

# Down Syndrome & Alzheimer's Disease

OCTOBER 25, 2024

Down Syndrome  
Indiana™



# Down Syndrome Indiana™



Down Syndrome Indiana is a one stop shop for information and resources related to Down syndrome. We support individuals with Down syndrome and their families from diagnosis through adulthood!

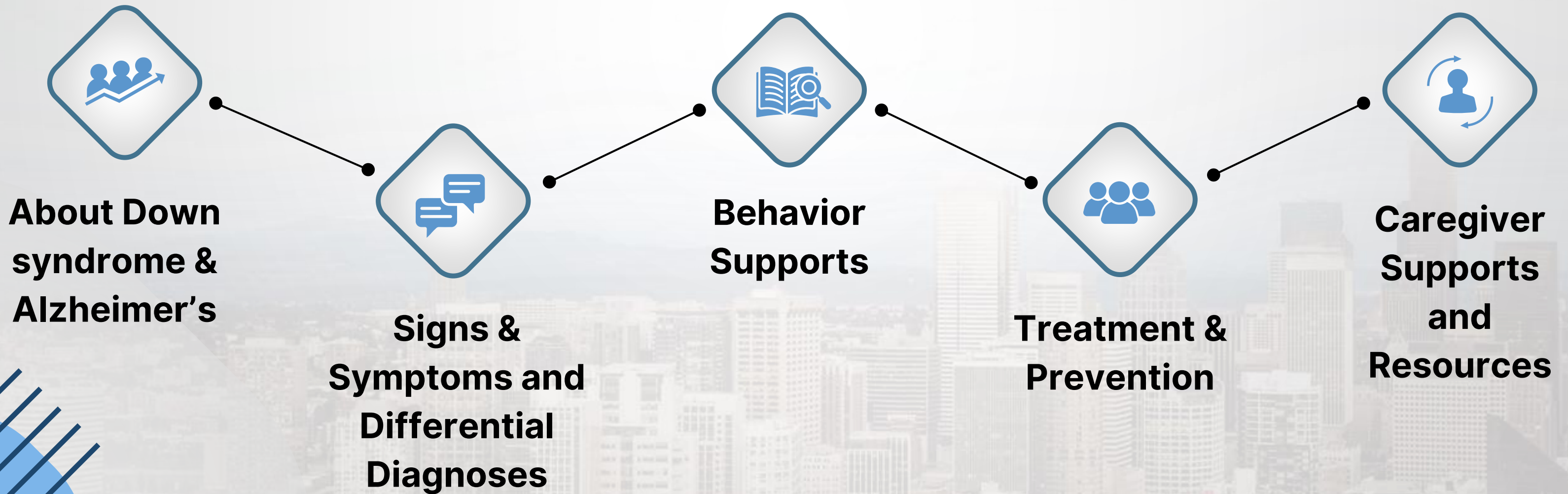


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# Today's Agenda





# Down syndrome 101

## Basics

- Named for the physician that first characterized the condition in 1865, Dr. John Langdon Down
- Individuals with Ds have an extra copy of chromosome 21 - giving them 47 chromosomes instead of 46
- The extra 21st chromosome is responsible for the differences that we see in individuals with Ds
- Despite those differences, individuals with Ds are more like us than different!

## 3 Types of Down syndrome

- Trisomy 21
  - Extra copy of 21st chromosome in all cells
  - Occurs in 95% of people with Ds
- Mosaic Down syndrome
  - Extra copy of 21st chromosome in some cells
  - Occurs in 1-2% of people with Ds
- Translocation Down syndrome
  - Extra copy of 21st chromosome attaches to other chromosome
  - Occurs in 3-4% of people with Ds
  - Only type of Ds that is hereditary

## Prevalence

- 1 in every 700 babies will be born with Ds
- About 7,000 - 8,000 individuals with Ds living in Indiana
- About 350-400,000 individuals with Ds in the US
- Indiscriminate of race, ethnicity, socioeconomic status

# Alzheimer's Disease

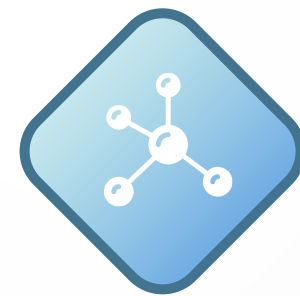
Alzheimer's disease is:

- progressive brain disorder
- the most common cause of dementia
- slowly destroys memory



## Irreversible Changes

Changes in the brain might begin 10 years before symptoms appear.



