MAPS Webinar Introduction

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Hi, welcome to the MAPS webinar series, Healing Tomorrow's Future. We are thrilled to be bringing you this series packed with valuable information and education within our community.

My name is Honey Rinusella and I'm the Executive Director of MAPS, the Medical Academy of Pediatrics and Special Needs. Within these webinars, we're able to empower clinicians on the knowledge and tools to support patients facing a variety of health challenges.

For more information on MAPPs or to register for a conference, please visit us at www.medmaps.org. That's www dot medmapps dot org. Thanks for joining us on this journey towards a brighter, healthier future.

Precision Genomics vs Genetic Testing

Hi, I am Dr. Emily Gutierrez and I'm a doctorally prepared nurse practitioner and i am part of the clinical education team for Intellix DNA. So we do look at precision genomics and have a tool to help clinicians make decisions with their patients.

The number one question that people have in mind is, how is this different from genetic testing? Well, genomics is different because you're looking at more common variants in the population.

So there is pathological genes that infer a great probability you will have pathologic disease. And then there are common variance that we have in population that can influence a pathway.

When you can come around those pathways with epigenetic modulation, you see improved patient outcomes. So some people think autism is a lifelong diagnosis or condition.

What is it that Intel X DNA can do to like modify that? Well, as you probably know, the earlier the intervention the better the outcomes. So knowing your genomic pathways and what variants you might have in some of the common pathways, you can come around and really understand what are the primary needs for that individual, things that you should be paying attention to that For example, we'd look at a shank mutation and one of the shinkopathies that we look.

If you give zinc very early on in much higher doses than the registered daily allowance, you can really help with scaffolding of neurons and connectivity in the brain.

So without that genomic data, You may never understand that there's a Shinkopathy that really needs more attention too. Yeah. With that said, is there certain patterns you see in genomics variations?

Common Genomic Patterns in Autism

And children with autism? Yes. Oh, absolutely. I mean, the three big ones to me are oxidative stress, problems with detox, which also plays in oxidated stress and problems, with methylation and foley abnormalities, Which plays into the two of them.

i think of autism as a messy Venn diagram of interconnected set of systems. And there's a lot of pathology in the way our kids are presenting. Our kids don't have a behavioral disorder, they have biological disorder and understanding their genomics gives you a step ahead of really knowing how to approach that individual in what pathway.

I think you mentioned methylation as one of the biological pathways. Are there other biological pathway that need to be considered? Absolutely. I mean, in neurodevelopment, there's many pathways that need to be considered.

What is our immune system doing? What's the nutrition behind the individual? And do you know that there are genomic variances around nutrition? For example, some people might have a genomics variant where they can't absorb magnesium in the gut.

And those are some of my absolutely most chronically constipated kids without any other etiology or cause. So they might require a lot more intervention around having a daily stool.

And as you know, having daily a stool is so important for detoxification. Children with autism are known to have much higher levels of oxidative stress.

So pooping every day is really important. Yes, it is. And it's a topic people don't want to discuss, or some people think it just normal not to go every.

It's strange. Or even twice a day. I want a touch on the subject of pathogenic mutations versus common variants. What's the difference? Well, there's a big difference.

So a pathogenic mutation might be less than 1 in 10,000 individuals, where a common variance might more than one in 100. Telix DNA looks for more common variants, and in those common-variants, things that have plausibility for epigenetic treatment.

A lot of our tool has a gene that you know the mechanism of it, and then mechanistically, we can come alongside it and say, based on the mechanisms of this gene, this intervention is likely to be helpful.

For example, if you have a problem making cysteine, which scaffolds in acetylcystein to glutathione or major antioxidant, you might want to give N-acetylcysteine to really enhance that pathway.

Targeted Testing and Supplement Guidance

N acetyylcystine, depending on your variant, might work better in that individual than somebody that doesn't have that variant. Interesting that you say that.

That could lead to, if I'm saying this correctly, more meaningful clinical outcomes. Am I saying that right? Yes. It's important because in this work of biomedical treatment for autism, You know, parents can be like, well, I heard sulforaphane works really well or there's resolvins or specialized pro resolving mediators.

And before you know it, you have a kitchen full of supplements and their breakfast is all their supplements, and it gets overwhelming. So genomics can really help you understand, what are the top three to five things that they really need based on their genomic that you shouldn't be missing?

Emily, I think you're saying this. I'm going to say it one other way, and you'll probably say yes, of course. But this sounds like precision medicine, where you are analyzing something and it's not a shotgun approach.

