Back to Basics: Guidelines for Management of Communication in Rett Syndrome

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- [Amy] At this time, it is a pleasure to introduce Dr. Theresa Bartolotta, who is presenting for Back-to-basics: Guidelines for management of communication in Rett syndrome. Theresa has been an SLP for over 30 years and specializes in working with children with complex communication needs, including autism, Down syndrome, and Rett syndrome. She holds a PhD in Health Sciences from Seton Hall University, a master's in speech language pathology from Queens College of the City University of New York, and a bachelors in speech from Mercy College. Theresa serves as a consultant to the Program for Research and Support for Rett syndrome at Monmouth University, is on the clinical staff of Tender Ones Therapy Services and teaches graduate courses as an adjunct professor. She is part of an international group of grant funded researchers who developed consensus guidelines for management of communication in Rett syndrome, and now serves as a consultant on communication to the International Rett Syndrome Foundation. So welcome Theresa. Thank you so much for joining us today.

- [Theresa] Thank you so much Amy, and I'm really happy to see all of the participants here and see all of this great interest in Rett syndrome, which is a topic that I've spent probably 20 years working on. So let's get started with some details. Here's my disclosure slide and then these are the learning outcomes that I have identified for today's seminar. So after this course, everyone who participates will be able to identify features of Rett syndrome that impact communication, and then be able to describe effective strategies for assessment of communication in individuals with Rett syndrome. And also be able to describe strategies in order to assist individuals with Rett syndrome to reach their communication potential, which I think we all know is so important through what we've identified as best practices in intervention. So I want to begin by giving a brief overview of what exactly Rett syndrome is, because it is very likely that if you are a clinician working with someone with Rett syndrome that you do

not have that many people with Rett syndrome on your caseload, because it is a rare disorder. So I'll just go into some of the basics, so that we have a good understanding of the fundamental information. So, Rett syndrome does occur primarily in females. It does occur rarely in males. There are males today living with Rett syndrome, but because it is an X-linked chromosomal disorder, males who do have that mutation do end up typically having a much more severe presentation and many males who are diagnosed with Rett syndrome, live a very short amount of time, and the mortality is very high.

So the majority of individuals living with Rett syndrome are females, so frequently throughout my presentation, I will likely say I might talk about a girl with Rett syndrome or a woman with Rett syndrome, and if you are working with a male or know a male with Rett syndrome, I do mean to be inclusive, but we tend to sometimes just default to talking about females because it is primarily a female disorder. The prevalence is approximately one in 10,000 live female births worldwide. The diagnosis is clinical and I do go through the clinical features in just a little bit, but as part of the clinical assessment, which is usually done by a neurologist or a team that is led by a neurologist. What is done after or as part of the assessment is a genetic test to see if the person has a mutation on what is called the MeCP two or the MeCP2 gene on the X chromosome.

And so often the genetic mutation confirms the diagnosis, but there are individuals who have a Rett syndrome diagnosis who have not been found to have the genetic mutation, yet they still meet the clinical guidelines. So most individuals with Rett syndrome are born after a typical pregnancy, and they look like they're doing really well, and there are no early signs in that neonatal period that there is something going on. What happens is there is a period of normal development that is followed by a regression and that usually affects both language or communication skills if the child is preverbal and motor skills. And the first regression can occur as early as the middle of

the first year, but it can last up until about between ages two and three. And it is quite common for a number of individuals who begin to show a regression to be thought as potentially having autism, because those of you who work with individuals with autism know that it is common for a child with autism to experience a regression. And so often people will look to see if they meet those initial characteristics. And there are a lot of similarities in the beginning between autism and Rett syndrome, but one thing that you can look at is that it is less likely that individuals with autism will have severe motor impairment and much more likely that individuals with Rett will have a significant motor impairment. So that can be a good diagnostic tool that you can use. So the diagnostic criteria were revised and updated in 2010. Previously Rett syndrome was considered to be an autism spectrum disorder, and it was listed in the DSMIV under the autism spectrum umbrella, but with the publication of the DSM V in 2013, Rett syndrome was deleted from the DSM and that has to do with the finding in 2000 that Rett syndrome is primarily caused by a genetic mutation, so it is no longer considered to be a disorder that is covered in the DSM. So the diagnosis as I said earlier is made clinically and then mutation is confirmed through the blood test.

And there are typically two kinds of Rett syndrome. There's classic Rett syndrome where someone meets all of the required criteria, and then there is a version that is often less severe that is called atypical or variant Rett. So these are the classic Rett main criteria, and all four of these are required in order to meet the diagnosis of classic Rett syndrome. So you have an early period of typical development. Then there's a regression which will last, it will begin before the third birthday and it may last say up to two years. So sometimes people are still regressing at four or five, but usually there's a stabilization somewhere between ages three and five, and then there is actually a regaining of skills, which is actually very positive. We used to think this was a degenerative disorder and it is no longer considered to be degenerative. So there are four required criteria. There must be a partial or complete loss of functional hand skills. So that means that someone with Rett syndrome is unable to engage in hands skills

that are part of activities of daily living. So for example bringing things to the mouth. As they get a little bit older using a utensil, playing with toys. So if you start to see these kinds of features it is a very classic of Rett syndrome. There is a partial or complete loss of spoken language skills. Some individuals never begin to speak. Others will begin to speak in that first year or a year and a half start to use some words, and then will usually lose all of their spoken words or most of them. The individuals who have retained verbal skills usually meet the atypical or variant Rett and they tend to have a less severe presentation.

