Fragile X with Liz Jewel and Monique Smith

Speaker Key:

CB Chantal Boyle

JL Liz Jewell

MS Monique Smith

00:00:20

CB

Thank you for joining me, Chantal, on The Sunflower Conversations. Joining me today is Liz Jewell, who is the family support counsellor at Fragile X Australia, and Monique Mitchell, who is mum to two children who both have Fragile X syndrome. Today we are going to discover what Fragile X is and the impacts that it can have. Welcome to both of you. Thank you very much for giving me your time today.

MS

Thank you.

JL

Thank you for having us.

CB

Liz, if I can come to you, please, first. Can you please tell me, what Fragile X is and what is the testing process in Australia?

00:00:59

JL

Sure. Fragile X is a genetically inherited condition that affects multiple family members. It's estimated that around 90,000 people in Australia are impacted by Fragile X as female or male carriers of Fragile X or living with Fragile X syndrome.

Fragile X syndrome is caused where an expansion occurs in an individual's Fragile X gene. The expansion of the FMR1 gene restricts the production of a protein which is essential for normal brain development, resulting in developmental delays and other characteristics associated with Fragile X syndrome. The gene change associated with Fragile X syndrome is passed on through a female parent who is a carrier of Fragile X.

In terms of testing in Australia, Fragile X syndromes is diagnosed by a DNA test of a blood

sample or a saliva sample. A GP or a paediatrician or a clinical genetic service can order the test and the results are usually made within a few weeks.

DNA testing for Fragile X syndrome is recommended to be included as part of a basic medical genetic assessment where a child has intellectual disability or developmental delay. GPs or paediatricians will also generally recommend a Fragile X test for a child who has a diagnosis of autism spectrum disorder, as Fragile X is thought to underlie up to one in 20 cases of autism, around 6%.

CB

It sounds as though there's a really good system in Australia, screening system to pick that up in an individual with the... And you said saliva test as well, so they can collect that. Is that through the DNA of the saliva test?

00:02:47

JL

Yes, a cheek swab. Some individuals are not comfortable with the blood test, so a cheek swab as an alternative is the more comfortable approach for them.

CB

What are the comorbidities that are commonly associated with Fragile X?

JL

Well, ASD, autism spectrum disorder, ADHD and epilepsy are commonly associated with Fragile X syndrome. Because the syndrome is a spectrum, an individual may present with a wide range of characteristics and vary from one individual to another.

The characteristics include things like developmental delay and learning challenges, as well as social, language, attentional, sensory, emotional and behavioural difficulties. Autistic behaviours and low muscle tone and difficulties with fine and gross motor skills. Anxiety including social anxiety is also very common. It's a very common trait in females and males. Behaviour and communication in social settings can present a challenge.

CB

That's interesting. Well, it's all very interesting, of course. But what I just picked up there was about the muscle.

JL

Yes, their muscle tone.

CB

Tell me a little bit more about that.

JL

I think in the early stages it's something that might not... Well, when I say early stages I'm referring to babies, essentially. It might not be picked up immediately because developmentally children do grow and develop at different rates.

00:04:24

This is something Monique could probably talk to more than me, but families or parents start to notice that their child perhaps isn't developing initially perhaps through feeding, for example. A baby is having problems feeding initially in that infant stage. That's perhaps an indicator. Then as the child becomes older, looking at their fine motor and gross motor development and being aware that something might be delayed quite significantly.

CB

Is that associated for people who have dyspraxia? Is that the same kind of thing? Do you know? I don't know if you call is dyspraxia in Australia. Do you call it dyspraxia in Australia? I know in America they call it something slightly different.

JL

Yes, we do. But whether it's a direct correlation, I don't know, to be honest.

CB

Okay. Monique, can you explain to me, the family gene, it's obviously... Is it genetic? Would we describe it as a genetic disorder?

JL

Yes, a genetic disorder or genetic condition.

CB

Who in your family is affected?

