107| Neuropsych Bite: Clinical Case 14 – With Dr. Wendy Kelso

November 15, 2022

NAVIGATING NEUROPSYCHOLOGY

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Speakers: Wendy Kelso, Ryan Van Patten, John Bellone

Intro Music 00:00

Ryan Van Patten 00:17
Welcome, everyone, to Navigating Neuropsychology: A voyage into the depths of the brain and behavior, brought to you by INS. I'm Ryan Van Patten...

John Bellone 00:26
…and I'm John Bellone. Today we speak with Dr. Wendy Kelso about a clinical case, a man with Huntington's disease, or HD. Wendy is a senior clinical neuropsychologist at the Royal Melbourne Hospital in Victoria, Australia. She's also

coordinator of neuropsychology services at the hospital. The first 15 minutes or so of this episode are a review of the case and then we step back and talk about Huntington's more broadly referencing Wendy's case along the way. We haven't covered HD yet on NavNeuro and this episode includes important clinical information about the condition. So, with that, we give you our discussion with Dr. Wendy Kelso.

Transition Music 01:06

Ryan Van Patten 01:15
Okay, we're here with Dr. Wendy Kelso. Wendy, thanks so much for making the time.

Wendy Kelso 01:20
Thanks so much for having me, Ryan. It's a great privilege and I really appreciate the opportunity to talk about some cases today.

Ryan Van Patten 01:28
Yeah, so tell us about your first case, the Huntington's disease patient.

Wendy Kelso 01:34
I thought I'd talk about the first person who was a gentleman in his early 40s that was gene positive for Huntington's disease and he had 46 CAG repeats. He found out that he was positive for Huntington's disease in 2007 and then in 2008 he came for an initial baseline assessment because he was having some difficulties at work and I think he was a bit concerned about if some of these were early symptoms of Huntington's disease. Just to briefly summarize, at that point, he was working as a police officer. He thought that he might not be making sensible decisions in his work and it was taking him longer to make high level decisions, and so he came for baseline assessment at that time. Essentially, when we saw him for assessment back then, he performed pretty normally on cognitive testing and he was very much probably premorbidly in the high average range. He had some difficulties on fine motor speed tasks and on logical memory. He had some difficulties learning stories and remembering over time, still in pretty much the average range, but that was thought to possibly reflect decline. At that stage, he had no motor symptoms of Huntington's disease and was still considered asymptomatic.

He was referred back about five years after that time for further assessment because he noticed increasing difficulties at work. At that time, he was still working

as a police officer and he was having more difficulties on the job with remembering the details of what he was meant to do each day. He was having difficulties remembering phone calls and conversations with his colleagues and with people that he was investigating. He was questioning his ability to make reasoned decisions and good judgments when interacting with the public. He was concerned that he was putting himself at risk and potentially his colleagues when he was attending different scenes. He wasn't always surveying the areas for the risks that were currently involved and he was concerned that this was a change.

With regards to the medical history, there was a maternal family history of Huntington's disease and his mother had become symptomatic in her early 50s and passed away in her late 60s. He also had a maternal uncle and cousin with the disease. The last years of his mother's life was spent in a psychiatric institution where she actually denied that she had Huntington's disease. He had a number of siblings, one was gene negative and two of his brothers had not been tested for the disease. He was married with three children, and two of his children were aware of the diagnosis. One of his sons had had genetic testing and recently found out that he was positive for the gene and the other son had not. One of his sons also had children of his own, which was of great concern to the patient because he realized that the condition would be passed down to the next generations as well.

In terms of the main cognitive complaints, the patient reported changes in how quickly he was taking information and explaining that it was taking a while for the gears to get going when he was on the job. He was having difficulties writing statements and reports and preferred complete silence so he could concentrate on the task at work. He hadn't noticed any changes to his language function, but he did know that it was taking him more time to prepare a response and to articulate what he was going to say if he had to give evidence. He had also noticed increased clumsiness and poor balance but he denied any change in motor speed or in handwriting at that time. As I spoke about before, there were a few incidences where he thought that he might have been putting himself at risk or the public, and he wanted to get assessed because he was concerned for the safety of other people in the community that he was working with.

In terms of his mood, he described becoming more upset more easily and fluctuations in days where he found that sometimes he just couldn't be bothered engaging in conversation with others. This was a significant change as he previously described himself as an extremely happy and joyful person. Over the past 12 months, he'd commented that he'd been more sad, particularly during the winter months, and his mood was about a 4 to 5 out of 10. He noticed that he was sleeping less and having difficulty waking up in the night with some motor

restlessness. He was now avoiding eating nuts because he was finding it difficult to swallow them.