So that's important to remember because most individuals with Rett syndrome are almost completely nonverbal. So if someone is on your caseload, and they are very young, it is always important to introduce augmentative communication, because it is very unlikely they're going to be able to use spoken language in a functional way. The third criteria is impaired apraxic gait or complete lack of ability to ambulate. So many individuals with Rett are completely unable to stand or walk. Some are but usually if they are able to walk, there are balance issues. They might have a very wide apraxic like gait, where there's motor planning problems, and usually their acquisition of motor skills are delayed. And then the fourth criteria, which is very classic, is stereotypic repetitive, non-functional hand movements.

And what these look like is usually it's bilateral hand movements and they can take different forms. So it is hand wringing or clasping the hands together or doing something with one hand, like twirling the hair and putting the other hand in the mouth, these are considered to be not generally under the individual's volitional control, though over time with appropriate intervention you can get some girls to use their hands in a functional way. And that's where working with occupational therapists is really very helpful. Along with the required main criteria there are supportive criteria. So these are conditions that very frequently accompany Rett syndrome and they're important for us to know about because as I talk about later, we will look at a lot of

these and think about their impact on communication and so it will affect how we assess and then what we use as an intervention program. So it is very typical to see respiratory disturbances and these can be characterized as rapid breathing or breath holding and as SLPs, we know that this can also impact feeding. And so often we'll see that some girls with Rett syndrome who have severe breathing issues and oral motor function issues have to rely on G-tubes for nutrition because they're unable to take appropriate nutrition by mouth. Teeth grinding is another feature, as well as impaired sleep patterns.

There's a pattern of very frequent sleeping during the day accompanied by frequent night waking. It makes it very difficult for the family. It also makes it very difficult for the girl to participate fully in educational or social activities because of fatigue. So we always want to be aware of that. Abnormal muscle tone is very classic. They are typically hypotonic, though some girls will have hypertonia. They have peripheral vasomotor disturbances. So they have poor circulation and often that is characterized by bluing of the extremities like the hands or the feet. Scoliosis is very common. It can start in late childhood or in early adolescence. Some individuals will have a very quick, severe progression of the curve. Some going from zero to 45 or 50 degrees of a curve in less than one year.

So surgery is often required, and so then that can impact their ability to ambulate or to sit up and be able to regard you know their communication partners, you know with good interaction. Many of them are smaller so they have a growth retardation. They tend to be a smaller height but that is not everyone in the syndrome. Some of the girls are bigger. However it is very classic for them to have smaller hands and feet. And as I mentioned before the peripheral vasomotor disturbances affect their circulation. So they typically have cold extremities. Often they'll engage in laughing or screaming spells, not appropriate to the context. They appear not to be under their volitional control, and it can really affect their ability to interact in social or educational activities.

They have a very high pain threshold, and then something that is very classic is what's called eye pointing or intense eye communication, regardless of whether the pattern of Rett syndrome is more or less severe, it is almost universal across everyone with Rett syndrome that their eye gaze is really excellent. You see it improve after they go through the regression, and so we as speech-language pathologists can use that for interaction and then also we can think about it in terms of when we move to augmentative communication trying to look at eye gaze as a potential way to assess ability as well as to enable them to communicate. So just to finish the description of Rett syndrome, atypical Rett, the person has to experience a period of regression and this occurs just at about the same time as I described earlier.

Somewhere around the middle of the first year up until maybe even age five. They must have a minimum of two of the four main criteria and five of the 11 supportive criteria. So someone who is diagnosed with atypical Rett syndrome may or may not have the mutation, but they do meet those criteria and they typically are less impaired than the more classic case. As we turn now to thinking about communication in Rett syndrome, I think it's important to emphasize that the early literature about Rett syndrome suggested that everyone with Rett was very severely impaired and incapable of communication, because they were assessed as being all pre intentional, so incapable of using anything in a symbolic way, and this would go back to the 1980s. Rett syndrome was first described in the world in the 1960s and the first case diagnosed in the United States was not until the 1980s.

So we still find that a lot of people don't know about Rett syndrome. There's not a lot of awareness about the potential for individuals with the diagnosis, but what we found is that once the gene was found, which was in 1999 and then the testing was used to confirm cases of Rett syndrome, that the spectrum of Rett syndrome actually became more diverse, so that people who have more ability were getting the diagnosis. So then I think that also shifted expectations and professionals I think are now more open to

considering communication potential. As I said earlier, most of the individuals are nonverbal and what we've seen in the last few years is that as advances in AAC technology have emerged, especially with electronic eye gaze systems, and hand-in-hand as a society, I think we have a greater understanding of the abilities of individuals with severe disorders. We're now moving to embrace a model of competence and that does not mean that there is a lack of impairment, but rather a shift in thinking to more positive opportunities and being more optimistic about potential for growth as we approach the individuals with whom we work. Now the literature about communication and Rett syndrome is still very limited. In 2019, Amoako and Hare published a systematic review of intervention studies on communication.

