

## **Overview of Genetic Syndromes Associated with Intellectual and Developmental Disabilities**

### **Slides 1, 2 and 3**

**No voiceover**

### **Slide 4: Mental Health Wellness for Individuals with Intellectual and Developmental Disabilities (MHW-IDD)**

Certificates of completion are available for each module. Please print or save your certificate immediately following completion of the course as they will NOT be available to you at a later time.

It is estimated that it will take you 50 minutes to complete this module. It **does** need to be completed in one sitting as your work will be lost if you close out this program during the middle of the module.

You can pause or back up at any time during the training but you cannot fast forward through slides. Slides will automatically advance once the voiceover is complete

Your computer must have speakers or you can attach headphones or ear buds, so that you can hear the information said by the presenter. For those who are deaf or hard of hearing, a narrative of each slide is available in the Resources section of this online training.

### **Slide 5: Overview of Genetic Syndromes Associated with IDD**

Welcome to the Overview of Genetic Syndromes Associated with Intellectual and Developmental Disabilities. This module is directed towards Direct Service Workers, or DSWs, as they are fundamental to supporting recovery and mental wellness in individuals with an IDD and genetic syndromes. However, the intended audience truly extends beyond those who are paid direct service workers. While the focus is on DSWs, the information in this module applies to all caregivers and other people who support individuals with an IDD.

#### **Objectives:**

In this module we will:

- 1) Provide overview of genetic syndromes
- 2) Provide awareness about shared behavioral traits and specific syndromes associated with Intellectual and Developmental Disabilities (IDD)
- 3) Present resources to learners on best practice treatments and supports

#### **Outline:**

Genetic Syndromes associated with IDD

- I. What is a Genetic Syndrome?
- II. How Genes Affect Behavior: Nature-Nurture Transactions
- III. What is a Behavioral Phenotype?

- IV. Common Genetic Syndromes Associated with IDD
  - Physical characteristics associated with the syndrome
  - Medical problems that can affect behavior, learning, and health outcomes
  - Behavioral phenotypes, including challenges and strengths, and support strategies
- V. Review of Genetic Syndromes and Strategies for Supporting Individuals

**Outcomes:** Learners will be able to identify common genetic syndromes associated with IDD, and be able to identify specific strategies to support people more effectively.

### **Slide 6: Introduction to Genetics**

Before we dive into the material for this module, it is important to note there are several resources available to aid in your learning and understanding of this exciting, but sometimes difficult material. Throughout the module we will refer to a table and a fact sheet that can be found in the Course Resource section of this module in the upper left hand corner of your screen. It may be helpful to print these out and keep them as resources during this training and for your future work in supporting people with genetic syndromes. Okay, now let's get started!

If we are going to learn about **Genetic Syndromes**, we need to understand a little about "genetics." **Genetics** includes how human traits are passed on from generation to generation, down the family chain. We inherit all kinds of traits and tendencies from our parents, including things like eye, hair, and skin color, as well as more complicated characteristics including potential risk factors like the risk of developing high blood pressure, or even, to some extent personality traits.

### **Slide 7: What is a Genetic Syndrome?**

Each of us is a product of the genetic material that we get from our parents. In each cell in the human body, half of the genes come from the mother and half come from the father, creating a unique genetic code, or biological blueprint, that determines our hair color, our body size and even some of our behaviors; but the code is not a simple copy of our parents' traits. Sometimes, people inherit genes for illnesses that the parents don't show; sometimes the genes of parents combine in ways that produce a condition in their child that neither parent may have, and sometimes the genes make errors, such as making too many copies, breaking off pieces of genes or recombining pieces that usually do not occur together. These can create changes in the genetic code that are called genetic syndromes.

### **Slide 8: Importance of Identifying Genetic Syndromes**

Genetic syndromes occur when there is an abnormality in our genetic codes. The genetic code we are all born with is like a blueprint. Sometimes, in the blueprint, there may be problems in the structure built from it.

A genetic syndrome is a set of characteristics and features, shared by people who have the same genetic blueprint. If a syndrome is present, such as Down Syndrome or William's Syndrome, two syndromes we will discuss in greater detail later, correctly identifying the syndrome is very important because we can then provide educated care specific to that syndrome. This is because syndromes cause risks for specific health and behavioral health challenges.

Knowing syndrome-specific risks for medical and mental health disorders allows us to provide better, more informed healthcare. Some genetic syndromes can increase risk for specific health and mental health disorders and may also be associated with certain behavior patterns and personality features. We especially want to know if a person has a specific syndrome that poses a high risk for certain medical problems, so we can prevent problems or catch them early to promote optimal health. When people are healthy, they are less likely to engage in challenging behaviors. Challenging behaviors are in fact sometimes the first or only sign that a person with IDD is experiencing physical distress or pain.

