50 chance, each child, of getting it. But it's also a family disease, you can not go through a whole course with Huntington's disease by yourself. You just can not go through this disease, from beginning to end, without somebody else being involved in your support. When the going gets tough, who we all lean on in the end is our families. It really becomes a family disease, in that way, as well.

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Martha Nance: It absolutely impacts ... A parent with Huntington's disease who has children at home are absolutely impacted by the disease. They can't not be. The first challenge for me, is to try to actually meet those kids. Did you ever go with your father to his doctor's appointment? No. Why would you?

Kevin Gregory: Right.

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Martha Nance: At what age and how do you get the parents to bring the kids with, so that I can meet them? One of the things I often tell parents is, one way that kids sometimes deal with Huntington's, is by writing a paper on Huntington's for some class at school or doing some kind of media project in school about Huntington's. I love to work with kids on those projects. And you know what? They always get an A. Why? Because they know more about the topic than their teacher does, and they speak from the heart.

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Martha Nance: And so, one way sometimes to get a kid, a middle school or high school aged kid, understanding a little bit more about HD, in a way that may not be quite so threatening, is to work on a paper for school about it. I always tell parents, "When your kid gets to be ninth grade or has a health class thing he has to do, bring him by. I'd love to talk to him." And then the problem you have is if you never meet the kids before they turn 18, then you never meet them because then they fly away. But you also try to listen for, "How are things going at home?" If it's a parent with Huntington's, how are things going with the affected person with Huntington's? But how's it going for the kids too?

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Martha Nance: There are resources available, both through the Huntington's Disease Society of America has its youth alliance and the HD Youth Organization, which is particularly near and dear to my heart because it was co-founded by the son of one of my patients. There are resources for kids. There are places for them to get accurate information and to find other people who have a shared experience. They shouldn't have to be alone. I try to direct parents or kids.

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Martha Nance: I think if you're a sibling of somebody with juvenile onset HD, I think that's hard. In some ways, it may be almost easier than having a parent with HD. One of the things we underestimate, kids are very resilient. Depending on the behavior issues, or the financial issues, or the family structure, or whatever, some living situations, if you have no money, and you're living with an affected parent, and the unaffected parent left. I mean, there's some very complicated and sad situations that people can get into. But there are also some very beautiful situations that kids can get into, and they are resilient, they learn things, and they sometimes become better people or different people because of their experience with a sibling or a parent who had a medical illness. It makes you a different kind of person when you grew up with sadness but you can actually grow from it, and become a marvelous person as a result.

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Kevin Gregory: Absolutely. Dr. Nance, I want to turn over to the topic of genetic testing. It's certainly an important issue to a lot of people. The question of whether to test or not to test for HD would seem to be the topic of greatest interest to those that are potentially at risk for Huntington's disease. Can you tell us why it's not really a question of yes or no to test, but really something that you should start with discussing and engaging a genetic counselor about first?

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Martha Nance: Mm-hmm (affirmative). Yeah. I've been involved with predictive testing for Huntington's disease since 1988 or something like that, so I've seen now ... Actually, we're coming on the second or third generation of people having predictive testing.

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Martha Nance: Number one, misunderstandings about genetics are still rampant among patients, families, and I'm sorry, doctors. People will sometimes come in with a misunderstanding or misinformation about what their risk is, or whether they're at risk, or why they're at risk, or even whether the family disease is Huntington's disease or not. It may be that somebody said it was, but did anybody ever actually get a gene test to prove that it really was?

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Martha Nance: That's why, to me, anybody who is thinking about having a predictive gene test, really does need to work with a genetic counselor. This is what genetic counselors do for a living, is they explain genetic concepts, they run through the family history, they'll look through or have you access the medical records, so that we know we're

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even testing for the correct disease.