MS

I'm a carrier and my mum is a carrier. It was passed down to me from my mom. We haven't been able to trace it back too far, obviously. But it looks like it would have come from my grandmother, so my mum's mom, just based on the testing that's been able to be done in the family with the living relatives. My mum and her siblings as far as we are aware are all carriers, so male and female, which is why we think it's come from my grandmother.

00:06:14

I'm a carrier and both of my children are full mutations. They have passed the number of repeats that would put them into the category of having Fragile X syndrome and they are the most affected. Obviously, once that diagnosis came through, we notified the family so

the cousins and everyone could go and get tested. That's where it's come...

CB

Have any of the cousins, have any of them tested positive for that?

MS

As far as I'm aware, and obviously people aren't always willing to share their diagnosis or even willing to get screened, so from what I understand I do have some cousins that have come up as carriers. As far as I'm aware at this stage, there aren't any other people in the family with full mutation.

CB

This is a question to both of you. Is there something that we know that we found out through research which triggers the mutation?

MS

Liz might have more info, but from my understanding from what I've been told by the genetics team is that when women pass it down it's less stable, so the repeats tend to get more. If you're say a carrier with 80 repeats and you're a female and you pass it down to your children, it's likely to be much greater than that.

CB

I see.

00:07:41

MS

It might be 120 or it might be 200. My count is about 107, I think, and the kids are well over. I think 200 is the cutoff. They're well over that.

CB

And that means that if they were to have children themselves, it could carry down to their children as well?

MS

That's right, yes.

CB

When did you start to notice that your son and your daughter displayed traits that weren't neurotypical?

MS

My daughter was my firstborn. She's 11 now. My son was my second and he's eight now. With my daughter, there wasn't anything that we really noticed in the first few years that we thought was particularly concerning. In hindsight, we can see signs now. But my son was probably the first one where we started to notice some things and that happened pretty early.

When he was about maybe... I actually remember quite distinctly, and I've spoken to Liz about this as well, when he was about six, seven months, around that age, I remember a tiny little thing, a little bell went in my head, started ringing and saying, I wonder if he's got autism because... But you sort of dismiss it because they're so little.

It was just little things like he wouldn't imitate us very much. He wasn't really babbling. It took him a long time to babble and to use words. It took him a while to develop clapping and waving and things like that. He was a late walker. Well, both of the kids were relatively late walkers. Just the way that he would interact with the world. He was quite fidgety with his hands and just a lot of those stereotypical autistic traits that they say, they raise a few concerns when they're little.

00:09:36

And so, we were wondering whether there were some developmental issues there, but he also had a lot of health problems when he was a baby. He had laryngomalacia. Funnily enough, there's been a few other Fragile X boys I know of that had laryngomalacia when they were born.

CB

What is that?

MS

It's a defect in the airway. Basically one... I'm not going to explain this very well, obviously, not being a doctor. But there's basically a valve that doesn't close properly or it's misshapen. And so, when they swallow, they can inhale liquids and foods and things. It can make it difficult for them to swallow and especially when they go onto solid foods. That's when we noticed it, when we started to introduce foods.

It self-corrects usually. It doesn't usually need surgical intervention unless it's quite severe, is my understanding. He also had grommets and needed adenoids out and all of that kind of stuff, which is really, again, quite common. It's just anecdotal, but I know quite a few Fragile X families who have had that.

00:10:43

He had a lot of those and we just assumed some of his delays might have been because he had multiple hospital stays. He had a lot of croup and things like that. But then it just got to a point at around two years of age where we just said, it's just not progressing, he's not catching up. He had a diagnosis then of global developmental delay and we had started speech therapy and OT and all of that.

We had him assessed when he was around three by a developmental psychologist and, again, confirming that global delay. Then our paediatrician decided to do a genetic screen just to be sure. He didn't actually think it was likely to be anything like Fragile X and then lo and behold, that's what it was.