Let's now move further into how our genetic blue print affects our behavior.

### **Slide 9: How Genes Affect Behavior: Nature-Nurture Transactions**

How can genes actually affect our behavior? Well, because of our genetics, we may be able to do or not do certain things better than others. But genes do not dictate a person's whole life story. Our genes interact with the environment around us to affect the total person and shape who we become. A poor, stressful environment can limit a person's growth and potential. Developing and living in an enriched environment can help us reach our best potential. The DSW has a very important role in creating an enriched environment for people with an IDD by adapting environmental conditions that support the individual's strengths and allow for successful teaching opportunities.

### **Remember, focus on people's strengths!**

We'll now move on to discussing behavioral phenotypes in people who have an IDD.

### **Slide 10: What is a Behavioral Phenotype? Introduction**

Behavioral phenotypes are the shared personality features, patterns of behavior and temperamental tendencies that are associated with having a specific genetic syndrome. Temperament is the combination of mental, physical, and emotional traits of a person, or their natural predisposition, to respond to his or her environment. The biological core of temperament remains relatively stable across someone's life, but the behaviors or actions can be modified by the environment.

### **Slide 11: Application Exercise**

### **Slide 12: What is a Behavioral Phenotype? Potential Difficulties**

In trying to best support someone with an IDD, it helps a great deal to know that a person may be more prone to have certain strengths or areas of greater challenge in terms of how they learn, plan and problem solve. Individuals with an IDD have challenges in learning, whether they have an identified genetic syndrome or not. These include difficulties with remembering, focusing attention, organizing, time management, impulse control, self-monitoring, problem solving and planning. Many people with an IDD have limited social awareness and social skills, as well as communication challenges. All of these factors can contribute to stress which can cause challenging behaviors.

### **Slide 13: What is a Behavioral Phenotype? Temperamental Differences**

For instance, a parent who has had multiple children may notice that brothers and sisters may seem to be born with different “temperaments.” Even though they come from the same family, grew up in similar environments and had many of the same experiences, one child is “easy going,” and doesn’t seem to get excited or upset easily; another child is “easily excited”, under the same exact conditions. One child is active, but not overly so; the other is on the go all the time. These “temperamental” differences may have a **genetic component** rather than being just a “learned” behavioral or emotional pattern.

### **Slide 14: What is a Behavioral Phenotype? How to Respond**

How we respond to someone, and their unique “style” of engaging the world, can make a big difference. If we panic when the child who is energetic and over-stimulated runs around in the store, we can expect that child to become even more excited or anxious. However, if we are calm in our approach, we are more likely to have a calming effect on the child’s behavior.

Once we understand the common shared behavioral characteristics of a person’s genetic syndrome and how the syndrome can impact the person’s temperament, we can adjust the way we communicate, teach and support that individual. It is important to build skills using the person’s strengths to maximize their success and quality of life, as well as their health and personal growth. It is important to understand the behavioral phenotype of a person with a genetic syndrome so we can better predict and even prevent common behaviors associated with that syndrome.

Let’s now move on to common genetic disorders associated with IDD.

### **Slide 15: Common Genetic Syndromes Associated with IDD: Overview**

The purpose of this section is to teach you about specific syndromes and to provide you with focused, positive strategies to help increase the quality of life and outcomes while reducing challenging behavior. For each syndrome, we will discuss:

- Physical characteristics associated with the syndrome
- Common medical problems that can affect behavior, learning, and health outcomes, and

- Common shared traits, or behavioral phenotypes, and support strategies.

### **Slide 16: Five Common Genetic Syndromes Associated with IDD: Examples**

Let's get started on specific genetic syndromes. We will discuss the following 5 syndromes:

- Down Syndrome
- Fragile X Syndrome
- William's syndrome
- Prader Willi syndrome (PWS); and
- Cornelia de Lange syndrome (CdLs)

In the Course Resources section of this module, you will find a document labeled "Common Genetic Syndromes Associated with IDD." More specific information about these and other genetic syndromes can be found on this table. There are also a number of web links that can help acquaint you with different genetic syndromes to help plan effective strategies.

### **Slide 17: Common Genetic Syndromes Associated with IDD: Down Syndrome No narration**

### **Slide 18: Common Genetic Syndromes Associated with IDD- Down Syndrome: Physical Characteristics**

First, we will discuss the different physical characteristics and commonly associated medical problems of Down Syndrome. Support strategies for this syndrome immediately follow.

