55| Neuropsych Bite: Moyamoya – With Dr. Joel Kamper

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Speakers: Joel Kamper, 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 00:25

...and I'm John Bellone. Today we have a Neuropsych Bite on a rare and fascinating brain disease, moyamoya. We talked with Dr. Joel Kamper, a board certified neuropsychologist at the James Haley VA in Tampa, Florida.

Ryan Van Patten 00:39

Today is our second Neuropsych Bite with Joel. These episodes are meant to be brief summaries, providing broad overviews of rare neuropsychological conditions. We hope that they can be of some help to both students and professionals who may encounter these conditions in clinical practice. We already covered Creutzfeldt–Jakob disease, or CJD, in episode 44, and we will be releasing two more episodes with Joel on Balint's syndrome and limbic encephalitis.

John Bellone 01:09

Before we get started, we want to give a brief reminder that select NavNeuro episodes are available for CE credits through INS. If you need CE's check out navneuro.com/ins for more information. And, with that, we give you our Bite with Joel Kamper.

Transition Music 01:25

John Bellone 01:34

All right, so moyamoya, or “puff of smoke” in Japanese, is a disease. Can you tell us about the neuropathological and clinical characteristics of moyamoya?

Joel Kamper 01:45

Absolutely. So it's a chronic progressive disease involving, typically, the arteries around the Circle of Willis. They call it "puff of smoke" because there are little occlusive lesions and, on angiography, it looks like a smoky, hazy presentation in the basal ganglia and the vessels around the Circle of Willis. It's relatively rare and more commonly seen in Asian populations. The word moyamoya itself is Japanese. It’s not exclusively seen in Asian populations, but there's a much higher incidence and prevalence in those populations.

John Bellone 02:25

What's the typical cognitive profile look like?

Joel Kamper 02:27
You know, there isn't a lot out there, neuropsychologically. In the research that we've looked into, it looks a little bit subcortical and diffuse, but with some focal findings. It's not like a major vessel stroke, but let's say you had an aneurysm that didn't catastrophically rupture, but ruptured a bit, and they went in and repaired it. So you had this diffuse, focal profile. It's kind of like that, but it tends to get worse over time. In the case I'll talk about in a bit, the patient was a veteran that we actually saw serially, a couple times over a number of years, and you could see some changes.

Ryan Van Patten 03:10
My understanding, which is similar to what you said, of the moyamoya "puff of smoke" is that the carotid artery tends to be blocked, or at least restricted, and then you have a number of small vessels that are opening up in its stead. And, on imaging, that looks like a puff of smoke. Is that accurate?

Joel Kamper 03:27
I believe so. Yes.

Ryan Van Patten 03:30
What are the incidence or prevalence rates of moyamoya? You mentioned that people from East Asian countries are most at risk, but it can happen to anyone.

Joel Kamper 03:39
Well, let's go with prevalence because I think that's more salient. So maybe about 5 per 100,000. So, not the most rare but certainly not common. There's a female predominance, about 2 to 1 or so. Interestingly, there is some genetic risk here. So 10 to 12% of patients are going to have a family history. And there's been some research about certain genes that may predispose someone to have this, which would make sense as to why it's seen in certain demographic populations but not others.

John Bellone 04:17
And it's a higher percentage of children that have it, too, I believe.

Joel Kamper 04:20
Well, because it's basically a genetic condition, it's often picked up on in childhood. Yes. I don't see children, so...

John Bellone 04:29
Right.

Joel Kamper 04:29
The few patients we've seen with it, it's picked up in adulthood. There is a second spike I think around age 40 or so. But otherwise, I think the peak age of symptom presentation or diagnosis is around age 10.

John Bellone 04:43
Right. Yeah.

Ryan Van Patten 04:44
Yeah. And the proximate cause of cognitive impairment are the strokes, right, to your knowledge?

Joel Kamper 04:50
Right.

Ryan Van Patten 04:51
Do we typically detect moyamoya only after someone has had a number of strokes? Or is it sometimes detected before the actual cerebral event?

Joel Kamper 05:00

It can be before the strokes, but they're going to have some symptoms. So let's say you have a 10-year-old who has a seizure, or has what they think is a stroke, but ends up being a TIA or something like that. It’s every parent's worst nightmare, right? You bring your 10-year-old in thinking it might be a tumor or something like that, and they don't find that. If they do a CTA or some sort of angiography, they can look at the vasculature and find it that way. Sometimes either an ischemic stroke or a hemorrhagic stroke is the presenting symptom. I would imagine in adults that's probably more likely.

John Bellone 05:40
Right. And there can also be recurrence. TIAs, transient ischemic attacks, too; it can look like a mini-stroke of sorts.