And the studies that they studied was a very small number. All of the studies reported improvements in communication, which is very positive but there were some methodological problems that plagued a lot of the studies. All of the studies had a small n, so a small subject number. It's hard to do anything if you're doing an intervention study with more than just a few subjects with Rett syndrome because it is so rare, and they do tend to be a very heterogeneous group. So we don't have a lot of data on communication intervention. So, about five years ago I joined up with three colleagues Gillian Townend from the Netherlands, Helena Wandan from Sweden.

They are both SLPs, and Anna Urbanowicz from Australia, she's an OT. We met at a conference in Europe about Rett syndrome, and we decided to try to see what we could do to advance awareness about communication potential in Rett syndrome. So we were recipients of a project that was funded by Rettsyndrome.org, which is also the International Red Syndrome Foundation. And in our project, our goal was to develop clinical guidelines for how to manage communication in Rett syndrome and what we mean by management is what are best practices in assessment and also intervention. So we try to pull the information together. So we have developed now what we're

calling consensus based guidelines and that's primarily due to the fact that the literature as I said, is very limited, but we did do an expansive review of English-language papers published all over the world, going back approximately 10 years. So we did a very comprehensive literature review. And then to add to that, we surveyed parents and professionals from all over the world who lived in a total of 43 countries and we translated the surveys into 14 languages and we had 650 people participate. And from that information that we gathered, we were able to put together guidelines for assessment and intervention. And we went through a Delphi process, we identified 36 experts from a variety of professional fields, including speech-language pathology, assistive technology, special ed and OT.

Also parents who knew a lot of individuals with Rett syndrome, so these were primarily presidents of parent associations representing particular countries. And what we did is we put together guidelines for statements about communication, and then recommendations. And then we have developed a handbook to enable parents and professionals to engage in best practices. So you see I have a reference there, Townend et al., 2020 that might have a list of references at the end of the slides and that refers to newly published guidelines by Rettsyndrome.org and you can go to their website to see lots of the information that they have on communication, including information on the guidelines, and we also have a journal article in press that is under final editing.

It has been accepted by the AAC journal and we're very proud of that and that will be out hopefully shortly. So what are some fundamentals? Fundamentals are is that the potential to communicators frequently underestimated in Rett syndrome as you think about some of the physical issues that I've described and the co-occurring medical issues, you can understand these individuals are really struggling often on a very daily basis with their bodies and so communication is very hard for them and if we don't really understand the impact of those features, we can underestimate what they can

do. An important point that I want to make and this is shared very widely by a number of AAC professionals is that there are no prerequisites needed for AAC. Someone should not have to demonstrate that they can function on a symbolic level in order for us to look at providing the individual an augmentative system so that they can then show us what they can do. Because Rett syndrome is rare, there's lots of people working with individuals with Rett syndrome who have never seen one before. The individual should be supported by a multidisciplinary team that can help troubleshoot all of the issues they might present with. That team should include an SLP and when needed seek outside expertise, such as a lecture like this, information from Rettsyndrome.org. Rettsyndrome.org also on their website has information about a North American clinic network, which they have developed. So there are specialty clinics across the US, where individuals with Rett syndrome can be supported by a team if they can be seen for regular assessments, and they can get information about AAC. So professionals who work with individuals with Rett should seek this kind of specialist knowledge that I was just talking about.

Connect with other members of the broader team, so that the individual and the family are supported and there's good consideration given to what their needs are and advice and recommendations are all coordinated, so everyone is working together. And then a key point which is true for everyone, who uses AAC that it is so key to train communication partners in techniques and strategies that will benefit the individual with Rett syndrome, and that is just something that we all should do whenever we work with someone who is not communicating verbally. So here I have an image for you of features of Rett syndrome and coexisting conditions that will impact communication, and I just want to go through these briefly with you. I'm going to start at the top on the upper left, just talking about each of these fairly quickly. I'll begin with dystonia and then go around in a counterclockwise fashion just to share with you some of the things that you should think about if the individual that you work with presents with any of these symptoms. So it is very typical for individuals with Rett syndrome to have motor

issues as I mentioned. So, dystonia which would be rhythmic movements that would be not under someone's volitional control, can affect their ability to use their hands, to use a part of their body with consistency. So it would be important to consider motor movements when you are planning on use of augmentative communication. And also if someone does have dystonic movements, to wait perhaps until they are not moving as much in order for them to give you a response that would be under their volitional control. With regard to this dyspraxia, we think of that as a motor planning issue, and we see that very commonly in Rett syndrome.

I talk a little bit later about a technique to use which is waiting for the individual to produce a motor response. We see that is very, very common for individuals with Rett syndrome to need extended response time. This can be as few as five seconds, up until a minute, and when you think about waiting an entire minute for someone to give a response, that's an extraordinarily long time. However, an educated communication partner who knows that it will take the individual more time to produce a response is really so key to the success of someone's communication development. As I mentioned earlier, there are hand stereotypies.