Some of the physical characteristics associated with this syndrome are

- Low muscle tone
- Short neck, with excess skin at the back of the neck
- Flattened facial profile and nose
- Small head, ears, and mouth
- Upward slanting eyes
- Wide, short hands with short fingers
- A single, deep, crease across the palm of the hand, and
- Short stature

### **Slide 19: Common Genetic Syndromes Associated with IDD- Down Syndrome: Medical Problems**

Some of the common medical problems that people with Down Syndrome may have are:

- Problems with vertebra in neck
- Obesity
- Heart problems, usually diagnosed as infant
- Increased risk of vision or hearing problems

- Thyroid problems including Hypothyroidism, or an underactive thyroid gland
- Gastroesophageal reflux disease, or GERD, which is a backflow of acidic stomach contents into the esophagus
- Constipation, and
- Increased risk of developing Alzheimer's disease at an earlier age, such as in their 40's and 50's

**Slide 20: Common Genetic Syndromes Associated with IDD- Down Syndrome: Medical Support Strategies**

There are many support strategies that can help people with Down Syndrome experiencing these medical problems. Let's review a few.

- Thyroid problems can cause irritable mood and lower the threshold for challenging behaviors. Screening for thyroid problems is part of "Best Practice" primary care for these individuals.
- Higher rates of **cervical spine abnormalities** could put an individual with Down Syndrome **at high risk of injury**, or even death, during physical interventions such as restraint. Cervical spine problems can also be associated with pain and cause the person to be irritable and more likely to engage in challenging behaviors.
- Additionally, obesity and congenital heart abnormalities have also been associated with injury and death in individuals who have been restrained.
- Obesity is a common medical problem, which can lead to other medical problems, including sleep apnea and heart disease, high blood pressure and diabetes.
- Because of difficulties with hearing, individuals with Down Syndrome may become frustrated, for example, if they cannot hear what others are asking them to do. People with hearing problems can seem like they are not being cooperative when actually, they may not hear what was asked of them. Also, poor hearing can lead to confusion. DSWs play an important role in recognizing when an individual with Down Syndrome has new or more severe challenging behavior, or irritable mood. It is important to let the individual's doctors know so they are checked for the many conditions listed here, before treating these challenges only with psychiatric or behavioral interventions.

**Slide 21: Common Genetic Syndromes Associated with IDD- Down Syndrome: Common Traits**

We will now move on to discussing common traits that people with Down Syndrome may have.

- The majority of people with Down Syndrome have mild to moderate intellectual disabilities; however some may have severe intellectual disability or none at all.

- Mental health challenges may relate to Obsessive Compulsive Disorder or Depression
- People with Down Syndrome tend to be socially motivated and make friends easily. However, they have also been described as inflexible at times.
- Being inflexible can lead to challenging behaviors including agitated outbursts. For example, people with Down syndrome may “dig their heels in” and not want to change their opinions! Rather than this being a challenge, this can actually be seen as a strength! That is, being persistent when you believe in something.
- For children with Down Syndrome, speech and language develop later and more slowly than in children without the syndrome, and their speech may be more difficult to understand.
- Some children with Down Syndrome have attention problems, and may need help to focus on tasks and when learning new things

**Slide 22: Common Genetic Syndromes Associated with IDD- Down Syndrome: Support Strategies**

In helping people with Down Syndrome, the following strategies can be helpful:

- See “oppositional” behavior as communication of frustration, and try to support the individual to better express needs. Be patient, and allow time for the person to learn. Always giving choices, even if just between 2 options, helps people have control.
- Also, acknowledging a person’s feelings without agreeing to their challenging behavior can help.
  - For example, rather than telling someone who is refusing to get off of the van that this is unacceptable and he must comply, you can say, “It seems like you are really worried about going in to the program today. How can I help you, because we will miss you if you don’t come!”
- Since people with Down Syndrome often do not state what they need in an effort to please others, it is important to be sure the individual knows you really want to know their opinion.
- For learning and attention problems, it helps to provide reminders and visual cues because people with Down syndrome may have communication challenges. Using multiple ways to get information across can help in the learning process
- Lastly, it helps to have a quiet place when teaching new skills and to repeat and reinforce steps in the learning process. It also helps to have the person repeat things back to ensure they understood and heard what was said.

**Slide 23: Application Exercise**

**Slide 24: Common Genetic Syndromes Associated with IDD:Fragile X Syndrome  
No Narration**

### **Slide 25: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Physical Characteristics**

Next we will cover Fragile X syndrome. We first list the physical characteristics and commonly associated medical problems followed by support strategies.