Martha Nance: I can tell you, over 30 years, I've seen every variation of, "Whoops. This is weird. This isn't what we thought." I've seen every variation of that, that you can imagine. We really want to make sure that we're even talking about the right disease. You need to know what the possible outcomes of the test are. It's more or less black and white, but there's a little bit of shades of gray. You need to understand that before you get the gene test. You need to understand what the test tells you and what it doesn't tell. Okay, you've got the gene, "So, when am I going to get it?" Well, I don't know. It will tell you that you have the gene, but it's not going to tell you when the symptoms are going to begin. In some sense, it replaces one unknown with another.

Martha Nance: And then, why are you getting the test? The question I always patients is, "Why are you coming in now?" The test has been available since 1993, and if you're anything over 18-years-old, why didn't you come in last year? Why not next year? What's the rush? Why are you here today?

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Martha Nance: Sometimes people are moved to get a predictive test because they think they're having symptoms or some kind of symptom, and then they're nervous that it could be a symptom of HD, and they ask for a gene test but what they really need is a neurologic exam. If you're having headaches, and blurry vision, and you're at risk for Huntington's disease, you don't need a gene test. You need a neuro exam and an MRI scan.

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Martha Nance: We often have people meet with the genetic counselor. Sometimes they meet on the phone for an hour before they even come into the clinic because there are also insurance issues, employability issues. There are laws, The Genetic Information Nondiscrimination Act, or GINA, that is supposed to protect a person against genetic discrimination but the laws only go so far. To sort through, how much does the test cost, are you going to pay for it, are you going to submit it to your insurance, what are the pros and cons of either of those possibilities?

Martha Nance: And then thinking through, what will you really do with the results? There's what you think you might do, and then there's unpredictable or uncontrollable things. You can't control other people's reactions to your results. You may think that your spouse is enthusiastic about you getting a gene test or that your mother will be thrilled if you test negative, but what if they don't respond the way you think they should? You need to be prepared for that.

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Martha Nance: So, a lot of thinking through. We actually spend more of our time talking about the negatives. Most people want predictive testing, barrel on in, and they've thought about it, and they've lived with this disease, and they know they want the testing. We spend a little more time exploring what are the potential downsides of being tested?

Martha Nance: I always liken the gene test more to a surgical biopsy than a blood test. We're taking a little snip of you and revealing it in a way that we can't undo, anymore than we can put your appendix back after we take it out. Once you have the gene test, you can't un-have it, so let's make sure that this is really going to be a useful piece of information.

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Martha Nance: Now, all of that may change. Obviously, if there's a treatment that changes the nerve cell degeneration. We're in this weird time right now, where there's starting to be research to develop treatments that we think may turn off the abnormal gene. But number one, we don't know if those treatments work.

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Martha Nance: Number two, you can't get into the research study just by having a gene test. For all the current trials that are going on, looking at gene-based therapies, you have to actually be symptomatic, diagnosed with Huntington's disease. A 20-year-old who has no symptoms of anything, who just got a gene test result and knows they have the gene, that alone doesn't qualify them to be in one of these research studies.

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Martha Nance: In this sort of weird time, where again, there's some misunderstanding that people want to get the gene test so they can get in the research study. Yeah, you can't get into a clinical trial of a gene silencing treatment just because you're gene positive. You actually have to have symptoms, at this point.

Kevin Gregory: Can you tell us about some of the reactions from people that have gone through the genetic testing process, that really surprised you, that you weren't anticipating, or that moved you in a way that you didn't think you would have that kind of reaction?

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Martha Nance: There are lots. They're very personal, and unique, and hard to describe quickly, and also hard to describe without giving out too many personal details. But I would just say, as I said, I've had ever variation of strange testing situations, so people who want to come in with their brother and sister, and all get tested together. Which we think is a bad idea because your brother gets a normal result, and you get an abnormal result, and you both thought it was going to go the other way around, and all of a sudden it's very awkward in the room together. We discourage that.