Joel Kamper 05:48
Correct. Yeah, I mean, sometimes these folks will have specific neurologic syndromes - you know, dyskinesias, dystonias, things like that. I haven't read into that myself or seen that too much, but there's some literature that suggests that. In a lot of ways, it's like a watershed infarct. So, for those who see more large vessel strokes, the pathophysiology is completely different. But that's kind of how it looks to us on our testing.

John Bellone 06:16
Is that a way to differentiate it, you think, from strokes that are not due to moyamoya?

Joel Kamper 06:21
Yeah, so it's unlikely you're going to have some of those big major vessel symptoms like you'd have in a large stroke. You're not going to get hemiparesis or neglect, some hard parietal signs, or speech loss like aphasia - none of those classic stroke signs or really big things that we look for. It's going to be more of the subtle stuff where it looks diffuse. But then they have some ideomotor apraxia or three step motor sequencing impairments, and you're thinking, "Oh, that's kind of weird." Could be executive but then there's some really specific things on testing, but it doesn't really fit a full subcortical pattern. It just looks weird. That kind of thing.

John Bellone 07:04
This would be hard to really pin down.

Joel Kamper 07:07
Yeah. The cases that I've seen or know of, that others have seen, all were diagnosed before they came to us, so that makes it easy. But for listeners who are doing board prep or thinking of a fact finding, it's something to consider when you have a weird case that you think could be vascular, but it doesn't fit MCA stroke or something like that.

Ryan Van Patten 07:31
That would be a rough fact-finding case if that was the answer.

John Bellone 07:34
Oh my gosh... [laughs]

Ryan Van Patten 07:36
Hopefully that person had just listened to this episode before going through that.

Joel Kamper 07:42
One of my colleagues, as a practice, gives students a case of osmotic pontine demyelination, which is mean. No one ever gets it. Which isn't the point, and that's sort of the lesson to drive home. But...

John Bellone 07:55
You said that was a mock session?

Joel Kamper 07:57
Yes.

John Bellone 07:58
[laughs]

Joel Kamper 08:01
Real cases are not quite that mean.

John Bellone 08:03
Yeah. Good.

Ryan Van Patten 08:04
A quick follow up. So the cases you've seen have already been diagnosed. Is this typically diagnosed through angiography or MRI, or how else?

Joel Kamper 08:12
Yeah.

Ryan Van Patten 08:12
Okay.

Joel Kamper 08:13
You know, just like anything, there's the occasional time when a patient may walk into your office complaining of headaches, and you know, this is seen

predominantly in women. So you get a 40-year-old woman coming into your office saying, "I have headaches. I've been having these dizzy spells", and you're thinking, "Okay, what's going on?" And the testing is just kind of weird. It's possible, you know. You could then refer to a neurologist who would do more tests and then diagnose it. But, typically, the referral will be “This patient has this condition and is reporting these symptoms, do those fit? Or could there be something else going on?” That sort of thing.

John Bellone 08:46
It might have been even diagnosed in childhood, and then we're seeing someone as an adult. Much later.

Joel Kamper 08:50
Right. Yep.

Ryan Van Patten 08:51
Yeah. And as Joel has said a number of times before with rare syndromes like this, they're very interesting, but sometimes if we don't know the answer, it may be tempting to look for the zebra, when we should look at base rates, too.

Joel Kamper 09:05
Absolutely.

Ryan Van Patten 09:05
Like, right now we're talking about moyamoya and it's important for people to be aware of it, but it's low base rate. It’s unusual, rare, and interesting. So if you're seeing a case, most likely, this isn't the answer just based on probability.

John Bellone 09:19
We should give some context to the “zebra”. So if you hear the sounds of hooves, you should assume it's a horse, right? If we're in North America, you shouldn't assume it's a zebra. So it’s much more likely it would be something that's more common - a large vessel stroke, let's say, or vascular dementia or something. You shouldn't automatically go to the very rare condition, which we call a "zebra". Just some context there.

Joel Kamper 09:46
Right. And there are people who are "zebra hunters". Certainly not in our field, but often some of our physician colleagues...

John Bellone 09:54
Like Dr. House. [laughs]

Joel Kamper 09:55
Yeah, I know a couple. We had a couple people who have since retired but they had the reputation where it's like, okay, everyone's got something weird. And, it's like, well, no.

Ryan Van Patten 10:07
Right.

John Bellone 10:07
Most of the time, we're gonna be wrong, right? [laughs]

Joel Kamper 10:09
Yep. When you hear hoof prints think horses, not zebras. That's very important.

Ryan Van Patten 10:14
So, moving forward in our conversation, what else is important for neuropsychologists to know about moyamoya? And how can we be of help to these patients?