Some of the physical characteristics associated with the syndrome are:

- Long and narrow face
- Large ears
- Prominent jaw and forehead
- Unusually flexible fingers

### **Slide 26: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Medical Problems**

Common medical problems associated with fragile X syndrome include:

- Ear infections
- Seizures
- High blood pressure, also called hypertension
- Hernias
- Premature menopause
- Heart problems
- Joint hypermobility where joints move beyond a normal range
- Misalignment of the eyes (Strabismus), and
- Poor coordination or muscle control

### **Slide 27: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Medical Support Strategies**

Let's go over a few support strategies for these common medical problems.

- DSWs are often the first people to notice that someone they support is unhappy and may not be feeling well. It is important to recognize that people with an IDD, including people with Fragile X syndrome, often are poor reporters of their own health issues.
- People with this syndrome may have few ways to show distress, so challenging behaviors at times are the only sign that a person is uncomfortable or in pain.
- Some clues that new or worse challenging behaviors are related to a health condition include when this occurs and at the same time, the person also has shown changes in their fluid or food intake, how much they sleep or looking tired, and having a different pattern of behavior.

The following several slides cover common traits and accompanying support strategies for people with Fragile X syndrome.

### **Slide 28: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 1 of 7)**

There are some common traits in people with this syndrome. Let's review a few.

- The presence of intellectual disabilities which may vary from mild to severe.
- Generalized Anxiety Disorder and Panic Disorder are common mental health conditions.
- In women, depression and mood disorder is also common.
- Autism Spectrum Disorder or autistic features are very common, affecting about 1 in 3 individuals with Fragile X syndrome. This will impact both communication and social interaction. See the table under course resources for more specifics on autism spectrum disorder symptoms.
- Additionally, a great place to learn more is at the website for Autism Speaks provided here : <https://www.autismspeaks.org>

**Slide 29: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 2 of 7)**

- People with Fragile X have sensitivity to noises. Crowds, and touch, are also a frequent trigger.
- To help with this, avoid excessive sensory or emotional stimulation, such as noise, and crowds. In general, be considerate to the individual's sensory sensitivities.
- Remember that sometimes noises that don't bother you may be too much for the person with Fragile X.

**Slide 30: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 3 of 7)**

Continuing with common traits and strategies,

- People with Fragile X may have challenging behavior because of anxiety.
- High anxiety can sometimes cause risk for "fight or flight" panic driven behavior. This can lead to aggression or self-injurious behavior. Hand biting is also common.
- Also, a person with Fragile X who is very anxious may also have rapid speech, which can be mistaken for a mental illness, specifically "mania."

**Slide 31: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 4 of 7)**

- To help with these traits it's important to realize that anxiety can come from not knowing what to expect. DSWs can help by being sure the individual is told what will be happening, and maintaining a predictable routine as much as possible
- Use tactics that help the person be less anxious about new things and transitions, and to be more relaxed. Examples might be swimming, taking

- walks, or using other relaxation tools, based on their person centered goals. A therapist can help DSWs have a plan for relaxation tactics.
- It is useful to provide reassurances often, versus “prompts.”
  - People with more severe functional limitations may need much more structured support, such as using a visual schedule. Visual schedules and structure serves to help reduce anxiety.
  - Lastly, set up predictable routines so the person knows when transitions will occur, and give warnings, use timers and as noted, use picture schedules or calendars, to show that a transition is coming and when to expect it.

**Slide 32: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 5 of 7)**

- People with Fragile X have often present with anxiety, impulsive, and hyperactive behavior such as fidgeting or impulsive actions. Attention deficit disorder (ADD) is common (which includes an impaired ability to maintain attention and difficulty focusing on specific tasks.)
- To help with this, consider what supports are in place for the individual and if they are appropriate or need modification. Are there things in the environment that could be increasing anxiety and contributing to challenging behavior?
- People with Fragile X can lead fulfilling lives, have friends, and employment. DSWs can make a difference in thoughtful planning and modifying the environment to meet the needs of the individual to set them up for success!

**Slide 33: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 6 of 7)**

- Another common trait in people with Fragile X Syndrome is being very shy.
- People are often fooled thinking that individuals with Fragile X don't want to be social when really, they may be unsure of how to make friends.
- They may have “social anxiety” meaning meeting new people causes them anxiety, but they will enjoy being social when given help and time to build relationships.
- Also, a distinctive feature common to people with fragile X is “gaze aversion,” meaning poor eye contact, looking down or away from you.

**Slide 34: Common Genetic Syndromes Associated with IDD- Fragile X Syndrome: Common Traits and Support Strategies (Slide 7 of 7)**

- To help with these traits, plan activities in the community that will not be overwhelming to the person and make sure that the right supports are available. Be thoughtful in planning goals and the demands that they may include, especially in the area of socialization.