So, those repetitive non-functional hand movements that may not be under their volitional control. What you can do sometimes is you can stabilize one arm or the other using a soft splint. Here's where we need our OT colleagues who can help us problem-solve, because sometimes if you gently in a very supportive way help the individual with Rett gain control over those hands stereotypies, they may be able to access a communication device or a switch. They have impaired oral motor skills. So again oral speech is frequently not a functional goal and so early interventions should not spend a lot of time on oral speech. If someone is vocal of course you want to encourage that, their use of as many modalities as they can get under their control, but remember we want to go to AAC early and as early as possible. I discussed the impact of the breathing problems and sleep on fatigue as well as scoliosis on posture. I want

to mention that it's very frequent for them to have seizures, either grand mal seizures, for which they are medicated or perhaps shorter periods of inattention. There's this other term that you might hear called Rett spells, which are not necessarily documented as being seizures but if you see someone has a lack of attention or a moment, where they do not seem to be fully cognizant, you want to talk to the members of the team about that to see if medication potentially could help. There is a lot of description that there is heightened anxiety in the population. This can be due to underlying physical issues, difficulties with regulation. So we want to make sure that we can provide a supportive calm environment, as often as possible to help limit some of the impact on that on interaction.

I mentioned fatigue. When we go to think about sensory regulation, you can have someone who is over or under stimulated and this again is where we can use the input from our OT colleagues in our handbook we taught, we've been grateful to Judy Lariviere who is in OT in California, who uses a traffic light system. So, red, yellow, green. So, green represents a state of homeostasis, excellent, alertness ready to learn. Somebody who's in a yellow phase is either mildly under or over stimulated and somebody who's in a red phase is totally over stimulated, unable to engage or totally completely under stimulated and perhaps almost asleep. And so by speaking to our OT colleagues, we can get some suggestions on how to move someone to a more regulated state, so they are in that green state.

So, they are ready to learn and we can use things like giving a break, giving a snack, using music to help calm them. Gastrointestinal issues are very common in the population, a lot of the girls have difficulty as I said earlier with eating and swallowing, but constipation is a huge problem. So they can have discomfort. They can have reflux, so that can make them upset. For hearing it is very difficult to of course get thresholds for levels of hearing, but we are able to get some gross hearing measures. So, that we can get a sense of whether or not they can hear. We want to consider perhaps that

they might have auditory processing difficulties, based on how they present, so you want to think about using your toolbox in terms of presenting multimodal stimulation, providing extra time for processing. And then lots of individuals with other neurological conditions have visual impairments. And so you want to think about vision and speak to experts in that area when you plan for augmentative communication. So now, when we think about intervention and assessment, there are some strategies for engagement that we can use with Rett syndrome with individuals with the Rett syndrome to maximize their potential. So you want to address and talk directly with the individual, don't talk over them. Acknowledge that they can understand you. Make eye contact and use multimodal communication. That can also be a teaching opportunity. Make AAC available at all times. We'll talk later about augmentative communication strategies there's a lot of interest in using high-tech eye gaze, but we should be aware that not everybody has the resources for a high-tech eye gaze device, and high-tech eye gaze devices are not always appropriate in every situation.

So somebody has a high tech eye gaze device, we also want to make sure that they have access to low-tech or no tech at other times when high tech is not available. We want to think about decreasing motor demands that they have to, that they have to engage in order to give us a response as cognitive load increases because of the very severe motor planning issues that we see, so if you are assessing and you present them say with a page set on a device with lots of icons, instead of asking them to indicate a particular icon, what you might want to do is use a technique like partner assisted scanning, where you as the communication partner point out the choices to them, then give them time to process and then offer them again and see if they can give you a response. So we want to be aware that as we increase the cognitive load, we want to back off on the motor response because of the motor problems. Remember they might need assistance to be regulated and offer that extended wait time. So, that you can help them maximize what they can show you. So, I have three videos for you today. I'm going to go into my first video now. This is a small

conversation group, so you'll see two girls with Rett syndrome and there are a couple of adults here. This video is from my colleagues in the UK. What they are trialing here in this video is a low-tech communication system. This is called a super core communication book and it is available at thinksmartbox.com. This is a European company. So what I'd like you to look at in this video is look at the communication strategies for engagement, that the adult uses and how she uses strategies of waiting, eye contact, talking directly to the girl with Rett syndrome and then acknowledging motor and vocal responses as communicative. And I'll chat briefly with you about this at the end of the video. It's just I think about two minutes.

- Yeah, so, we can actually use the core words and on hands for these, okay, good. Okay who, I, it, you, he, she, can you see?

- [Child] Yes.

- Who.

- Who.

- Yes. You want to , 'cause you were kinda sleepy before, why don't you? So this is Tilly. I think you've met Tilly once before. And she was a . This is Tilly. She is coming . And this is Dee.

- [Dee] Hello Sylvia.

- She is Tilly's mommy and this is Daisy, and Daisy is the lady that made this book.

- [Dee] Can we go?

- Is that good?

- [Dee] Can we go Tilly?

- Do you have any wallets? That's right. You do, okay.

- You want some more?

- Is it a cool wallet I guess. Yes. Who, I, it, he, she.

- See you later.

- [Woman] See you later.

- Goodbye.

- What's am, go, help, hat.

- Have some more.