Joel Kamper 10:23
I think because it's rare, they might get a diagnosis - and it's a definitive diagnosis, we know what's going on - but they're not going to have that lightbulb moment of "Oh, this explains the symptoms you're having". And especially since it tends to be seen in middle-aged women, I think sometimes it's these patients who have been through - you know, they get diagnosed with fibromyalgia and all these other things, and they kind of have this "No one's taking my symptoms seriously." Even though they had this diagnosis, they haven't had that connection of "No, these are real symptoms and it's related to this." It's amazing to me how people will say - and maybe it's just memory loss, but I believe it - but they will say, "No one's really ever explained it to me like that". Kind of validated and explained what's going on, and

why, and how it's not something else. It's this and here's why. I mean, like with anything, patients really appreciate that explanation.

John Bellone 11:23
Yeah.

Joel Kamper 11:25
But often, for whatever reason, a few of the neurologists that I work with like to send these sorts of folks to us to help corroborate the diagnosis. You don't need a neuropsych to diagnose moyamoya by any stretch. But I think it's just to lay eyes on them, we spend more time with them, we can get that history. We can explain what the neurologist thinks is happening, and we can see if the profile fits with the limited neuropsych data we know.

John Bellone 11:49
Yeah, great. So let's talk about your case a little bit. Tell us what the presentation was and what the cognitive profile was.

Joel Kamper 11:58
Yeah, so I had a female veteran, middle-aged, who had the diagnosis. So this wasn't one where we got to help track that down, but that's just fine. I had the diagnosis but she was having additional symptoms. I think she had had it for, let's say, 5 or 6 years. She was having some additional symptoms and they had done a repeat angiography and said, "Well, there's no change." So what's going on with the symptoms? And, you know, that can certainly happen, but the neurologist hadn't taken that leap. So they sent her to us. She walked with a walker, quite slow. Not like a Parkinson's patient slow, just kind of carefully. Carefully is a good way to put it. And we got a typical subcortical profile with some odd executive-type stuff. You know, she didn't present as terribly dysexecutive, most of the scores were okay. But there were a couple of weird focal findings. I think she had some dysdiadochokinesia, which is one of my very favorite words. It's problems with rapid alternating movements.

John Bellone 13:03
Your hands, usually.

Joel Kamper 13:04
Yes, exactly.

John Bellone 13:05
Up and down. Palm up, palm down. Yeah.

Joel Kamper 13:07
Yep, there's different ways to do it. The "palm up, palm down" is the one that I do. It's basically stopping one movement and starting another, and it localizes typically either to the posterior frontal areas or it can be cerebellar. But she had a couple of those sorts of neurobehavioral signs. And it was like, "Well, that's weird". And then she was a little slow and a little dysexecutive. So we put our heads together, consulted the literature, it fit pretty well with what we'd expect. She had never had a neuropsych evaluation before and I think she had some additional life stressors and sleep problems that were relatively recent and that explained why she thought the symptoms were getting worse. She had never had a baseline study done with us, so we got to explain, "Yeah, these symptoms are there. They're real."

John Bellone 13:10
But, that's nice, though. That you could parse out some of the effects of sleep and mood, and that's a perfect point of intervention then for you.

Joel Kamper 14:03
Yeah, thank you. I harp on that. I love when we're able to take something like that, that's muddy, and be able to parse out, “Well, this could be due to the moyamoya, but this aspect is probably related to the sleep, and this to the mood.” You can't always be that fine grained, but it's nice to be able to provide that level of detail to folks.

John Bellone 14:20
Or at least the intervention. I mean, we can't do anything about the moyamoya, but there are some great interventions for insomnia or depression.

Joel Kamper 14:28
Right. There is hope. People want that hope for improvement. Yeah. So she came back about 5 years later for a follow up. We hadn't recommended to follow up unless needed, but she was concerned things were getting worse. She had had a couple events between when we saw her the first time and the second time, some little strokes. She was definitely worse. A lot slower. Looked almost - I mean no tremor or anything, but the cognitive profile looked a little Parkinson-like. She was very, very slow. A little dysexecutive - poor attention, that sort of thing. But, again, some of those focal neurologic signs were still there. She did relatively better on

some of the more formal executive measures, which makes sense, because this is often around the Circle of Willis and basal ganglia, so you're not going to hit those dorsolateral prefrontal areas as much. You could hit the pathways, which is that subcortical profile, but you're not going to hit them directly. So it fit, but definitely worse. And since we had done that initial study, we could say, "Well, looks like you've had declines in these areas. And that makes sense. How are you sleeping?" [laughs]

John Bellone 15:32
Right.

Joel Kamper 15:33
Still room for intervention. Always. But we were able to make that comparison, which was nice.

John Bellone 15:39
Okay, Joel. Well, thanks for talking through that case. It was really helpful.

Joel Kamper 15:42
Absolutely.

Transition Music 15:42

Ryan Van Patten 15:47

Well, that does it for our conversation with Joel. Be on the lookout for future Neuropsych Bites on Balint's syndrome, limbic encephalitis, and several rare pediatric disorders as well. As always, thanks for listening and join us next time as we continue to navigate the brain and behavior.

Exit Music 16:05

John Bellone 16:28

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Ryan Van Patten 16:40

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