- While generally we want to look someone in the eye to make sure he is paying attention, this may heighten the anxiety in a person with Fragile X syndrome. Be thoughtful in your social interactions.
- Next, we can arrange the person's environment in a way that maximizes individual learning style. For example, sit next to them as opposed to sitting across from them, when working on tasks. Use praise and encouragement to enhance and build upon their strengths.
- Try to arrange a good fit between strengths, skills and interests, and daily activities. For instance, a person who is very shy and dislikes noise will work better in a situation where it is not too loud and crowded.
- Teach simple social skills like how to greet someone you are meeting for the first time and taking turns when talking to others

### **Slide 35: Application Exercise**

### **Slide 36: Common Genetic Syndromes Associated with IDD: William's Syndrome**

### **Slide 37: Common Genetic Syndromes Associated with IDD- William's Syndrome: Physical Characteristics**

The next syndrome we will cover is William's syndrome. We will start with physical characteristics and commonly associated medical problems. Some of the physical characteristics associated with the syndrome are:

- An "Elfin" facial appearance with a small upturned nose
- Short stature
- Wide mouth with full lips
- Partially missing teeth, defective tooth enamel, or small, widely spaced teeth, and
- Sloping shoulders

### **Slide 38: Common Genetic Syndromes Associated with IDD- William's Syndrome: Medical Problems**

Some of the common medical problems people with William's syndrome may have are:

- Heart valve problems
  - High blood pressure
  - Problems with coordination
  - Gastrointestinal problems; chronic constipation, stomach pain, rectal prolapse,
  - Urinary tract infection, or
  - Urinary incontinence
- In general, if you notice changes in the behavior or mental health of the person you are supporting, always rule out medical issues that may be occurring

**Slide 39: Common Genetic Syndromes Associated with IDD- William's Syndrome: Common Traits and Support Strategies (Slide 1 of 6)**

Now let's look at some shared traits associated with William's Syndrome, and discuss how we can provide focused and positive support. In general, people with this syndrome are diagnosed with

- Intellectual disabilities which range from mild to moderate.
- ADHD, Anxiety, Phobias, and Depression, are common mental health conditions that are associated with Williams Syndrome.

**Slide 40: Common Genetic Syndromes Associated with IDD- William's Syndrome: Common Traits and Support Strategies (Slide 2 of 6)**

- Continuing with common traits, individuals with this syndrome tend to be extremely sociable and endearing, and tend to take an extreme interest in other people. Speaking and social skills are big strengths.
- However, they are often described as having an overly friendly demeanor, and may trust strangers when they should not.
- People with this syndrome often have difficulties understanding social cues and this makes it harder to form lasting relationships. Because being social and having friends is so important, not being able to form meaningful and lasting relationships with others can lead to depression.

**Slide 41: Common Genetic Syndromes Associated with IDD- William's Syndrome: Common Traits and Support Strategies (Slide 3 of 6)**

Let's talk about some support strategies you can use for these common traits.

- Because people with William's Syndrome can be overly-friendly, it is important to protect them from unsafe interactions such as leaving with a stranger or telling a stranger too much about themselves
- Opportunities for social interaction are important. Support individuals to have meaningful social experiences in their daily lives, such as getting involved in community activities, church events, sports teams, or other types of social events.
- Many adults with this syndrome can contribute to their communities as volunteers or paid employees. For example, working at assisted living homes, hospitals, and libraries, or as store greeters provide opportunities for social interaction as well as meaningful employment.

**Slide 42: Common Genetic Syndromes Associated with IDD- William's Syndrome: Common Traits and Support Strategies (Slide 4 of 6)**

- Another common trait is that people with William's syndrome tend to do well on tasks that involve spoken language, music, and learning by repetition

(rote memorization). Language abilities, facial recognition, and short-term auditory memory are also big strengths.

- However, they can have challenges with visual-spatial tasks such as drawing and assembling puzzles, poor hand-eye coordination, and fine motor control. This can make doing relatively minor tasks very difficult, causing frustration and anxiety.
- Other difficulties in learning have to do with numbers, perceptual planning, and abstract reasoning, which can make daily tasks a challenge. Keep in mind that people with William's Syndrome often have relatively advanced language skills that can mask all of these cognitive challenges. This may mistakenly give a DSW the impression that they are more capable than they actually are.
- When teaching skills that may be more difficult for the person with Williams syndrome to learn, try to make it fun, social and use a tool the person enjoys such as music.
- Be thoughtful in setting expectations and working on skill building. Because of a desire to make friends, the person you're supporting may take on more than he is able, which can increase overall anxiety. Instead, get to know the person so you can recognize challenges AND strengths. Be careful not to make assumptions based on first impressions. In this way, you can help to establish the best suited goals for that person.