When we interviewed his wife, she had not noticed any significant deterioration in functioning, although had noticed slight changes in speech with the patient becoming slightly more clumsy. There was now more saliva present when he was talking. But apart from that she hadn't noticed any significant difficulties and, importantly, she thought that his memory was actually intact. I think it's important to note at this time, I thought that the wife might be underreporting the cognitive changes in the patient because she was wanting the patient to be able to remain employed for financial reasons. I thought it was possible, to protect him, that she was wanting to make him come across in a way that showed that he didn't have many cognitive difficulties.

John Bellone 07:05
Wendy, to clarify, this was prior to the second evaluation? These symptoms that you're...?

Wendy Kelso 07:10
This is prior to the second evaluation.

John Bellone 07:12
Okay.

Wendy Kelso 07:12
Yeah. So, on assessment, he was a casually dressed man, friendly in demeanor, and easily engaged in conversation. His affect was appropriately reactive and mood was euthymic. His speech was of reduced tone but normal rate and there was occasional mispronunciation of words with minor slurring evident. Expressive and receptive language were considered grossly intact on a conversational level. There are involuntary movements noted in the neck and hands. He is able to provide a coherent history and sequence of events. There was no formal thought disorder or visual or auditory hallucinations. He did fatigue throughout the assessment, and hence we had a break in between.

In terms of the core findings of the assessment, he was at least of the high average range premorbidly and he was fully orientated. In terms of processing speed, there was mild to moderate reductions compared to the previous assessment five years previously, and basic motor speed was now in the average range. Performance on more complex psychomotor tasks was in the borderline impaired range. Language

processing skills were sound, and confrontation naming was preserved. There was no evidence of word finding difficulties or paraphasic errors or agrammatic speech, but there was evidence of subtle motor changes impacting speech production including mild dysarthria. With regards to visual spatial functioning, there was no gross visual spatial disturbance; however, there was an inattention to detail. There'd been a significant reduction on Block Design where they'd been from the superior range to the average range - so from scaled scores of 17 right through down to 10 - and this was really due to difficulties with planning and problem solving rather than difficulty with visual construction, specifically. Fine motor coordination decreased. In terms of memory performance, really his memory had declined a little bit but he wasn't amnestic. So he's slower to learn new information than he was five years previously, but he was still able to retain the information over a delay. It was much more a profile of difficulties with retrieval rather than rapid forgetting. In terms of executive functioning, there'd been a decrease over time where there'd been declines in verbal abstract reasoning skills, decline in both letter fluency and category fluency, and decline in problem solving and planning over time. At a behavioral level, he was not disinhibited or impulsive at the time, but he was possibly slightly overfamiliar during the interview. In terms of self-report measures of depression, anxiety, and stress, normal levels of depression and anxiety and mild to moderate degrees of stress.

The patient was seen again recently for review evaluation. At this stage he was in his early 50s and the assessment was to identify strengths and weaknesses to assist with planning for the National Disability Insurance Scheme, which is a scheme in Australia to support people that have disabilities. By this stage, he retired from work as a policeman and he'd been medically retired at the request of the police force. There'd been a gradual deterioration in skills in this role where he'd been able to change roles over time to accommodate his changes in cognitive functioning, but was to the point now that he was having such considerable difficulty that he was fully medically retired and was not working at the time of this evaluation. He was living in a rural community, and he was quite isolated. He was still driving a car and this was a concern to his wife and also to the medical professionals that were looking after him at this stage.

When he was seen in the final assessment, there were very significant changes in cognition and now it was much more global in terms of the domains affected, and that included changes in immediate attention, psychomotor speed, and executive function with significant declines over time. At this presentation, insight was considerably reduced and the patient, while quite depressed, was less concerned about the cognitive changes that he had been previously. At this stage, he was definitely slower to learn new information and was having considerable memory difficulties in terms of his functional daily living; however, again, he was not

amnestic and the pattern of memory was, again, a retrieval rather than rapid forgetting pattern. He described having a slightly shorter fuse where he was more irritable and easy to anger, and that he was making errors when he was trying to fix things around the house. He gave me an example of having great difficulty lighting fires when he was to provide heat where he was living at the time, having more difficulty fixing things around his property, having more difficulty actually operating more complicated appliances, and also having more difficulty driving where he had to turn off all distractions otherwise he was concerned about potentially having a crash or running into someone else. Again, at this stage, his memory was a relative strength and his language and spatial skills were still relatively preserved; however, there were significant changes this time in speech articulation and volume. The patient had more slurring when he was talking and also his voice was quite considerably softer than it was in the initial assessments.