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Martha Nance: I've had ... Again, people with Huntington's are quite resilient. We tend to encourage them to bring a companion with them. We don't require it, but we certainly encourage people to bring somebody with, so you've got somebody else in the world that knows what news you got today.

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Martha Nance: I had one patient once, where the patient halfway expected to get the result they got. The companion, who was just one of their buddies, broke down and was balling. My poor patient was trying to support the support person, who was just

devastated by this result.

Martha Nance: We've had people get ... I actually recently had somebody who delayed coming in for a long time. They had a sibling who had been tested 20 years ago, and the person finally was brave enough to come in and get the gene test. I had taken care of the person's mother, who had HD, and it was really fun to give good news. The person just about fell off their chair because I think they really had lived a long time just thinking they were going to get it.
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Kevin Gregory: Oh.

Martha Nance: It's really fun when you can give happy news.

Kevin Gregory: Yeah. Well, there's definitely a lot to consider, for anyone that wants to go down this path or has been thinking about getting tested for a while. It's definitely worth the longer discussion.

Kevin Gregory: Dr. Nance, I just want to wrap it up with a couple more questions for you, just for you personally. I'm really interested, you've had just an amazing wealth of experience and work in the field, but is there one thing in your life that you would consider or classify as your proudest accomplishment?
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Martha Nance: Yeah. Well, we talked about Huntington's disease being a family thing. I mean, without a doubt the most important thing I did was raise my two boys to be the marvelous people that I think they are. That's actually the most important thing I've done.
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Kevin Gregory: Awesome. And then, I think I may know how you will answer this but I'm going to ask you anyways. Who do you most consider your mentor?

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Martha Nance: Oh, geeze. You probably don't know the answer to that.

Kevin Gregory: Oh, okay.

Martha Nance: There's not going to be a single person but sort of a number of people. I think we all ... The field of Huntington's disease would not be anywhere near where it is without Nancy Wexler, who was a role model for two reason. One, obviously, a leader in the field of Huntington's disease but also, a role model I think for a woman going into medicine and research at that time, back in the '70s, '80s. That would be one person.
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Martha Nance: Actually, a role model, in terms of neurogenetics is Tom Bird, out in Seattle, who we don't talk that often but I think he knows that he holds a special place in my heart and mind for ... He's also just the kind of person you would want to be as a physician. He's soft spoken, and listens, and thinks, and is articulate, and actually,

[00:56:30] he just wrote a book that came out last year, which everybody should read, about his years of experience with some of the stories that he has to tell about patients with Huntington's disease.

Martha Nance: I don't know. If I start naming more people, I forget somebody but I think those two people probably stand out as unique individuals who I became aware of at a formative time in my life.
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Kevin Gregory: I appreciate it. I'm going to claim partial credit because I was thinking Dr. Wexler, just based on having met her at the HSG annual meeting in Houston, when she was there receiving the lifetime achievement award. Partial credit.

Martha Nance: Yeah, okay. All right.

Kevin Gregory: Well, Dr. Nance, thank you so much for taking time out of your schedule. I've just had an absolute pleasure to speak with you on all these topics today, and just really appreciate your time with our audience.
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Martha Nance: It was my pleasure too. We should all move forward, probably with our heads up. Huntington's is a bad disease but the more we talk about it and the more we don't hide or heads in the sand or feel like there's a stigma, let's just move forward and get better solutions.

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Kevin Gregory: Absolutely, great advice and words to live by. All right, Dr. Nance, thank you very much.

Martha Nance: Great. Thanks, Kevin.

Kevin Gregory: Well, I absolutely enjoyed that conversation with Dr. Nance. We chatted for almost an hour. I felt like we could have easily gone on to a second hour, and I promise we'll have her on again to pick up the conversation, hear about more stories and experiences that she's had with patients and her clinic. Just a delightful person to talk to.
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Kevin Gregory: We hope you enjoyed this episode. Stay tuned and look for another episode of the HD Insights podcast coming in a few weeks.

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