**Slide 43: Common Genetic Syndromes Associated with IDD- William's Syndrome: Common Traits and Support Strategies (Slide 5 of 6)**

- People with Williams Syndrome may be hypersensitive to loud sounds, and
- Loud sounds or physical contact can sometimes cause anxiety.
- As a support strategy, it is key to avoid places with loud noise.

**Slide 44: Common Genetic Syndromes Associated with IDD- William's Syndrome: Common Traits and Support Strategies (Slide 6 of 6)**

- People with William's Syndrome also often have special musical talents! There is even a Band comprised of all individuals who have Williams syndrome and who have wonderful musical talents. The band is called the KanDoo Band and can be found at the following link. <https://williams-syndrome.org/marchapril-2012#Kandoo%20Band%20Releases%20Original%20Song%20on%20iTunes!>
- In supporting these people, remember that anxiety is often at the core of any kind of challenging behaviors. Check on what might be causing anxiety, and be supportive and reassuring. Music is a great way to promote wellness and create a calming environment for these individuals.

- **Celebrate the individuals you support.** Promote their strengths and reinforce their accomplishments!

**Slide 45: Common Genetic Syndromes Associated with IDD: Cornelia de Lange Syndrome**

**No Narration**

**Slide 46: Common Genetic Syndromes Associated with IDD- Cornelia de Lange Syndrome: Physical Characteristics**

Next, we will discuss Cornelia de Lange syndrome. Common physical characteristics of this syndrome include:

- Small and widely spaced teeth
- Small upturned nose
- Low-set ears
- Arched eyebrows that often meet in the middle
- Long eyelashes
- Some limb abnormalities, as in their legs or arms
- Excessive body hair
- Short stature, and
- Microcephaly, which is an unusually small head

**Slide 47: Common Genetic Syndromes Associated with IDD- Cornelia de Lange Syndrome: Medical Problems**

Common medical problems include:

- Gastro-Esophageal Reflux Disease, or GERD
- Heart problems
- Vision problems
- Hearing loss
- Sleep disturbances
- Seizures and a
- Cleft palate, which is an opening in the roof of the mouth

Medical problems seen in people with this syndrome are highly variable from one affected person to the next!

**Slide 48: Common Genetic Syndromes Associated with IDD- Cornelia de Lange Syndrome: Medical Support Strategies**

- Because of GERD, it is recommended not eating close to when someone will go to bed and knowing what foods make this condition worse. For example, spicy or acidic foods or caffeine may be triggers.
- Wearing tight clothes can also make the symptoms of this medical condition worse
- It is important to recognize that challenging behaviors may be provoked by pain or discomfort from GERD. So when you observe someone with Cornelia

- De Lange Syndrome becoming very irritable or having new or increasing challenging behaviors, ask that they be checked for health problems.
- Some clues that GERD may be a problem include problems sleeping as this medical condition gets worse when people lay down and having challenging behaviors after eating.
  - Some people are described as vomiting on purpose when really, they do not have control.
  - Due to the many medical issues commonly associated with this syndrome, individuals with Cornelia De Lange Syndrome need to be screened for a number of health problems. DSWs may advocate for this to happen and help support the person if he or she is anxious about medical appointments.
  - If this does occur, using stories and visual boards to explain what will happen at the medical appointment can be helpful to reduce anxiety.

**Slide 49: Common Genetic Syndromes Associated with IDD- Cornelia de Lange Syndrome: Common Traits**

Now let's look at some traits that are commonly associated with people with Cornelia DeLange Syndrome, and discuss how we can provide focused and positive support.

In general, people with Cornelia DeLange Syndrome have:

- Intellectual disability that typically range from moderate to severe
- Depressed mood and anxiety are also common
- Obsessive-compulsive features and compulsive self-injurious behavior, specifically skin picking, can be a very difficult challenge for people with this syndrome.
- Additionally, Cornelia DeLange Syndrome is correlated with high rates of Autism Spectrum Disorder, significant sensory abnormalities, and social shyness. However, people with Cornelia DeLange Syndrome can also enjoy making friendships with others.

**Slide 50: Common Genetic Syndromes Associated with IDD- Cornelia de Lange Syndrome: Support Strategies**

Here are some specific support strategies for these common traits.

- Because of obsessive compulsive features and anxiety, the use of visually presented, predictable, structured, daily schedules and tactics are very helpful
- Riding, swimming and other similar activities help sensory challenges.
- Use visual aids, pictures or technology such as an I-PAD to help with communication.
- To help reduce anxiety, it is useful to very slowly introduce new things.
- Lastly, helping people have fun and meaningful activities as well as helping insure that health problems are addressed can help prevent depressed mood.