Ryan Van Patten 13:17

All right. Thank you, Wendy, for that summary. I will really briefly review who we have here. We have a male patient in his early 40s, who was initially a police officer, who was found to have 46 CAG repeats, which we'll get into in a few minutes what that means. He has Huntington's disease. One of the initial symptoms that he reported was that it was taking him longer to make high level decisions at work and that change, as far as I heard, came even before any motor changes for him. A few other aspects about him: His mother had Huntington's disease as well as maternal uncle and cousin. He's married with a few children and at least one grandchild, and we can talk about implications there and concerns. He reported some changes in processing speed, apathy, and depression, which are definitely changes for him from the past because he was previously a happy and joyful guy. His wife initially minimized or denied some changes in him, but this may have been due to other incentives around her wanting him to continue working.

You saw him three times, as far as I heard. The first time for the neuropsych eval, most test results were normal. Some maybe subtle difficulties on fine motor speed and logical memory. Five years later, there were some reductions in processing speed. This is a second neuropsych eval that you did with him. Lower psychomotor scores, but other skills were preserved like confrontation naming, visual spatial skills. Some mild declines in memory, but not not amnestic. And then the third time, which was a few years after the second time, the third neuropsych eval, he had medically retired from his work as a police officer, continued deterioration in cognitive functioning, starting to look more like global decline. We can keep in mind Huntington's is a degenerative process that's going on for him under the surface here. So some variability in his cognitive profile, but it sounded much more concerning. Maybe visual spatial skills were somewhat preserved, whereas

attention, speed, executive functions, and the subcortical memory skills were quite low.

So that's my off the cuff really, really brief summary. Let me know if I got anything wrong there. [laughs] I'd love to hear about any neuroimaging he had.

Wendy Kelso 15:51
Sure, Ryan. This gentleman had baseline MRI imaging and then was seen subsequently at each evaluation and had an MRI taken at that stage as well. So, often in Huntington's disease, in the early stages, the MRI imaging is actually normal. You may see changes, for example, particularly in apathy and depression, and some cognitive changes in fine motor control and processing speed even before the changes on MRI. For this particular gentleman, when he was first seen, his MRI imaging was considered normal and he was considered asymptomatic for Huntington's disease. When he was seen five years ago, at that stage, he had very subtle bilateral caudate atrophy, but otherwise his MRI was considered normal. And when he was seen in the last examination, he had more significant caudate atrophy on MRI.

John Bellone 16:52
That atrophy sometimes is referred to as boxcar ventricles, because once there's significant neurodegeneration it makes the ventricles look sort of box-like.

Ryan Van Patten 17:01
They lose their contour.

John Bellone 17:03
Yeah, the contour of the part of the basal ganglia. Sounds like that tracks with the cognitive decline over the 10 years, roughly, that you had been following him, too.

Wendy Kelso 17:12
That's right. That's right, John. So there's really what we call "classic", I suppose, MRI caudate or striatal atrophy on imaging. Sometimes we get people to have PET imaging as well, which obviously shows reduction in terms of the basal ganglia, but usually MRIs are the most freely available in terms of imaging in this cohort.

Ryan Van Patten 17:38
Let's transition and talk about Huntington's disease more broadly using your case that you presented to us as a launching off point but then thinking about the condition generally. So, for our listeners, Huntington's is an autosomal dominant

trinucleotide repeat disorder. So, autosomal dominant, meaning if one's parent has the Huntington's gene then there's a 50% chance that you will have it. You alluded to, Wendy, that he had 46 CAG repeats. Fourty or more CAG repeats on chromosome 4 leads to Huntington's disease. My first general question to you is if you would talk through the Huntington gene, the protein, the pathophysiology - what do we understand about this disease?