- No. Why, won't, stop, write, think. . Why? Why? Do you want to write that huh? Yeah. I think so, Tilly also knows Samona. I know that you are going to see Samona tomorrow. Well Tilly is also going to see Samona. Yeah? And that's why Daisy is here, because she wants us to try out this book and tell her what we think, yeah? I know you don't want trying things out. That's why you told me that would be okay. So, that's why.

- [Theresa] So I hope that in that video you were able to see a number of the strategies that were used by the adults. So the young woman in green shirt, you can see she's

talking directly to the young girl with Rett syndrome, who has the red sweater on, and she was acknowledging head movements, eye gaze as volitional and meaningful actions, and she was inferring communicative intent on that. The mother who was feeding the other little girl was also using her eye gaze, as a way to acknowledge her daughter's wanting to get more. They were trialing this, this would be considered a low tech AAC system. They were trialing core words and core words or words that are used in everyday activities. They're very common words and you could see how she was using the technique of partner assisted scanning, so she went through the choices. So, the little girl knew what they were and then she went back to see which one she wanted. So, I think that was a real nice example of that, of partner assisted scanning and using lots of modalities for communication. So, we can go back to the slides. Thank you all right.

So now let's talk about some fundamental principles of assessment. Assessment should be a team process, and you want to have the communication partners who are in the individual with Rett syndrome's life, part of it. We want to assess in as naturalistic settings as possible and this can also help with regulation and anxiety, just like with anyone you would assess for augmentative communication, you want to assess for opportunities. What are the opportunities in their environment? Who are their partners? Who do they interact with?

And then barriers to communication. Who can help them with their communication? Who is available? Who can be a good communication partner? A key point to make is that standardized assessments may not accurately reflect an individual's underlying ability. I know you're aware that standardized assessments can be adopted or I'm sorry adapted, so that you can, if you have a plate with pictures on them you can take those pictures and you can reproduce them to make them larger or move them further apart. So, someone can indicate through eye gaze, of course you have to note that in your assessment report. So, there is a way to adapt standardized assessments for use of

people with severe disabilities, but still they may not accurately reflect exactly what their underlying abilities are. So dynamic assessment is a principle that is used with a lot of individuals with complex communication needs and can be used well with Rett syndrome. So if some of you may not be aware of this, so dynamic assessment is a three phase process. So there's a testing process, that's phase one. That's where you provide the opportunity for the individual to show you what they can do. So, you ask them a question and then of course you wait for their response and remember they're going to need extended wait time. Now instead of just noting oh they can't do it, as you would in a formal assessment, then dynamic assessment has a second phase where you intervene and you provide some teaching strategies to assist the person in making the appropriate response.

So this might be using modeling, partner assisted scanning, you might offer the opportunity to use pictures, to use a switch. You can trial eye gaze, and then you see if the individual is able to produce a response using any one or all of those strategies. And then you do a retest, which is the phase three, where you provide the opportunity again, you ask the question, but then in addition to having the question, they also have at their disposal some of those strategies that you've trialed with them, and then you see if they're able to produce the response with that support and then that becomes part of your assessment report.

So this is a good tool to use whenever you're assessing someone with complex communication needs and can really be a nice model for assessment in Rett syndrome. Now because of the intense eye pointing and strong eye gaze we see with this population, eye gaze is usually the best access method. Not always. I want you to know that eye gaze can be very fatiguing if any of you have ever trialed an eye gaze device, it can be very challenging to use eye gaze for an extended period of time. So it's something that should be trialed in short periods initially and then you can increase the time that someone has access to an eye gaze device. Also a barrier to eye gaze

often is calibration, and there are ways to move to using, trialing an eye gaze system without requiring that calibration be established. So if you can't calibrate, it doesn't mean that someone can't use the eye gaze system. You want to give them the experience of it. So you want to think of providing a range of symbol systems and depending on where you live, what may be school district you're working in, what access you have to different vendors. There is a range of symbol systems that are available. Sometimes it's not necessarily you know a particular symbol system that'll help. It's more the particular layout you choose, may be beginning with a small number of vocabulary items initially and then going to large, I mean there really are two schools of thought here. There is a school of thought that you begin with a very small amount of vocabulary, like a core vocabulary and then as someone demonstrates they can master that, then you expand and there's another school of thought that says no, no you're limiting their ability to communicate initially.

Let's give them lots of vocabulary, but we'll just show them how to use it. So that's going to vary based on the needs what you see the individual able to do and also how the family is able to model and use it. Trial periods are essential. Think about the motor planning problems, the regulation planning, the regulation issues we've talked about. You're going to have to expect inconsistency.

This is not a population that you can write a goal that says they've got to do it or 70 or 80 or 90% of the time in order for us to acknowledge that they have that skill. That is often an unfair expectation that you might put on an individual with Rett syndrome, because of the other issues that are going on with them, and that's why as long a trial period with a device is as you can get would be important so that the person has the maximum opportunity to try the device. When we move towards intervention, our goal should include development of nonverbal, low-tech and high-tech communication strategies, across multiple modalities. And I've got a couple of videos, where we look at some other modalities of communication but remember if you have an ambulatory

person, they may not be able to use an eye gaze device all the time because depending on how they move, they may not be able to trigger the cameras. They may only be able to use their eye gaze when they're seated. So then when they're ambulating, they've got to have another way to communicate and we have to think about that. So because these individuals have really complex communication needs, and we want to be able to acknowledge all potentially communicative behavior, it's really important that we think that they're going to be communicating using multiple modalities. So when you're beginning to learn about a person with Rett syndrome, take a look at how they use their body, facial expressions, moving their trunk forward, that could be an acknowledgement, that could be a yes.