## Beta-Amyloid Plaque

A hall mark of Alzheimer's disease is the presence of protein clumps called beta-amyloid plaque



## Where Does It Come From

The precursor to Beta-Amyloid Plaque, amyloid precursor protein (APP) is produced by the 21st chromosome.

# The Ds / Alzheimer's Connection



## 21st Link

Because people with Ds have a 3rd copy of chromosome 21, they produce more APP



## Aging & Ds

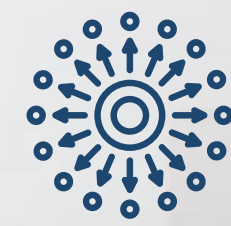
Most people with Ds will have beta amyloid plaques by the age of 40



## Numbers

But not everyone with these plaques will develop symptoms of Alzheimer's disease

4-5:30/6:00.



## Prevalence

It is estimated that 50% of people with Ds will develop Alzheimer's related dementia.

# Signs and Symptoms

## Changes in Behavior

- Increased confusion
- STM problems (ie: repetitive questions)
- Change in ability to do ADL (reduction or loss of ability)

## Other S&S

- Difficulty with coordination/walking
- Decreased ability to pay attention
- Personality and behavior changes
- Change in sleep
- Decreased fine motor control
- Difficulty navigating familiar areas/locations



# Diagnosis

Diagnosis can be challenging because:

- Most individuals with Ds can't self report memory concerns
- Assessing thinking-skill changes is challenging in individuals with IDD

Recommendations:

- Baseline adult function by age 35
- Look for changes in day to day function
- Evaluation by professional in dementia if concerns arise (but cognitive function testing shouldn't be the only benchmark)
- Rule out differential dx





# Differential Diagnosis

- Vision loss
- Hearing loss
- Hypothyroidism
- Obstructive Sleep Apnea
- Osteoarthritis
- Atlantoaxial Instability

(Alzheimer's Disease and Down syndrome: A practical guidebook for caregivers p 7)

## **Other things to consider....**

- Depression
- Vitamin B-12 deficiency
- Celiac disease
- Heart disease
- Medication side effects/interactions
- Metabolic disease (kidney, diabetes, calcium abnormality)

(National Down Syndrome Society, n.d.)



# Ds Regression Disorder

What is it: DSRD occurs between individuals between 10 - 30yo with Ds. Characterized by loss of skills (including thinking, socialization, and ADLs).

## Signs and Symptoms:

- Loss of adaptive skills
- Difficulty talking
- Depression
- Change in obsessive behaviors
- Increase in repetitive behaviors
- Fatigue, headaches, irritability,
- Anxiety
- Aggression
- Attention seeking behavior
- Self injurious behavior
- Difficulty concentrating
- Over active bladder
- Change in food habits
- Stubbornness
- Prefers to be alone

## Treatment:

- Treatment of medical conditions contributing
- Benzodiazepines, Antidepressants or Antipsychotics
- Immunotherapy or Electroconvulsive therapy



NDSS has a great  
DSRD Symptom Checklist!



# Behavior Supports

Get back to the basics....



# Behavioral Supports

## Communication

- Simplify verbal communication as disease progresses
  - shorter sentences, smaller words
  - pair words with gestures, gentle touch, or pointing
- As verbal skills decline, individual with Alzheimer's Disease will rely on:
  - emotional cues
  - tone of voice
  - facial expression
  - body language



# Adaptive Strategies

It can be helpful to “learn the language of dementia”. Successful interactions depend on caregivers and providers adapting language, tone, and messages to suit the individual.