**Slide 51: Common Genetic Syndromes Associated with IDD: Prader Willi Syndrome**  
**No Narration**

**Slide 52: Common Genetic Syndromes Associated with IDD- Prader Willi Syndrome: Physical Characteristics**

Next, we will move on to Prader Willi syndrome. Some of the physical characteristics associated with Prader Willi syndrome are:

- Almond shaped eyes
- Thin upper lip
- Small downturned mouth,
- Small hands and feet
- Nasal sounding speech
- Short stature

**Slide 53: Common Genetic Syndromes Associated with IDD-Prader Willi Syndrome: Medical Problems**

Common medical problems associated with this syndrome are:

- Obesity, including risk for type 2 diabetes mellitus; high blood pressure and other consequences of obesity
- Sleep apnea
- Bone density problems
- Seizures, and
- Low muscle tone
  
- To provide support for these medical problems, promoting exercise is one of the most helpful strategies, since this is very important to promote overall wellness.

**Slide 54: Common Genetic Syndromes Associated with IDD- Prader Willi Syndrome: Common Traits and Support Strategies (Slide 1 of 6)**

Now let's look at some traits that are commonly associated with people with Prader Willi Syndrome and discuss how we can provide focused and positive support.

In general, people with Prader Willi syndrome can have:

- Intellectual disabilities that are mild or within the range of borderline intellectual functioning. It is also the case they may have no intellectual disabilities at all.
- Common mental health conditions include anxiety, obsessions and compulsions and to a lesser extent, depression and bipolar disorder. Although psychotic and impulse control disorders are also seen.
- People with Prader Willi Syndrome may engage in many repetitive and obsessive behaviors, including hoarding, over-concern with symmetry and exactness, and difficulty with changes in routines.

- In addition, individuals with this syndrome can have in some serious self-injurious behaviors, especially skin picking.

**Slide 55: Common Genetic Syndromes Associated with IDD- Prader Willi Syndrome: Common Traits and Support Strategies (Slide 2 of 6)**

As a support strategy, psychoactive medications can be helpful at times, but in some cases they can make symptoms worse, especially if the medication used increases appetite. It helps when DSWs use other tactics such as encouraging exercise, assisting the individual in having a meaningful day, and following an individual's positive behavior support plan (when applicable) to promote mental wellness and reduce anxiety.

**Slide 56: Common Genetic Syndromes Associated with IDD Prader Willi Syndrome: Common Traits and Support Strategies (Slide 3 of 6)**

- The most distinctive trait in people with Prader Willi Syndrome is an excessive food drive.
- Challenging behavior is often driven by obsessions with, and anxiety around, obtaining food.
- The obsession with food is so strong in some people with PWS that they will even eat food out of the garbage and frozen foods.

**Slide 57: Common Genetic Syndromes Associated with IDD- Prader Willi Syndrome: Common Traits and Support Strategies (Slide 4 of 6)**

Support strategies for these traits may include:

- Providing carefully controlled diets and support around access to food, because of severe health risks if not provided.
- Planning meals ahead of time, including a regular and dependable meal schedule.
- Trying to avoid being vague about meal times. For example, avoid saying things like, "we'll eat dinner, **later**" or "We're not going to have lunch, **right now.**" Be specific and say instead, "We're going to have dinner at 6:00." In some cases, a watch may be helpful to alleviate a person's anxiety about food and mealtimes.
- It is critical to be sensitive to the fact that a person with Prader Willi Syndrome cannot help the desire for more food. Their brains do not signal that they are full and they really feel very hungry.
- DSWs can use tactics such as providing predictable routines, and small healthy snacks more often. Do not give individuals treats or rewards as this may actually cause them more anxiety.
- Remember to always follow the dietician, physician, or psychologist's orders regarding food for each individual.

**Slide 58: Common Genetic Syndromes Associated with IDD Prader Willi Syndrome: Common Traits and Support Strategies (Slide 5 of 6)**

- Other common traits for people with this syndrome include being very social.

- People with Prader Willi Syndrome often have mild cognitive challenges and are aware of their challenges and differences.
- They may feel shame about their challenges, and can have poor self-esteem as a result.
- They tend to have strengths in the areas of long-term memory, reading, and visual-spatial functioning, especially completing jigsaw puzzles!