CB

It's like you said, all of those medical procedures that he had to undergo at such a tiny age, you think, well, actually, he's having little setbacks all the time, isn't he, so give him a break sort of thing.

I'm really impressed that you guys managed to actually pick up that there was a difference and get the diagnosis. Three is still very young, isn't it? I think that's really good and it obviously goes to show how on it you are and the healthcare that you guys can receive in Australia.

MS

I think we were very fortunate and our paediatrician was very responsive to our concerns. He wasn't dismissive. He was aware of Fragile X. We weren't at the time. When we showed concerns, he took it seriously and we weren't dismissed.

I think he wanted to reassure us and give us all the answers that we could possibly get. Then when the diagnosis came through, he was incredibly supportive and we were in touch with the genetics team within 24 hours. We got a lot of support through that process, which was really good and so important, I think.

00:12:50

CB

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We think that Fragile X displays differently depending upon your gender. Can you talk me through some of the differences that you've experienced as a family?

MS

Yes, sure. Of course, as Liz said, it's a spectrum, so every child is different. But speaking generally, boys are typically more affected than girls, I guess. It's not always the case, but it seems to be typically the case. That's definitely reflected in my children.

My son is definitely more affected. His language is quite delayed. He's eight now and he probably has similar language to maybe a three- or a four-year-old. He can have a very basic conversation about everyday things and he does a lot of imaginative play. He is quite creative, it's just his expression is a little bit more difficult for us to understand. Whereas my daughter was a little bit slow to start talking but once she got going, she took off and she hasn't stopped since. She's very articulate and a very good communicator.

00:14:11

My son probably, if you were to meet them, you would probably pick him as someone probably with autism. He has a lot of those stereotypical features. He flaps. Obviously, the delay in the language. He's fidgety. He has a few other repetitive behaviours and he struggles in noisy environments, crowded places, new places. We had a long period of time where he wouldn't walk through doors that he wasn't familiar with without a lot of struggling.

He's got a lot of those sorts of features whereas my daughter appears a lot more typical. It presents in her mostly with anxiety. She's an incredibly anxious child. She also has ADHD. And so, it's very difficult for her to focus at school. She has an intellectual delay as well, so academics is very difficult and she has an adjusted learning plan. For her it's more ADHD and anxiety and for my son it's much more the ASD features.

CB

Are they in schools for children that need additional needs or are the in mainstream schools?

MS

My daughter is in a mainstream primary school at the moment. She's in Year 6 here in New South Wales, that's the last year of primary school, and due to go to high school next year. My son is in a school for special needs kids, so he's in a tailored school. But with...

CB

And does...? Sorry.

MS

Sorry, I was just going to say for next year we're not sure. We're working through that process for my daughter because we're trying to figure out what would be the best fit.

00:15:59

CB

For your son, with his communication delay, does that create a lot of frustration for him?

MS

Yes, it does. Especially when he was littler. Probably when he was at preschool age. Three, four, five-ish. Around that age. He had a lot more desire to communicate whereas before that he just didn't really want to say much. But when he got to that age, he started wanting to be able to communicate a bit more and he struggled for people to understand him and to form the words.

We did see a lot of frustrated behaviours and aggression, things like that, and him trying to

communicate using physical means. That was very frustrating for him and for us as well, not being able to understand.

CB

What tools did you employ to help him get across what he was trying to convey?

MS

Well, we actually did PECS, a picture exchange programme. We did that with a speech therapist. He would have a range of pictures of his favourite things, his favourite foods, his toys, all these sorts of things, and just daily tasks. He could take one of those pictures and give it to us. If he wanted an apple, he would bring a picture of an apple and then we would encourage him to say apple and then we extended that to a sentence strip, which would be I want an apple.

That actually worked really well. He started to get a bit sick of using it, I think, so he would use his words so that he wouldn't have to use the pictures. Then once he got quite good at doing that, he ended up just hiding his PECS book because he didn't want to use it anymore. He started using words because it was easier.