## **It's Not Anyone's Fault. It's A Disease**

If frustrated, avoid raising your voice, expressing disappointment, repeating rules, etc. This will give you the opposite result.



## **Stay in the Moment**

Expect to repeat yourself, offer multiple reminders, listen to the same questions/stories over and over.



## **Make Simple Requests**

Focus on strategies that reinforce sense of calm, reassurance, safety, and security. Avoid future based rewards which depend on short term memory!

# Dealing with Negative Behavior

## Examine the Behavior

- What was the behavior?
- Did something trigger it?
- What happened immediately after?
- Could something be causing pain?
- Could this be related to med or illness

## Explore Potential Solutions

- Are the individual's needs being met?
- Can adapting the surrounding comfort the individual?
- Can I change my own reaction or approach?

## Try Different Responses

- Did my new response help?
- Do I need to explore other potential causes and solutions?
- What can I do differently?



# Behavioral Supports

## Try this:

- Include the individual in conversation
- Take time to listen
- Talk directly to the individual - face to face
- Use humor
- Allow time for response
- Be patient and supportive
- Offer instructions one step at a time
- Ask one question at a time
- Ask yes/no questions rather than open ended
- Give visual cues
- Look to the emotions rather than the words

## Avoid this:

- Negative tones/words
- Criticizing or correcting
- Arguing

# A Meaningful Day

When planning for a meaningful day focus on:

- Person
  - do activities that the person enjoys and adapt as needed
  - provide choice
  - consider physical limitations/problems (ie: vision/hearing loss, OA)
- Activity
  - Focus on enjoyment not achievement
  - Be flexible and adapt as needed - time of day is important
- Approach
  - Give support, cues, and supervision
  - break activities into steps and do one step at a time
  - don't criticize or correct
  - try again another day/time if unsuccessful
- Place
  - choose safe activities
  - minimize distractions, have supplies ready
  - ensure nothing in the environment will be frightening

(National Down Syndrome Society, 2019, p.33)





# Treatment

- Significant research is happening across the country!
- Treatments are made challenging by difference in metabolic factors in people with Ds (slower resting metabolic rate), people with Ds absorb meds differently, and are at increased risk of adverse reactions and poor outcomes
- Some drugs that are proven effective in the general population have not shown efficacy in the Ds population
- Recently, a few new promising anti-amyloid drugs have been developed and approved by the FDA but individuals with Ds were not included in the clinical trials\*





# Prevention

- Regular cardiovascular exercise
- Stay mentally stimulated and engaged
- Don't smoke
- Lower risk factors for stroke and heart disease
- Avoid preventable head trauma
- Eat a healthy, balanced diet
- Get rest
- Take care of mental health
- Stay socially engaged

# Caregiver Supports

Encourage caregivers to

- Practice good self care
- Stay active
- Stay mentally stimulated
- Remain socially engaged
- Create a support network
- Be kind to themselves



# End of Life Considerations



## Promoting Dignity and Choice

Medical decisions and death  
How individual chooses to spend time



## Advanced Directives and Decision Making

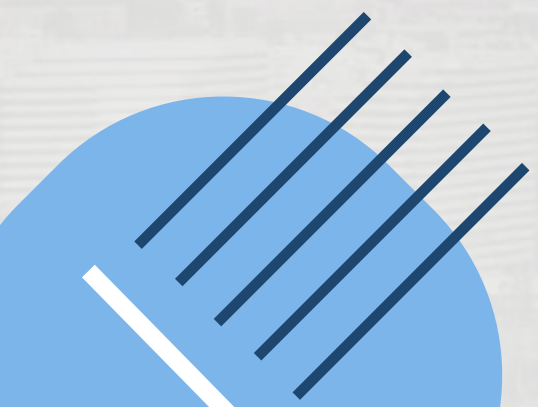
- Do Not Resuscitate (DNR) or Do Not Intubate (DNI)
- Medical Orders for Life Sustaining Treatment (MOLST)
- Healthcare Proxy v Guardianship