Wendy Kelso 18:33

In Huntington's disease, you're considered to have symptomatic Huntington's disease if you have greater than 40 CAG repeats. What we call a zone of reduced penetrance, or increased risk range, is from 36 to 39. So if you have CAG repeats in this range, the majority of people will manifest the disease within their expected lifetime, but potentially may have slow progression or less severity, and if they live long enough, they may not actually show the disease in life. What we call intermediate alleles on the Huntington's gene, on chromosome 4, are between 27 to 35. These are usually associated with what we call the normal phenotype. So the person doesn't develop Huntington's disease in life, but they are prone to changes and it's possible that the next generation may inherit Huntington's disease. You're more likely to have an earlier onset of disease if you inherit the gene from your father versus your mother, so paternal versus maternal transmission. If you have less than 26 CAG repeats, you're considered normal and that you will not develop Huntington's disease. There's a rarer form of juvenile Huntington's Disease, which you see in children, and they generally have significantly increased CAG repeats which are greater than 60.

In general, Ryan, the number of repeats is related to the severity of the disorder. So the higher the number of the CAG repeats, the earlier the age of onset with Huntington's disease and the more significant the disease pattern. We've seen people here at the clinic that might be in their late teens or early 20s, where a parent's had a significant number of repeats but they've inherited from their father and then they develop the disease 5 or 10 years younger than the parent and become very symptomatic. Often these people have CAG repeats in the 50s or 60s. The majority of people have CAG repeats probably in their 40s, and those people generally develop the disease in the age of their 40s to 50s is the most common.

We're talking about Huntington's disease being a trinucleotide repeat disorder. So it's the CAG, or the nucleotide letters, that form the code that's read in a group of three. For Huntington's disease, it's cytosine, adenine, and guanine. These have too many in Huntington's disease. So, for example, it's caused by stretch of the letters in the CAG gene, which is repeated too many times. If you have, for

example, under 26, you have CAG under 26 times, but with Huntington's disease, the CAG repeats reflects a number of repeats. So, for example, if you have 60, then you've got 60 CAG repeats which is an increase from normal and this is considered toxic. The mutant Huntington protein forms clump in brain cells and causes them to become damaged over time and die. The most vulnerable part of the brain in Huntington's disease is the striatum, which controls movement and mood and memory. It's the damage to the striatum, particularly the caudate, over time which causes the symptoms of Huntington's disease.

John Bellone 22:08

That follows what we had talked about for your patient. Over time, the degeneration on imaging became more and more prevalent. Can we talk about the ethical issues around genetic testing and genetic counseling in general in people with Huntington's and their biological relatives? It's extremely relevant here, the genetic testing and counseling aspect.

Wendy Kelso 22:32

Yeah, sure, John. We have what we call a predictive clinic here at the hospital. When someone knows that they've got a risk of Huntington's disease - because either parent has got the disease, or it may be that one of their parents has died early, or there's non-paternity but there might be Huntington's disease in the extended family, so for example, in a sibling or an aunt or an uncle - those people therefore, tend to come to neurogenetics and have counseling to determine whether they want to find out if they have the gene or not. As it's autosomal dominant, if they have the gene, they've got a 50% chance of inheriting the disease. And it is fully penetrant, which means that if you have the gene you'll get the disease.

There's a number of risks and benefits about knowing if you have Huntington's disease. The benefits are that people can plan their life. Often people worry excessively knowing that they're at risk and so would prefer to have some certainty and some control over their future. The risks are obviously that you can find out that you are going to develop a terminal neurodegenerative illness with no cure often many, many years before you actually become symptomatic. The risks are that you may, for example, have predictive testing in your early 20s and you may not develop the disease until you're in your late 40s. So you've got two decades knowing, where you're constantly hypervigilant for symptoms when you may not develop symptoms till much later. Often people that find out that they do have the gene make significantly different life choices. It affects choices in life such as will they get married or not? Will they decide to have children or not? If they do decide to have children, will they then go through the IVF process so the embryos that they

decide to have will not have the HD gene? So there's a number of different ethical decisions that are important to know.

I think one of the other things that's important is that we often see people when they come through to the genetic testing process where we have to assess risk about whether they are psychologically suitable to go through the genetic process. Some people if they've got a history of very significant depression, the family is unstable at the time, there's a risk of self-harm, or they don't have enough support, often those people are advised or counseled that maybe now is not the time to go through genetic testing and it would be better to delay the process until they're feeling more psychologically stable or there's enough supports in the community that if they found out that they were positive, they would be able to manage that. The risk in terms of suicidality for Huntington's disease is quite high. It's been estimated up to 10% of people with Huntington's disease actually commit suicide. The most common periods for that to happen are around the time of finding out you're gene positive and then very early on in the disease process itself. When the disease continues over time, people have a reduction in insight, which means that the people around them often have great difficulty looking after them and there's a lot of grief and loss, but the person with the disease themselves doesn't have full awareness anymore so it's less psychologically distressing as it was in the early stages of the disease.