00:18:02

CB

Well, that's good because he's encouraging himself there, isn't he?

MS

Yes, that was really useful. And lots of just labelling and engaging with him and just responding to him even if you don't understand what he's saying.

CB

Liz, what support exists in Australia for families like Monique's? Is there ongoing support also? It's great that they're able to diagnose this at such a young age, but of course, you have Fragile X as a child, but you also will have it as an adult. And so, what's the support like for adults?

JL

Many families within the Fragile X community first hear about Fragile X associated conditions when they get a diagnosis as either a carrier or having Fragile X syndrome. It can be a really emotional time for families and individuals and can bring about a lot of uncertainty. But by the same token, it can be affirming to have an actual diagnosis.

The Fragile X Association's purpose is to provide support, counselling and information. Making contact through our helpline offers reassurance for many families and services and health professionals. We also offered facilitated online peer support sessions whereby we group individuals either with or caring for someone with Fragile X, which enables them to

talk about their experience and build connections and feel less isolated, regardless of where they're based in Australia.

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Information is power and there is a wealth of information on our website where we also have a wide range of webinars with experts presenting on Fragile X, which can be easily accessed by families, health professionals, educators and therapists working with individuals affected. The National Fragile X Foundation and the Fragile X Society in the UK websites are also terrific as a source of support and information.

Because, as you said, Fragile X is a lifelong condition, early intervention provided by speech therapists, as Monique mentioned before, occupational therapists and behavioural therapists can be ongoing and helpful to individuals with the syndrome as they move through the cycle of life. Speech therapy to help with ongoing communication, occupational therapy to help with sensory processing and deficit.

The environment has a significant effect or impact on the social and emotional development for a person, whether that be in a learning or social capacity. The workplace is often where many Fragile X individuals create and maintain social relationships. Females in particular often need additional support and encouragement in meeting and forming relationships outside of work because of their social anxiety.

It's helpful to focus on a person's interests and strengths when seeking social opportunities. But I guess the underlining theme throughout the lifecycle is that early, ongoing interventions provide the best outcomes and therapies and management strategies for helping are going to be helpful lifelong. Having a multidisciplinary team around you, and including family, of course, is going to help that individual.

00:21:20

CB

You've mentioned again about the social anxiety that females can experience and that is a recurring trait, that is quite a distinctive thing that females will experience. What therapies do you suggest for girls and women to assist them in that way?

JL

Well, it often goes under the radar because females, your strength is your weakness and vice versa quite often. It's a real strength of theirs to be able to mimic and mask what's perhaps happening internally whilst their anxiety is at its peak.

They'll present really well. One of the difficulties I think for the people around them is to actually pick up on the fact that they are experiencing anxiety and are feeling overwhelmed. Of course, enabling them to express that is difficult for them also because they might not necessarily be aware of it or be able to recognise it.

I think working with a speech therapist, again, on communication is really helpful. I've set up

a peer support group for young women with Fragile X syndrome for that very reason, so that they can connect and support one another through day-to-day events that come up.

Obviously, as they progress through life, like any other person, they'll seek out intimacy and relationships. Quite often because they don't pick up on the social nuances that other people would pick up on, they can find themselves being in very vulnerable positions.

00:23:22

Within that group we talk about how to keep yourself safe and how to remove yourself from a situation where you're feeling particularly vulnerable, which is difficult for somebody with a high level of anxiety to be direct. They will avoid confrontation at all costs quite often.

It's a really difficult thing for them to navigate and because they present so well, it adds another element of challenge for them, I think. But, look, a lot of women with Fragile X syndrome will tell me that it feels like they have an invisibility, that other people can't see what they're struggling with. It is a real challenge because they often fall through the gaps because they present really well in a lot of situations.

Speech therapy. I think finding a network, a peer support group where they feel safe and can be themselves. And armouring them with some key phrases as well to use when they're on the spot and in those difficult situations where they're feeling challenged.