## Late-Stage End-of-Life Care

- Palliative Care
- Hospice
- Comfort Care
- End-of-life doula

# Resources



## Psychosocial Assessment Overview & Common Presentation Checklist

This checklist or “toolkit” is intended to support the health of adults with Down syndrome directly or through their caregivers. We encourage you to share this checklist with your medical provider. Per the GLOBAL Medical Care Guidelines for Adults with Down Syndrome, adults with Down syndrome should undergo a psychosocial assessment each year. A psychosocial assessment is an interview, usually performed by a social worker or psychologist, where they ask about the patient’s mental, physical, and emotional health factors. Below is a chart outlining the 3 basic areas of a psychosocial assessment, and the changes associated with those areas of functioning that may prompt a caregiver or parent to make a special appointment with a medical or behavioral professional such as a doctor or psychologist familiar with adults with Down syndrome.

3 Basic Areas of the Psychosocial Assessment:	Examples of things to notice in behavior, function, and psychosocial areas:	Things to Review with a Medical Professional:
<b>Behavioral Factors</b>	<p>TAKE NOTICE OF CHANGES IN:</p> <ul style="list-style-type: none"> <li>• Self-talk</li> <li>• “Groove” behaviors (tendency towards sameness or repetition)</li> <li>• Personality</li> <li>• Ability to focus or pay attention</li> <li>• Activity level</li> <li>• Social motivation or engagement</li> <li>• Mood and/or temperament</li> </ul>	<p>Type of Changes:</p> <ul style="list-style-type: none"> <li>• Increase or decrease of the number of times a behavior is displayed in self-talk, groove behaviors, etc</li> <li>• Increase or decrease of levels around focus, activities, social engagement, etc.</li> <li>• Start of new behaviors</li> </ul> <p>Behaviors that should be immediately addressed:</p> <ul style="list-style-type: none"> <li>• Self-harm, Harm to others</li> <li>• Loss of interest in things they previously enjoyed</li> </ul> <p>Past psychological history (diagnosis and/or medication)</p>
<b>Functional - Adaptive Factors</b>	<p>TAKE NOTICE OF CHANGES IN:</p> <ul style="list-style-type: none"> <li>• Social skills</li> <li>• Communication skills <ul style="list-style-type: none"> <li>○ Receptive (Listening to others and understanding)</li> <li>○ Expressive (Speaking or otherwise communicating to others)</li> </ul> </li> <li>• Activities of daily living (ADLs) including bathing, dressing, eating, toileting</li> <li>• Sleep patterns/Sleep hygiene (bedroom and routines that promote consistent, uninterrupted sleep)</li> <li>• Strengths &amp; weaknesses</li> </ul>	<p>Types of Changes:</p> <ul style="list-style-type: none"> <li>• Ability to learn new skills</li> <li>• Eating more or less (appetite changes)</li> <li>• Difficulty doing things they previously could do (or needing more support from others)</li> <li>• Confusion around familiar people and places</li> <li>• “Obsessive Slowness” (almost looks like intentionally moving in slow motion)</li> </ul>
<b>Psychosocial Factors</b>	<p>TAKE NOTICE OF CHANGES IN:</p> <ul style="list-style-type: none"> <li>• Family/friends/support staff</li> <li>• Key relationships</li> <li>• Housing arrangement</li> <li>• School/occupation/vocation</li> <li>• Community involvement</li> </ul>	<p>External influences that change the level of participation in activities:</p> <ul style="list-style-type: none"> <li>• Changes in medication</li> <li>• Divorce, breakups, grief</li> <li>• Changes in routine</li> </ul> <p>Immediately report instances of trauma or abuse to police/child services and/or medical professionals, and seek supports</p>

# Psychosocial Assessment



## Evaluating Challenging Behaviors in Adults with Down Syndrome

Down Syndrome Medical Interest Group: DS-ASD Workgroup

**Challenging behavior escalations (e.g., aggression towards self or others) in an adult with Down syndrome may indicate the need to:**

1. Explore etiology of the behavior and any recent environmental changes or perceived stressors.
2. Rule out a medical cause.
3. Evaluate mental health.
4. Consider an autism or regression assessment as indicated.