Am I saying that correctly? Yes, yes. And not only is it helpful for interventions, sometimes it is helpful to know what testing to order. So, for example, if you have a mannose-binding lectin pathway, which is a gene that helps you identify that there's a pathogen on a cellular surface, and if have you a double-copy variant in this, you've reduced opsinophagocytosis, Which means you just don't clear infection as well.

I might say, oh gosh, with this gene variant, you're more likely to have candida overgrowth, I really want to do an organic acids test and look to see where our fungal markers are.

So it also can help you not to just have every single test, but really to say which tests are leading me to be more important. The ones that I've found to be most important in the tool that i've seen over and over again in my clinical practice is when you need more glutathione and it increases your risk for heavy metal toxicity in those patients really doing a provoked metal challenge, it becomes more essential.

And what I have seen, over, and, over, and, again, is their mercury or their lead will be high, whereas somebody with that variant that wasn't discovered, that might not be one of their oxidative stressors.

Well, autism is definitely a hot topic in the news and the media. It seems like it's a priority of the current administration. But I think there sounds like there's some misconceptions or misinformation.

Can you talk a little bit about that? Oh gosh, thank you for asking me about that. It's so on my heart because yes, I believe CNN came out yesterday saying lukewarm was approved for cerebral folate deficiency, which is incredibly rare.

Only less than 50 cases in the United States and it's linked to the FORL1 gene. So IntelliX DNA does look for the Forl1 in Two copies are a homozygous variant.

It's in 0.4% of the population and a heterozygos variant is in 11.8% or 11 3% percent of population. In my practice, I've recently done a cohort study looking at genomics and frat testing or if they have those antibodies that can lead to cerebral folate deficiency.

Cerebral Folate Deficiency and Leucovorin

And what I found in 143 people were 11 more homozygous, which is about 8%. And of those 8% that were homosigous nearly 60% of them had fully receptor antibodies.

And they were the type that we're binding, blocking or floating. They were more severe. So yes, there's a correlation, but it's so much more common. then the news is saying it's this uncommon thing.

And the new is also saying, leucovorin is hard to resource. That's absolutely not true because there's lots of beautiful compounding pharmacies that have the wrong ingredient.

Let's be clear, Leucavorine is folinic acid. It's vitamin B9. Just a special form of folate. So just to make sure, because some people may be wondering exactly what you said, let me see if my analogy, if you would agree.

The difference between not having a folate receptor and one that's blocked or binding is like kind of saying if walk up to your home, you don't have a door.

If you have no door, then you can't get in. That's different from if the door is stuck. Am I saying that right? I love the door analogy. I loved the house analogy, yes.

So either that methylated folate isn't able to cross through the brain through to the choroid plexus, so we need to give folinic acid, which is leukovorin, the drug, and that goes through reduced foliate carrier, like the back door to that house.

You can get folates into the body, but then it has to continue to be methylated into methylfolate to Do its magic with DNA repair, growth, neurotransmitters, redox status, glutathione, etc.

Well, I'm sure you have helped many, many people. But is there a case that sticks out really well? Yeah, that's a great question. Yes, there's case, and she loves me.

The mom loves to me to talk about his name. Says, use Miles. I want everybody to know his story. And when I started seeing Miles, he was around six years old.

About six months into our treatment, we hadn't done any kind of folate testing or genetics, started having seizures. These were the seizures that she was calling 911. He wasn't breathing.

he had multiple stays in the hospital. And eventually we did the genomics and we also, which led me to do a folate receptor antibody test and he had a floating receptor and a blocking receptor.

This little boy went to the epileptologist, was on two different seizure medications, still having breakthrough seizures. Mom wasn't sleeping. She was like sleeping with one eye open with him.

she was so afraid that he was going to have a seizure and not be okay. We started Luke of Oran and not only within six months of a higher dose than typical because he had such a severe form, did all of his seizures disappear, but he started talking like he'd never talked before.

His language improved tremendously. And then mom got to sleep at night because the seizures settle down and he is thriving. He is doing really well in school and all the inattention stuff that was really a big problem for him too has so much better.

So for Him, Luca Warren was an incredibly effective treatment for his development. Yeah, and we're not trying to diagnose or treat anyone here. And I think you would agree that testing is very important in order to understand what is it that you're dealing with.

It's not, again, a one-size-fits-all, it's a precision medicine. That's right. So there's going to be a lot of people who see this video and they're going wonder, well, how do you reduce their anxiety or uncertainty about this test?