Fragile X individuals generally, male and female, like to please people and sometimes that amounts to them getting into a little bit of strife and trouble because they're saying what's expected of them rather than the reality of what's actually happening.

CB

That's actually quite a dangerous position to put yourself in, isn't it, to be that people pleaser and not to think what's right and what's actually safe for you in that particular scenario?

00:25:15

JL

Exactly.

CB

Monique, is that something that you are having to firefight, if that is the right expression, with both of your children on a daily basis?

MS

Absolutely. Especially with my daughter. Not so much with my son at this stage. But that vulnerability to be taken advantage of is definitely difficult. I think because she is in more of a mainstream world. She's in a mainstream school, she has neurotypical friends, all of those

sorts of things.

There have been incidents where she's been taken advantage of and she's extremely loyal and caring and she wants to do what her friends want her to do. That has been taken advantage of a couple of times and she's at that age, I suppose, where we have a little bit more control and oversight and we can intervene.

But it's definitely something that I worry about for the future and something that we talk to her about and we work on a lot around her recognising what's okay and what's not and when she's uncomfortable, what she can do.

CB

For a lot... I'm not going... I was going to say for a lot of women, but I don't know if that's necessarily true. I think for a lot of people it's difficult to say no anyway.

00:26:49

The other day I was being followed by somebody in a car and basically he just liked the look of me. He fancied me and I think he thought it was complementary to have the window down, drive really slowly, be talking to me out of the car window. This happens quite a lot and it's incredibly frightening. It's really made me feel very vulnerable.

However, I don't say to this guy, can you go away, you're making me feel vulnerable because I'm worried about upsetting him and then what might happen. I think equipping your children with those tools to be able to protect themselves in all situations and scenarios is really important and you are doing a great job.

MS

I just say it's a work in progress.

CB

Work in progress, yes.

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What would you say are your children's strengths, Monique?

MS

Again, they're both very different. Actually, both of them are very loyal, it just comes out in different ways. It's funny because they will rip each other's hair out, but if somebody wrongs the other, they'll be the first in line to take them down.

They are very loyal and they really love each other and it's beautiful. They wear their hearts on their sleeve. They miss each other when they're not together...

00:28:30

CB

Oh, that's lovely.

MS

And they're not ashamed to express that. They love with all of their being, which is really nice.

They're also, I think it's a bit of a common thing that people say with Fragile X, is they're really funny. They have a really good sense of humour. They like to laugh. They're very creative and quirky in their own ways. Very imaginative and love to get a laugh out of people. They're little comedians. Which is funny, because they are both quite anxious, especially my daughter, very anxious. But at home she's comfortable and she will play the part.

CB

Really just be herself.

MS

Yes. She's a very creative kid. She loves working with colour and she's very artistic. That's kind of her strength. She's not going to be a mathematician, but she might be an artist. She has that kind of a brain, I suppose. And they both have exceptionally good memories, which you really need to be mindful of as a parent because if you promise something when they're four, they'll remember when they're ten.

CB

Do you know what, I've heard this before with a friend. Her child is autistic and he's in his twenties and he recalls the absolute detail of a day out when he was, as you say, four. That's incredible.

00:30:07

MS

Yes, you have to be very careful because they'll remember. Really good memories. It's hard for them to learn certain types of things because it just doesn't make sense in their brains. But if it's something that they can learn well and they're given the time to learn in the way that works for them, once it's in, it's in.

My daughter, she gets quite interested in sometimes current events and things and certain news things, stories or whatever. She'll remember all about it and if she learns about... They recently did the whole First Fleet and convicts thing here in Australia and the white settlement in Australia. She would come home and tell me all about it because she was...

CB

Is really interested, so that's why she was able to absorb it and it stuck.

MS

And so, she remembers all the details and once that's there, it's not going anywhere.

CB

So, you're learning from her.

MS

Yes, for sure. Homeschooling during COVID was quite an eye opener.