### Behavioral Assessment

#### **Important considerations:**

- In order to help reduce the risk of exacerbating anxiety and escalating behaviors, remember the need for a gentle tone and longer processing time when clinically assessing individuals with DS.
- Sensorimotor behavior (stimming) can be a stress reliever in individuals exhibiting challenging behaviors, necessitating tolerance of this behavior in the office.
- Be sensitive of your tone when working with the caregiver.

#### **Explore etiology of challenging behaviors:**

- Many behaviors do not have a medical or psychological cause; rather they are situational.

#### **Review any recent environmental changes or perceived stressors:**

- Consider abuse/neglect.
- Individuals with DS tend to remember events like a movie: the emotions, words, actions that were stressful may continue to replay in their minds as a relived experience.
- Timing of behavioral escalations may help identify the cause.

# Evaluating Challenging Behavior in Adults with Ds



# Resources

Alzheimer's Association. (n.d.). *Down syndrome and Alzheimer's disease*. <https://www.alz.org/alzheimers-dementia/what-is-dementia/types-of-dementia/down-syndrome>

Bulova, P., Capone, G., Chicoine, B., Harville, T., Martin, B., McGuire, D., Mckelvey, K., Peterson, M., Tsou, A., Tyler, C., Sie Whitten, M., Gelaro, B., & Wells, M. (2018). *GLOBAL medical care guidelines for adults with Down syndrome*. <https://www.globaldownsyndrome.org/medical-care-guidelines-for-adults/>

Mass General for Children. (2023). *Down syndrome and regression*. <https://www.massgeneral.org/children/down-syndrome/down-syndrom-and-regression>

Moran, J. (2023). *End-of-life and Down syndrome*. Washington, DC: NDSS.

National Down Syndrome Society. (n.d.). *Alzheimer's Disease & Down syndrome* - <https://ndss.org/resources/alzheimers>

National Down Syndrome Society. (2019). *Alzheimer's Disease & Down syndrome A practical guidebook for caregivers*. Washington, DC: NDSS.

National Institute on Aging. (2020). *Alzheimer's Disease in People With Down Syndrome [Fact Sheet]*. chrome-extension://efaidnbmnnnibpcajpcgclefindmkaj/https://order.nia.nih.gov/sites/default/files/2021-02/alzheimers-down-syndrome.pdf

Van Pelt, K., Head, E., Schmitt, F., Koehl, L.M. (2020). *Alzheimer's disease and Down syndrome*. Academic Press. <https://doi.org/10.1016/B978-0-12-813955-4.00020-9>





# Resources to Share

General Information about Down syndrome and Alzheimer's:

<https://www.alz.org/alzheimers-dementia/what-is-dementia/types-of-dementia/down-syndrome>

<https://ndss.org/resources/alzheimers>

<https://dsindiana.org/aging-down-syndrome/>

Caregiving a loved one with Down syndrome and Alzheimer's:

<https://ndss.org/resources/alzheimers-disease-down-syndrome-practical-guidebook-caregivers>

<https://www.dsrf.org/resources/blog/practical-preparations-for-the-aging-caregiver/>

<https://www.youtube.com/watch?v=uzVByHcjuhc>

<https://ndss.org/resources/aging-and-down-syndrome-health-well-being-guidebook>

<https://www.alz.org/help-support/caregiving/daily-care/communications>

<https://www.alz.org/help-support/caregiving/safety/home-safety>



Feel welcome to email Macy Pohl, M.S. OTD at [info@dsindiana.org](mailto:info@dsindiana.org) to request specific resources or for any questions regarding aging in place!

# Q&A Session



# Thank you from

## Down Syndrome Indiana™



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