Ryan Van Patten 25:55

Helpful overview, thank you. This is obviously a very heavy topic and probably made more so by your presentation of the case initially, where we hear about your patient having three children, two of them were aware of a diagnosis and then a grandson. For the patient you talked about and just in general the patients you see, by the time they come to you do they typically know that they have Huntington's? Or they know that they might have it, they understand that they have a 50% chance of having it based on a mother or father getting it? What's your understanding for a lot of the patients you see the time course of their awareness and understanding of this disease?

Wendy Kelso 26:42

It varies enormously, Ryan. For some people, when there's a younger onset of Huntington's disease in generations of families - for example, people become symptomatic in their 20s, 30s, or 40s - the majority of those people often know that there's a family history and, hence, are well aware and have looked after family members for generations. There's considerable grief and loss and huge anxiety about developing the disease, particularly if they've had direct involvement of caring for a family member with Huntington's Disease themselves. Often these people are

caring for a parent, whether they're in their teen years, a very difficult time to be looking after a parent. Because of the nature of the disease and because it's progressive and it affects motor, cognitive, and behavioral changes, it can be quite a difficult condition to look after people well. It also can be really distressing because the person that they thought was their parent has changed into terms of personality over time. Towards the end of the disease, there's often marked behavioral changes and personality changes. So the mom or dad they're looking after aren't the same mom or dad that raised them to begin with.

If there's later onset of the disease in the family - so I talked before about if people have lower CAG repeats through generations of family, sometimes you may not have the disease. A person may only manifest the disease in the 80s or they may never have actually had the disease in life, but then the child has a disease earlier. Those people often are less aware of their risk because the parent may not have actually had the disease in life or the parent may have died early. There's often not good education from their point of view about Huntington's disease and they don't exactly know what's going on.

In some families, particularly historically, Ryan, Huntington's disease had a huge amount of stigma around it. So families hid the disease from other family members. When they were getting married, they often didn't tell the person about the risks involved because it was considered something that had no treatment. There was a lot of mystery around the disease, particularly in historical times. A lot of people with HD were often found to be very disabled over time where they were put in psychiatric institutions due to changes in behavior. So there was a lot of stigma around that presentation.

John Bellone 29:16

I'd like to talk a little bit more about the typical motor symptoms involved, especially how this is a hyperkinetic syndrome which differs from Parkinsonian conditions such as Parkinson's and PSP, CBS. Can you talk about the typical motor symptoms for HD?

Wendy Kelso 29:38

Yeah, sure, John. The typical motor features, what we call chorea, are really "dancing movements." It used to be called Huntington's chorea, where people have involuntary movements that are unintentional, choreoathetoid movements, and they don't have control. These can start off sometimes looking as twitching or jerking and they can be quite frightening for people that do not know about Huntington's disease to become used to in the clinic. You sometimes find, particularly with people with Huntington's disease when they progress and they may need more

supportive residential care, that the people caring for them are concerned about them because they often make unintentional movements that are quite jerky and twitchy and they may look like they're actually trying to lash out when they're not at all, but they just don't have control of their movements.

It's quite different from Parkinson's disease, where there's really a lack of movement or a great slowing of movement. You also do get slowing of movement in Huntington's disease, but chorea is the most common feature. It's often seen in the face, in the trunk, and in the arms and legs. I often look, for example, if I'm seeing someone with Huntington's and it's early, I often look under the table, look at their feet to see if their feet or toes are actually twitching as one of the common symptoms. Another common symptom is that people have chorea particularly in their hands and arms, but they try and hold their arms or close their arms together. They try to minimize the chorea so it looks more socially appropriate on interview.

John Bellone 31:17
Did your patient show signs of the choreiform movements?

Wendy Kelso 31:22
That's right. So, John, at the initial assessment, not so much. But there were very early changes in swallowing that were evident and slight slurring, and it obviously affects swallowing as well. The sort of classic features of Huntington's disease is that they have changes to eye movements. They have what we call changes, or saccades, where they have difficulty initiating saccades, or horizontal eye movements. They can be quite slow to do that and that can sometimes give them an appearance where they look like they're also staring or they have difficulties with their eye movements. They often have difficulties with swallowing and also dysarthria, control of motor speech. And choreiform movements in the room, where it can also mean that if you're giving them some neuropsychological tasks, they have the tendency to be clumsy and they drop things or things move around the room because they unintentionally can't control and things fly out of their hands. Often people in the initial and early stages are quite self-conscious about these types of movements and try and hide them from other people. But as the disease goes on, they lose insight and they don't see the chorea as bad as other people around them.