CB

I can imagine. Let me ask you, what do you think about the hidden disability sunflower?

MS

I had heard about it quite a while ago, actually. I mostly associated it with airports because I think that must have been the context that I had heard about it, for travel. Then I looked into it a bit more when we first started speaking and I think it would be really useful... I can see so many times that I would use it, especially with my son.

00:31:50

CB

Give me the scenarios.

MS

Because my son, he struggles a lot more in these sorts of new environments. And so, airports obviously is a big one because you tend to do a lot of waiting in airports and then it's very fun for kids.

And so, places like that. Even just restaurants and movie theatres. Everyday things that you want to be able to do with your kids that everybody else can do with their kids. Go bowling. Go to the zoo. Whatever it happens to be that you want them just to have a lot of those normal childhood experiences.

It's great when people are aware that they might have different needs and that things might be tricky for them. So, if they're having a tantrum in a line it's not because they're naughty, it's because they're struggling. Sorry, did I answer that? I feel like I went off on a tangent.

CB

No, you did answer it. That's what I wanted to find out, where do you think it would be of

benefit to you and your family and the reasons why. You've answered that perfectly.

MS

I think one thing I also would say about it is what I like about it in some ways is there's some certain amount of subtlety to it as well because it respects... With both of my children, for example, my son it's very clear that there's things going on with him that he finds things difficult.

00:33:21

With my daughter, she can cope fairly well in certain situations. Things like airports and such she's completely fine. And so, she's a lot more sensitive to making people aware of her Fragile X, so she only likes people to know who need to know or that she's chosen to know.

I would say that in that way it's good to have something that while it's known and it's understood by the people that are there to help, it's not too labelling. Does that make sense?

CB

Yes, it does. The thing is with the Sunflower, you can wear it as a lanyard or you can have it as a tiny little pin badge and you can take it on and off as to when you want people to be aware of it. We also have these Sunflower cards which has the name of the disability on. But, again, we only produced those because people have asked for them.

Some people like to have the thing on so that somebody can read easily, okay, this person has this and then they might put on the back the type of support that they might require. Other people are just like, I don't want to tell anybody what my condition is, I just want to be met with some understanding, an offer of help and kindness and just given more time to do what that action is that I'm trying to do or if I need somewhere to sit down, for example. That's the beauty of it that I think you've managed to recognise there.

00:35:02

MS

Definitely. I think that's important. I try to give the kids that autonomy as much as possible. Especially my daughter because she's so much more aware of what it is and how it affects her. I like to give her some choice in these sorts of things.

CB

I think that's really important, isn't it, that the person who has the disability has a voice in choosing what they want to share with people and when they want to share it.

We've had some questions from the Sunflower community about Fragile X that we wanted to put to you two. I'm going to ask the question and whoever feels they're most able to answer it, then please go ahead and answer.

The first question we had was this. I'm just going to read it out as it came in. Since having our son, who has Fragile X, we have discovered the chromosome came from my dad, passed on to me, so I am a carrier. He has tested as full mutation, same result as my son. However, he has led a normal life. So, his gene FMR1 must not have been affected. Surely people like my dad must create an interest when it comes into studying how to turn the FMR1 gene back on. Can't wait to hear your thoughts.

JL

I guess my immediate thought or thinking is it's a good question for a researcher.

CB

I'm sure you'll be able to funnel that through through your connections with the Fragile X Association.

00:36:49

JL

Yes, and luckily there are many researchers around the world interested in Fragile X. I guess some of the research involves conducting clinical trials to evaluate medications, which might be helpful for some symptoms associated with Fragile X syndrome which create challenges in daily living, such as anxiety.

But there's other researchers exploring whether the FMR1 protein, which is essential for normal brain development and function but is switched off for people with Fragile X syndrome can be switched back on. Other researchers interested in genetic conditions are also exploring whether gene editing can be helpful in correcting a genetic disorder.