Do they make any gestures? Are they vocalizing? Can we assign meaning to those vocalizations just as we would with a very young child? Are they able to do some spoken words, even if they're repetitive, even if they're echolalic, acknowledge them as meaningful. And if they look at something or someone, then acknowledge that that eye gaze to that object or a picture or person is meaningful, and let's think about using combination of symbols, pictures or text. There's lots of interest in providing this population with access to literacy, as there should be, and we want to think about that as a possibility. So here I have a video for you where we have an individual who uses vocalizations to communicate, who uses lots of nonverbal communication, and I think you'll see here some really nice, the need for wait time with this individual okay? And I'll talk about it at the end of the video. This is just under a minute.

- [Therapist] Hey Lisa, hi, hey hi. Are you looking at your iPad? Oh, Lisa, do you want to watch a movie?

- Mm.

- [Therapist] You do? What movie do you want? I know you're excited. Are you gonna pick a movie?

- Yes.

- You are, okay. You know what? Why don't use your hand? Use your hand, there you go, pick a movie ah. There you go you did it. Good job, Lisa.

- [Theresa] Okay so in that video you see this young lady. She's actually 30 years old and she is ambulatory, does not use an eye gaze device. You can see that she does have some preserved hand function. Her hand function has actually gotten better as she's gotten older. I'm gonna go full disclosure here. This is my daughter, and this is why I got interested in Rett syndrome, so many years ago. She was diagnosed when she was 11, but you can see that she uses a lot of body movements. She moves her body forward. She uses eye gaze and vocalizations and we assume that they are intentional and we acknowledge them as being communicative, and she does need extended wait time, and you can see that with the command to use her hand, after a few seconds she did do that. So, now we're moving--

- [Therapist] Hey Lisa.

- [Theresa] Sorry can we go on to the slides? That was my error sorry. Thank you. So now we'll go on to talk a little bit about AAC. So AAC can be aided or unaided, and as you just saw, Lisa did with her iPad, she can use direct selection. She does have an isolated finger that she can point but some individuals can activate a switch. Partner-assisted scanning which is a very widely used technique with AAC. Now it's a great technique for a Rett syndrome, where you read out, as the partner you read out the options available to the individual and then they indicate their choice. And this is especially important when they're not able to use their eyes to let you know exactly

what objects or symbols they want. It could be because there's a lots of options on a page and you can't read which one they're looking at or very early on when you're just getting AAC going and you want to teach them that by looking at something that means that they are communicating. We can use modeling or aided language stimulation to show them how to use a device. So we get lots of questions about individuals who either aren't using devices even as adults or individuals who are using very simple systems, and they want to move towards more complex systems. And aided language simulation, where the communication partner acts as the AAC user. So, they point to the symbols so they activate the symbols while they also talk add a verbal message, as well as they indicate the symbols can really show the individual how to use the system to communicate. So that's a great technique. When we begin to develop communication is very important to establish a yes-no.

So there's been a lot of talk about determining a best yes and Susan Orwell, who's a special educator, who's really an expert in Rett syndrome talks a lot about a best yes, and that means the individual you work with may not be able to give you a conventional yes like nodding their head or vocalizing, that sounds like yes, but they might be able to look away for yes or close their eyes for yes, or look at a green check. Some people do well by learning to use a yes/no strip, and that can be something that you could start initially, ask them about something that you know they like a lot, and see how they respond and you could take that as their best yes, especially as you're starting communication and then you can go from there.

One thing I did not write down in this, but you may have heard about a communication passport, which is a document that people will create for an individual who is an AAC user, will you talk about their interests and you talk about how they communicate. And so if an individual has a best yes or yes, that is kind of unique to them and not standard. You definitely want to make sure you have that information available to anyone who could be their communication partner. As we move from developing

yes/no when we go to more complex vocabulary, you want to consider establishing a core vocabulary which is the vocabulary that's used every day in lots of situations. That's considered sometimes devices will call them quickfires. You can go to a quick-fire page where you have that core vocabulary. So that's words that can be used in multiple activities. Fringe vocabularies is vocabulary that's more specific for a specific activity. You can design activity boards. So, I have an activity board on the left. So this would be for a specific activity that somebody would be engaged in, whereas a context based board is a board that where the vocabularies they are just for a context, like going to school.

So, you can customize the vocabulary for the individual based on what their activities of daily living are, and then provide those vocabulary items to them in those particular situations, using aided language modeling, to show them, how to use the device and then assisting them in their communication with partner assisted scanning. In terms of access as I mentioned earlier, eye gauze works for most, but it can be fatiguing. So, you might want to think about an alternative system for when the person's ambulating or they don't have access to an eye gaze device, or they don't have access to electricity. We can't assume that everybody has access to these devices. If someone has some preserved hand skills, we want to try to maximize those. We used to think as I said earlier this was a degenerative disorder. It is not.