**Slide 59: Common Genetic Syndromes Associated with IDD- Prader Willi Syndrome: Common Traits and Support Strategies (Slide 6 of 6)**

- In supporting individuals with Prader Willi syndrome, it is important to help these individuals focus on their strengths and positive accomplishments.
- Positive Behavior Supports can be very helpful with focus on individual strengths and interests to engage, have fun and divert the focus from food, or other obsessive tendencies like hoarding things.
- Use of a reward system can be very helpful to reinforce positive and replacement behaviors, but do not use food as a reinforcer.
- Use of visual daily schedules to help set up expectations can alleviate anxiety. Including meal schedules on the calendar can also help.
- Never tell a person with Prader Willi Syndrome that you will withhold food due to behavior. This will increase their anxiety and increase the likelihood that they will have challenging behavior. It also keeps them from progressing toward mental wellness.
- Lastly, always follow an individual's Positive Behavior Support Plan!

**Slide 60: Application Exercise**

**Slide 61: Individuality**

We have now reviewed several genetic syndromes so that you can provide the best support possible to the individuals you serve. However, it is important to remember, a person is not defined by their syndrome!

**Every individual is still unique!** Even people with the exact same micro-deletion will have differences in how they develop, depending on environmental influences and life experiences. Every individual has unique skills and strengths as well as challenges, unique dreams and visions and unique potential.

**Slide 62: Case Study Judy**

Judy, a 28 year old woman with Down syndrome and moderate intellectual disability, had been struggling with depression off and on for a few years. Judy lived in a group residence, but spent many weekends with her parents, and had regular visits from her sister. Judy also had hypothyroidism, and had a heart problem when she was a child that was surgically corrected. Recently, she started to look very sad and depressed again, after the family pet (a dog) passed away. She did show some improvement when her anti-depressant medication was adjusted, but was not really getting back to her usual presentation. It also turned out that her thyroid

medication dose needed to be increased. This was done and she seemed less depressed, but still, not the usual Judy. When feeling well, Judy was a very warm, funny and social person who was very creative. She loved painting and most any type of arts and crafts. Her DSW found out that a local church was having a holiday event, and people were going to be selling their handmade items for gifts. She arranged for Judy to have a spot, and helped her get started making ornaments. Judy really brightened up after this and began to be more active and happier.

### **Slide 63: Supporting Individuals with Genetic Syndromes**

Understanding a person's genetic syndrome provides valuable information in helping you to establish a foundational relationship and using strategies to maximize a person's opportunities for success. But remember, getting to know the individual for who they are, outside of their genetic syndrome, will help you to truly support their mental wellness and help them to achieve positive outcomes.

You can make a large difference, even for people with a definite genetic syndrome, by helping them have an enriched environment and positive experiences that build on their unique individual strengths.

Medical treatments helped Judy to some degree, but these were not enough to get her back to feeling well again. Ultimately, the efforts of her DSW were the most powerful influences on her feeling and functioning better.

### **Slide 64: Genetics Syndromes: Summary**

By understanding unique traits, strengths, and challenges associated with Genetic Syndromes, we can provide more informed and targeted care. Many examples were discussed that include how we can:

- Recognize when someone may have a genetic syndrome associated with IDD.
- Know the individual's medical conditions and recognize how feeling ill may be shown in mood and behavior, or may even look psychiatric on the surface.
- Understand the importance of learning about these syndromes and how they impact a person's physical and mental health
- Follow professionals' recommendations;
- Listen, communicate and be patient (do not assume the person is "doing it on purpose") when they may have trouble remembering, learning or understanding.
- Provide more reminders and use tools like visual aids to help the person understand what is expected and what will happen next.
- Pair verbal instructions with modeling, as well as visuals, and repeat instructions.

### **Slide 65: Genetics Syndromes: Summary, Continued**

- Be sure people can have fun when learning and that they are taught practical skills with a focus on their strengths, interests, and learning styles, to help them live happier and healthier lives.

- Use Positive Behavior Supports to teach and reinforce skills, and make changes to the individual's environment to set them up for success
- Recognize the powerful influence a DSW can have on outcomes, even for people with a genetic syndrome, because enriching experiences change outcomes for the better
- Develop opportunities for people with an IDD to help them be engaged and connected socially, that are informed by our understanding of the person's developmental profile and behavioral phenotype
- People with a genetic syndrome and an intellectual and developmental disability are more like you and me than they are different. Focus on how "normal" they are instead of how "different" they are. Syndromes are a medical label, not who the person is. Be sure and get to know the person and then read their medical chart.
- In the case of all of these syndromes and disorders, DSWs are able to make a strong and powerful impact on quality of life for the people for whom you provide care. DSWs can help people reach their most optimal potential by providing a positive atmosphere with lots of opportunities.
- YOU can make a difference!

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