It's a developing space in terms of research and a moving space, which is really hopeful. I guess we are learning more and more as we go. I guess this is a little bit off topic, but when I first joined the association four years ago, there were very few younger families coming forward with a diagnosis. Whereas now it's very commonplace to get an earlier diagnosis.

Which means out there in the testing and professional world that more and more GPs and paediatricians are attuned to the condition and have more awareness when they're presented with a child or an adult individual around the possibility that they might have Fragile X in the family or a development delay, for example. That's all encouraging and it's movement within the Fragile X space, definitely.

CB

So, that research is going into this anyway, so the lady's question, it is being looked into, it is being researched. But maybe if she wanted to contact you guys directly, you could maybe pass her to the relevant research people to find out a little bit more [overtalking].

00:39:04

JL

Yes, definitely. In America, in the States there's researchers, and elsewhere, obviously, who are looking into this and who are better placed.

CB

Next question is, and it's a bit similar. I have the gene in my family. If I want to have children, how likely is it that they'll be affected?

JL

Well, with each pregnancy, female carriers have a 50% chance of passing on the Fragile X gene change to a child of either sex as a carrier or Fragile X syndrome, whilst male carriers of the Fragile X premutation will pass on the premutation to all of their daughters, but not their sons.

In that scenario, I would recommend if there's a known history that you consult a genetic counsellor who would be better placed to explain the inheritance and organise testing for yourself and current and subsequent family members. What do you think, Monique? Have you got any further thoughts on that?

MS

For our second pregnancy we went through the IVF process and we didn't know about me being a carrier at that point in time and so it wasn't really an option for us. But knowing what we know now, if we were to go and have another child, we would definitely be... Which we will not. But if we decided we might, we would go down that path and discuss that with a specialist.

00:40:38

CB

Thank you. Next question. How can I tell the difference if my child has sensory issues or is being fussy or difficult? Maybe Monique wants to answer that one first.

MS

I think the thing with sensory issues, you'll start to recognise it because it's persistent. It may come off as fussiness or a colicky child or a difficult child or something like that, however you wanted to phrase it, but you'll find patterns in it. It might be that the environment is noisy, it's too bright. Maybe certain textures of foods are rejected or sought. It may be a seeking behaviour rather than an aversion.

My son always loved touching everything. He always had to touch everything. It's settled down a little bit as he's gotten older and he's learned a bit more about what's appropriate. And he would lick things all the time and everything would go in his mouth.

I think just from my perspective as a parent, what triggered it for me in terms of thinking that there were some sensory things going on was that consistent and persistent seeking or aversion to a certain sensory input.

JL

I agree totally, obviously, with Monique in that children with Fragile X syndrome have sensory sensitivities to lots of things. Parents will often say that they knew nothing about Fragile X at the time of diagnosis, but have since learned to become experts and I often refer to parents in that way.

00:42:36

Having an occupational therapist as well can help identify some of the triggers and strategies to help calm and regulate the nervous system when it comes to sensory sensitivities too. That's something to explore for families if they haven't already thought about it.

MS

OT has been amazing for my children. It's been really great for helping us understand what they need.

CB

That's what it's doing. It's the nervous system that's being affected when the stimulation through sensory touch or the tap sound or whatever, that's what's happening. And so, it's trying to calm down that nervous system, Liz.

JL

Yes, they're regulating themselves basically with a need to, as Monique was saying, to perhaps touch something and to make them feel safe and secure. I think you had a lovely way of describing it the other day when I was talking to you, Monique, about the pathways, the way that you explained that to your children. Did you want to share that?

MS

That was probably more around, well, yes, it applies for the sensory stuff as well, but also just for their academic stuff and their learning. Because, as I said, my daughter is more aware of her condition and what it means for her. And so, we do have problems with her anxiety and with her being quite down on herself about her ability.

The way that I explained it to her is when she says, for example, why can other kids do this when it's so much harder for me to do that, and that might be something that is sensory, so eating things that she's not comfortable eating. Or being in a place that is really noisy that she really struggles with, why is it okay for them but it's not okay for me?