There is potential for them to get better at hand use and individuals can certainly use more than one access method. So this is the final video I have for you. This is a mother/daughter duo. This young woman is in her mid-20s. She started using an eye gaze device probably when well, probably when she was about 19 or 20. So, it's fairly new for her. With her the communication partner is key to her success because the communication partner believes that she is intentional. Look how nicely her mom waits for her and reads her behaviors. So, if we can go to, oops, can we go to the video? Thank you. So this is again just over a minute.

- Cookies, cookies.

- You talk.

- Cookies, bite.

- Cookies.

- More cookies.

- More.

- More, okay.

- More, yum.

- That was yummy. That's yummy. Glad to hear that it's yummy.

- More.

- Okay.

- More, all done, all done.

- All done, okay. Awesome.

- [Theresa] Okay, thank you. That's the end of that video. We can go back to the slide so you can see in that mother-daughter duo, the mom is really using some great

strategies there. She's waiting. She's looking at her daughter. That pair was part of a study that we did several years ago where we worked on coaching of the communication partner to wait and to acknowledge any behavior that they saw by their daughter as intentional, and I really think that was the key to moving the daughter and advancing her and using her electronic device. So they've been really wonderful to work for, to work with and I think it's also really, it speaks to the idea that it's never too late to begin to move toward a more complex communication system. So the final point I wanted to make is about literacy. Just like with any other learner, we want to provide access to literacy to print, because you never know how high they're going to go. Augmentative communication, symbol systems have icons available with the print paired with it to provide multimodal stimulation.

Let's not assume that they can at least learn some sight words, and who knows how far they'll go. There's lots of information on using literacy systems and reading programs to really develop sight word vocabulary, phonemic awareness, and writing skills. And I want to end by saying it's never too early to begin communication with this population, but it's also never too late. We've seen lots of adults who as they're older still or healthy, the individuals with Rett syndrome are living longer. We used to think that their mortality which is higher than their same age peers, but we thought their mortality was, their lives were very short that many of them were going to have an early death but with an improved health care and management of some of the physical symptoms that can affect health, they really are living longer and everyone deserves access to communication. So I then have a list of some references here for you and I want to thank you so much for your participation in this, and for your interest in Rett syndrome, and for your work with these individuals. Thank you.

- [Amy] All right well thank you Theresa. It's always great to hear your expertise in this area. So let's go ahead and address some of the questions.

- [Theresa] Yes now I had seen the first question and I clicked on it and it went away. So I do want to address it very quickly. It was that someone wanted to know if genetic testing in the prenatal period would identify Rett syndrome? And yes if you can get DNA material from the child with Rett syndrome, yes it would identify it. Prenatal testing is not done for a Rett syndrome, because it is such a rare disorder. And I should also say that it is also most often not inherited. It is caused by a spontaneous mutation, so it's like Down syndrome. It's something that, you know there's really no, nothing from the family history that could predict that Rett syndrome would occur. Okay, so should I just go through the questions? Amy how should we handle this?

- [Amy] I'm happy to read them for you and then you can just focus on the answer. Okay thanks. So when does the period of regression begin?

- [Theresa] So, it can begin as early as five months. So right in that very middle of the first year, when they almost have not gained any skills, and it can start as late as about 30 months.

- [Amy] Okay, is there an age that is too late to work on hand use for switches? I work with a child who uses eye gaze AAC device, a device, but would also like access to switches for other activities or when her eye gaze device isn't accessible, but she doesn't have hand splints.

- [Theresa] Okay this is a great question. So, you saw that video of my daughter who's 30 years old. Right after she got the diagnosis of Rett syndrome when she was 11, because she's not, she wasn't considered a typical person with Rett syndrome. She was diagnosed after the gene was found. So her OTs started her on what we would call a hand strengthening program. And I really have to credit the OTs with working with her to be able to gain more hand control, and so as an adult now she can hold a cup, pick a cup up, put it down, use a fork, use a spoon, open a door. She could do none of that

when she was younger and she is not alone. So I would say absolutely this is something that could be addressed. There's always the potential for learning hand use, but I really think you need to consult with at least an OT or a PT. You want to identify which is the dominant hand, especially if they're doing any kind of hand clasping or repetitive hand movements that are considered not to be intentional. So, you want to see if you can do something in a very gentle not restrictive way to break that pattern and then give that girl access to trying to use her hand in a meaningful way.

- [Amy] Okay thank you. All right the next question is when I've worked with my student with Rett in the past, we used PODD or P-O-D-D with partner assisted scanning, which was very similar to what was presented in the video. It was really difficult to get responses that seemed on topic or appropriate, as they were in the video that you showed, such as asking why and who. I've heard with Rett's this sort of habituation effect, and things need to be constantly made new, exciting. Everything comes with a personally engraved invitation. I'm not sure that my student wasn't able to use PODD or PAS but that was boring or she didn't want to because she thought it was too simple. What are your thoughts?