Ryan Van Patten 32:44
Another motor symptom, just for our listeners to be aware of, that commonly happens in Huntington's disease is motor impersistence, where the person is not able to maintain a static posture for a while. A neurologist might test this with a tongue protrusion, having the patient stick out their tongue and if the patient is

unable to hold it there for a duration of time. Thank you for describing some of the motor features of Huntington's.

Another big and important clinical aspect of the condition would be neuropsychiatric symptoms, Wendy, which you talked through regarding your patient. In the literature, I've seen a wide variety of different psychological or neuropsychiatric symptoms that are possible in this condition. We've talked about apathy, depression, you mentioned grieving, coping with the diagnosis, suicidality. There's also irritability, aggression, and anxiety. It can get more severe with psychosis, mania, disinhibition, dysexecutive symptoms, OCD spectrum. You mentioned loss of awareness. There can be disturbance of sleep and the sleep-wake cycle. What would you like to tell us about these symptoms? How frequent are they and how do they impact quality of life?

Wendy Kelso 34:03

Probably the neuropsychiatric symptoms and the behavioral changes are the most disabling for people with Huntington's disease, much more disabling than the motor symptoms combined with the changes in cognition. Often, the changes in cognition start quite early. The most common neuropsychiatric symptoms at the beginning are often apathy and depression - probably the two most common. There's varying rates of depression in the literature, but it's extremely common in people with Huntington's disease and there's different reasons for that. Some people believe that people with HD are more likely to have what we call a biological depression due to the changes in the neurotransmitters in Huntington's disease. But obviously, there's also reactive depression due to loss of function, changes in relationships, and due to changes in personality and empathy and social cognition. It can mean that staying in a relationship is more difficult and there's higher rates of divorce and higher rates of intergenerational difficulties between family members.

In terms of when we get towards the later stages of the disease, there's increased behavioral change. Very significant insomnia, as you spoke about, where people have great difficulty sleeping at night and often tend to sleep a bit more on the day. About the significant behavioral changes, as the disease gets more severe over time, changes in people can become much more aggressive and much more irritable than they were before. They can become much more disinhibited and they often tend to get stuck in set and have some stereotypical behaviors and perseveration and utilization behavior. They require a lot of assistance from family members and paid carers to actually try and change their track of thinking because they are unable to change it themselves. They require considerable changes to the environment, to be able to modify it, to function at their best and use some

techniques from positive behavior support type of therapies to try and increase the quality of life and reduce behavioral changes.

Ryan Van Patten 36:18

Let's talk for a few minutes about management of Huntington's treatment, if you will, a treatment as symptomatic not curative. As John had mentioned earlier, Huntington's is a hyperkinetic syndrome so there is excess movement, the chorea, which differs greatly from hypokinetic syndromes, which are the other Parkinsonian syndromes we commonly think about. In Huntington's, because of the nature of the pathology in the basal ganglia, there's actually an excess of dopamine in the nigrostriatal system instead of a dearth of dopamine in PD. This is going to impact pharmacological treatment. We don't want to increase dopamine in Huntington's disease. Also, if we consider symptoms like psychosis, that's another reason to not increase dopamine. Generally stepping back, when someone comes to you and you go through a neuropsych eval and at the end you're talking through various treatment options, imagine that they haven't heard all this from a neurologist already. Medical, nonpharmacological, what options are there?

Wendy Kelso 37:27

It depends a little bit on the type of symptoms, Ryan. Often, for chorea, one of the treatments is tetrabenazine and some people also give Risperidone. Risperidone can also be given to try and modify some of the behavioral changes in small doses. The only difficulty is if you give too much Risperdal then you get some phrasing and more of a Parkinsonian type of features, which you have to be concerned about because otherwise it increases the risk of someone falling if they become too stiff.