00:44:39

I explained to her that for some kids, getting from A to B or learning something is more of a straight line in their brain and for her, she takes the scenic route. Her path is more windy and she has to climb some obstacles and go under some things. But she can get there in the end, it's just that she'll have a more interesting path to get there.

And so, that's how I explained it to her in that she will feel different. Some things will feel harder and some things will be easier and it will feel different along the way, but she can get there too.

CB

It's really a great way to help somebody identify their emotions and their view of the world by visualising it in that way, isn't it? That's a good analogy. I think that's the word, isn't it?

JL

Definitely.

MS

Yes, and that sort of thing works for her. She can imagine that, so that is a good way of explaining it to her.

CB

Yes, that's some good mumming there, Monique. Finally, my final question, what research is being done into effective medication?

00:45:52

JL

Well, because Fragile X syndrome is a syndrome or a spectrum, people living with Fragile X can be affected by various symptoms and in different ways, which is what we said before.

There's no one overall treatment or medication for all the symptoms associated with Fragile X, but certain existing medications can be helpful as targeted treatments for specific symptoms, for example, medications for ADHD or anxiety. Medications are used in combination with other treatments, such as therapies.

A multidisciplinary approach tailored to an individual, incorporating both medical and behavioural management and therapies is what most families tend to work towards. Would you say, Monique?

MS

Yes, I think so. I think medication is... Sometimes if I talk to other Fragile X parents and maybe they're a little bit less along the line than me or smaller kids or they're just entering it, we talk about medications.

I describe it as they're just tool in a toolbox. They're not a be all and end all. They sometimes give you that 5% benefit that allows the child to engage with other skills and to open them, to calm them enough that they can learn better skills to be able to regulate themselves or those sorts of things.

In terms of my kids, we do use medication. Both of my children are on medication for anxiety. One is on fluoxetine, one's on sertraline. We also use ADHD medication. Vyvanse for one and Ritalin for the other. We run quite a pharmacy every morning.

00:47:47

Also, my son is on a clinical trial through the children's hospital. I think that's a global one as well and that is a cannabidiol gel that's applied to the skin twice a day and that is to target anxiety in children with Fragile X. He's been on that for about three years now. It's been quite a while. We use a combination of medication and therapies and other supports.

CB

Do they need medication to help them sleep?

MS

Mine don't, but it's not uncommon. We have had problems with sleep in the past, but we've never used medication for it because the type of sleep problems, often people will use melatonin and it's more about sleep initiation, I guess, and trying to get off to sleep.

Whereas the problems that we had with my son in particular was waking somewhere between 11 PM and 3 AM and he would wake up and scream for an hour or two. It was just something we had to ride out in the end and it has settled now and he sleeps pretty well. But, yes, we haven't used it, but it's not uncommon.

JL

Actually, just on that note, we did recently run a webinar on sleep in relation to Fragile X syndrome with Honey Heussler, who is a developmental paediatrician, very experienced in sleep and Fragile X. You can access that on our website, if anybody has an interest.

CB

Thank you. That was what I was going to ask you next. How do people find you?

00:49:34

JL

Well, they find us through our website, www.Fragilex.org.au. People are welcome to have a peruse and share with family members, educators, therapists, anybody who'd like to know more about the condition.

It is sometimes educators, for example, who pick up on the fact that a child might not be developing, there might be some sort of developmental delay and they might actually bring that to a parent's attention. They're very involved, obviously, in the child's learning progress. We really invite educators and support workers and anyone who has an interest or otherwise to have a look at the website.

CB

Thank you, ladies. Thank you so much for your time. Monique, obviously you're a busy working mum as well, so massively appreciate you taking the time out to share your family's story with us and with everyone. I think it's been hugely educational and thank you so much as well, Liz.

MS

Thank you. Thank you very much for having us.

JL

Thank you. Thank you so much.

CB

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