Case Study: Miles' Treatment Response

About which test. About just doing the genomics. Genomics, I like to think of it like this, just because you test doesn't mean that you make something happen.

It's already there. And having that knowledge to me is empowerment. You know, it's empowerment that can come along and do something beside it or because of, or you can really tailor your treatment to help improve the outcome.

So being anxious about what already is there is almost denial to be, right? Just because you're testing to understand where you are at doesn't mean you created something that wasn't already there.

It empowers you to make choices because now you understand the physiology of the child better. Excellent. Yeah, very good. So, Intel X DNA is collecting a lot of data and data sats.

How is this going to change? I mean, it's just adding on to the data set or is something going change in the future with the approach? Well, first of all, we would get informed consent from every single individual to use their data.

And if we did, it would be de-identified and no personal information at all. But we get consent if use anybody's data, I think that it's great that we have so many integrative providers from neurodevelopment to neurocognition using this tool, because our data set is incredible.

I Think artificial intelligence in the future not open evidence outside of the tool but within the tools and you know that having something that can process large language models is going to be really helpful to clinicians to really help their understanding to decrease the burden of I have all this information how do I process it and how to I treat my patients.

And one thing that's in Intellix that so wonderful is they have the hot spot. And so what that is, is that they take the

genes that have highest odds and the greatest and most rare variant within those pathways and they tier them.

So really the top 5 to 10 genes out of that whole report become really important to pay attention to. In a lot of my patients, we just work from the hotspot primarily.

Okay, you said a hot word in that hot spot. AI, do you feel like this is going to be a good integration?

Using Genomics for Empowerment and AI

I do if AI is trained by a team. So open evidence, if you're using Perplexity or ChatGPT or whatever AI model, it can get it wrong. But within a company, If you have a Team training that AI Model, It can become super powerful.

I look forward to innovation within the tool and FelixDNA is at the cutting edge of innovation. You better bet that we're going to see improvements in the Tool in near future.

You know, some very questions I think a lot of parents would have is that, is this covered by insurance? It is not covered insurance, and the cost of the tool depends on which panel that you do.

The wonderful thing is your genes don't change, you know just like a stool test can change. Thankfully, if you one and you have a lots of dysbiosis and inflammation after treatment, If you repeat it, it should look normal.

But your genomics don't change, so you can always go back and look at them over and over again so it holds its value. But you got to also think, you know, the cost of what you're doing now versus the costs of a tool that could help you do it better.

You know therapies in itself, like the cause of having a child with autism is tremendous. So you want to really tear your therapies to what is the most effective because that over time will reduce the I think it's a very worthwhile investment in a child's life.

And in the end, I thinks it reduces your cost over time. Yeah, you said a key word there. It is an investment. You should consider it like that. Because you only have to do it one time, don't have keep repeating it.

So where can people find more information about all of this? So IntelliX DNA has a website. They have providers that you can look and find a clinician that can order a test for you.

A clinician, a certified medical provider, does have to be able to order the test. And there's a depository or a list of providers, that are using Intellix DNA.

You can find one on their site and they will reach out to that provider and say that, you are interested in seeing them. So they can help connect you So the name of the website is?

Intelix, I-N-T-E-L-X-S, DNA.com.

Cost, Access, and Where to Learn More

And on there, do you have also, is there some information like people can read research papers or case studies? Yes, there's lots of different testimonials on the website.

As far as their research depository, I'm not sure what is listed there. The tool is so researched based on every single intervention or multiple resource studies, you know.

supporting it. That's one thing that I really love about the tool is that it's just not, hey, there's this snip and there is no research and, you know, all of it is evidence based, based on research.

So we're not making it up. It definitely is based science. Yes, it not voodoo medicine. No, I wouldn't say it's voodoo

medicine. And Dr. Sharon Housman Cohen, that's one of the founders along with Carol Village, she is a Harvard physician and I would say that she's gone to a woo school or she has woo-woo medicine at all.

Right, right. One more time before we go, please tell us the name of that website. Sure, IntellicDNA, i n t e l l x x d n a dot com. Super thanks, Emily.

I hope people check it out. There is a lot of utility in doing precision genomics. And like I said, it would be precision medicine because it is not a one size fits all, and I hoped everyone will utilize that.

Well, thank you for being here. Thanks for having me. Thanks so much for joining us today. I'm Honey Renaissancella, and this is MAPS. As we heal tomorrow's future, we appreciate you joining on the journey.

We'll see you next time.