In terms of treatment for cognition, usually what we do in the early stages is have more of a cognitive remediation approach or compensatory approach to cognition. Some people do brain training, but most people find that the most useful are people in their support team changing the environment around them and using compensatory techniques in the early stages to maintain them at work. We often have quite a lot of contact with someone's workplace if they have spoken to their employer that they actually have Huntington's disease to try and modify the work duties or modify the type of work roles to be able to then maintain their employment as long as possible. Those types of things include allowing more time, working part time, making sure that things are written down, and showing rather than talking to them in terms of learning new procedures. Due to the changes in speed of processing, often people with HD take longer to take on new information and learn things. Often these things have to be accommodated for.

In terms of behavioral changes towards the later stage of the disease, again, it's usually non-pharmacological management is preferred. Often people are put on a range of different medications from antidepressants, antipsychotics, to try and change behavior, but there's not always good evidence for all of them. Often we find that in residential care facilities where they don't have much experience treating people with Huntington's disease, they're put on a cocktail of medications which can cause a person to become quite sedated to try and modify some of the challenging behaviors. But this can very significantly impact the quality of life for the person.

John Bellone 39:49
Is it common for neuropsychologists like yourself to be involved in a Huntington's disease clinic? That's my understanding - that you're in a HD clinic. Is that right?

Wendy Kelso 40:00
Yeah, that's right, John.

John Bellone 40:01
I'm curious about your role and if you're involved in the intervention, or is it mainly assessment of the cognition?

Wendy Kelso 40:09
So, John, our role changes a little bit through the disease process. We often see people at the very beginning, and it's probably different from other types of neuropsychology because for most people with Huntington's disease, the disease is known. So it's not diagnostic clarification. We know what the disease is. It's much more about staging in the early stages. When people know that they've got a gene for HD, we do a baseline assessment. Then we use that to try and find out when they're symptomatic over time. That can be helpful when people are trying to plan families and trying to plan work if they notice a couple of symptoms and they've had a baseline assessment. Cognition is often one of the first symptoms that changes, and it can be much more sensitive than early imaging changes and certainly before motor symptoms. So if we think that there's changes in the cognitive ability, we say, "Look, we suspect on the basis of probability that you're becoming symptomatic. It may be many years until you have significant symptoms, but you may want to think about the future and do future planning at this stage."

Towards the middle and end stages of the illness, people don't often have to have more ongoing neuropsychology done, but they would often get support with behavior. Also, we often provide education to family members, to the person with

HD themselves, and often to residential care staff that have absolutely no experience in Huntington's disease, giving an explanation about cognitive changes, psychiatric changes, and motor changes, what to expect and how to manage it the best, and also trying to get them to put themselves in the position of the person with Huntington's disease. Imagine what it must be like for someone that is living with a disease in midlife and having these significant changes and also no control over their future.

In terms of the role in the clinic, we're also involved in cognitive remediation, as I spoke about, in the early and mid stages. Once people come very symptomatic, often it's more about symptom management and the main things are focusing on things that keep people alive, such as swallowing, not falling, nutrition, balance, bowel and bladder function - more of those core types of features where we provide education to the family. Much less cognitive testing at that stage.

John Bellone 42:28
That education and intervention piece is so important for both the patient and the family, like you mentioned. Any other recommendations that you made to your patient that we talked about that you wanted to mention? Are there - I'd imagine support groups, organizations that would be helpful to become a part of?

Wendy Kelso 42:48
There's Huntington's Disease Associations across Australia and across the world and they're often a fantastic provision of education support for families. They often have support groups involved as well, where people can go along with living with Huntington's disease and also for family members to learn more about it and to gain support from people that have a shared experience of living with a rare neurodegenerative condition. In Australia, we also have the National Disability Insurance Scheme for people under the age of 65, which provides financial support to maintain people's independence and maintain a good standard of living. That will fund activities such as speech pathology, occupational therapy, physiotherapy, neuropsychology, counseling and clinical psychology, and a range of other activities. We often get people to support them in applying for that so that they can actually have the best possible life they can. That can also help people have someone come and take them out on outings when they lose their license, be able to have a much more enriched type of environment to keep them at the best they can for as long as they can be.

Ryan Van Patten 44:00
Great. I think that's good for case number one. That was excellent, Wendy. Thank you.

Transition Music 44:05

John Bellone 44:09
Well, that does it for our conversation with Wendy. If you'd like to support what we're doing here, please leave us a rating on whatever podcast app you're listening to this on. And, as always, thanks so much for listening, and join us next time as we continue to navigate the brain and behavior.

Exit Music 44:25

John Bellone 44:50
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Ryan Van Patten 45:01
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