- [Theresa] You know I've heard that from families that they say that their daughters if they're being talked down to or the topic is not of interest to them, that they will shut down, fall asleep. So, if that was the case I would try a few different things. I might first of all find out what are the topics of interest, see if you can, see what she looks like when she's really excited about something. I know there's lots of interest, you know there's lots of people say well you know we should only present age-appropriate activities. I think you want to find interests that are really specific to that individual and regardless of the age level of it. So, I'd find out what really turns around, what does the parents say you know she'll get excited about and it could also be that the vocabulary might have to be made simpler on a page. Some do find that the range of choices on a

page might be too many and that could be a turn-off as well. So, those would be some of the things that I would try with that young lady.

- [Amy] Okay thank you. We have quite a few more questions, and so I would like to invite those who need to log off, you are more than welcome to do so. You will be given full credit for the course. Theresa do you still have time to address a few more questions?

- [Theresa] Yes absolutely.

- [Amy] Okay so with your information about eye gaze, is there a limit on amount of time a child should be in front of an eye gaze device to communicate, such as 20 to 30 minute blocks, five to eight times a day?

- [Theresa] Well I think that's going to vary. I do think that you want to give them the opportunity to look at something that's further away. Think about what happens to us when we're looking at our computer screen for a long time. We get eye fatigue. So we want to take the same principles that apply to everyone else in terms of doing that very close work and think about giving breaks 20 to 30 minutes I think sounds very reasonable. I know that adults who sit at computers are told to get up at least once an hour. What you want to do is you want to try to work up to the 20 to 30 minute block. You would never want to start somebody out that that an intense a period. You might want to just start for a couple of minutes and then direct their attention to something else and then come back much later, but yeah you want to have for good, you know visual hygiene, you want to be very cognizant about giving them breaks.

- [Amy] Great thank you. Is communicative intent also an important factor for differential diagnosis between ASD and Rett syndrome?

- [Theresa] Wow that's a really good question. I think, so what we usually see with autism, we don't see all of those motor problems though some of the, some kids with autism have you know motor planning problems, but they don't those non-functional hand movements. The problem with assessing communication intent in Rett syndrome is that it is often very hard to see that it's I think sometimes harder to see because of the physical issues that you have, the regulation issues, the fatigue issues, and the delayed response. In autism I think it sometimes can be easier to identify something that a child with autism is interested in, even if it's you know a very small core of items like cars or blocks or colors or something or food. And it's sometimes going to be harder to see that in a child with Rett syndrome. So, that's why for so long people thought that they were incapable of communicative intent, where I think what we see in autism is we don't see the shared attention. Lots of kids with autism have intent. They want what they want but they're not interested in sharing their ideas about it with you whereas in Rett syndrome, you get the really good intense eye gaze. So that's what I'd look for. Great question.

- [Amy] Okay, great, thank you. All right just a couple more and then if we're unable to address your specific question today, you're more than welcome to email those questions to me, Theresa I'm guessing it would be okay if I forwarded them on to you if you're able to address them that way maybe that's an option.

- [Theresa] Yes absolutely. Be very happy to do that.

- [Amy] Okay so the next question is would physical movements such as swinging help with regulation in responses during speech therapy?

- [Theresa] It might, it might. Because of the difficulty with staying in that green zone swinging could help stabilize someone, put them into that homeostasis, so they're ready to learn but for other individuals, it could very likely throw them into that over

stimulated, yellow into red zone, where they might have trouble coming back. So that's again where I think we need to talk to our OT colleagues to help with, to give us suggestions on techniques that can help bring them into regulation, but you can try it.

- [Amy] And we'll take this last question.

- [Theresa] Sorry.

- [Amy] Okay sorry about that. That's okay no problem. Are standardized assessments considered a valid measure of language and cognition in individuals with Rett syndrome?

- [Theresa] Usually not and because it is so difficult to assess them in a standard way that what you would be, what you would learn from the assessment will give you such a low score, that it likely incredibly underestimates their potential. Just like individuals who have very severe cognitive disability often are not good candidates for formal assessment of cognition, because there are actually things they can do in activities of daily living that indicate they have a higher level of cognition. Then they would be assessed at on a standardized test because you cannot accept qualitative responses in a standardized test. You have to stick to the instructions. I'm not suggesting that all individuals with Rett syndrome have severe cognitive impairment. That's a big topic for a discussion. There's lots of ideas about that, but the prevailing thoughts right now is that the very severe motor impairment, which then of course impacts somebody's ability to engage in the environment and learn, coupled with the physical problems about regulation, fatigue, seizures et cetera. And then the severe communicative problem that it could very well likely mask cognitive potential. So, if you could stay away from standardized tests, if you can use criterion reference tests, that describe behaviors and can actually be helpful in developing an intervention plan, if that can be

accepted by your institution, wherever you work, whether it's you know early intervention or a school, that's what I would recommend.

- [Amy] Great very, very helpful. Okay I think we're going to go ahead and wrap it up there for today. Again thank you so much Theresa for joining us. It's always a pleasure to have you here and learn from you, and I'd like to thank all of our participants for joining us today and asking some wonderful follow-up questions. We certainly do appreciate everyone's time and look forward to seeing you again soon. Again if we were unable to address your question today, please feel free to email them to me ahanson@speechpathology.com and we'll see if we can get them answered for you. All right we'll go ahead and wrap it up there. Thanks everyone, have a great rest of the day.

- [Theresa] Thank you so much.