CARE DOWN SYNDROME
national down syndrome society

CARE Down Syndrome

May 20, 2026

Speakers:
Anthony Sciullo
Dana Sciullo, MOT, OTR/L

Anthony Sciullo

- 33-years-old
- Works at an auto shop
- Outdoors
- Photography
- Going to the gym
- Travel

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Self Advocacy in Healthcare

Here are a few ways medical professionals can make me feel more comfortable:

- Greet me and introduce themselves
- Speak directly to me
- Treat me like an adult
- Tell me what's going on and what they're doing
- Speak positively about my situation
- Explain medical terms to me
- Show interest in what I like to do outside of the doctor's office
- Know about Down syndrome and how it might affect my health
- Ask permission before touching me
- Avoid medical jargon
- Ask me if I have any questions
- Allow me to have a caregiver present, if I want

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Self Advocacy in Healthcare

Speak up to your medical team about what you need

- For example:
  - I ask for a butterfly needle when having blood tests done
  - I ask my doctors to speak slower and louder because I am deaf in one ear

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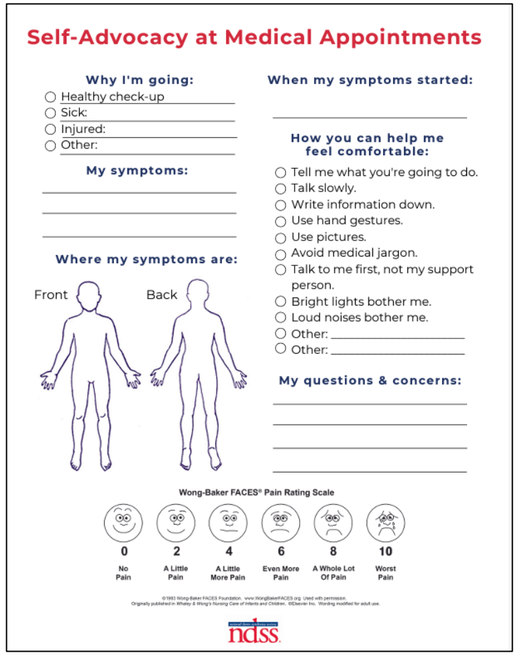
Clinician Resources



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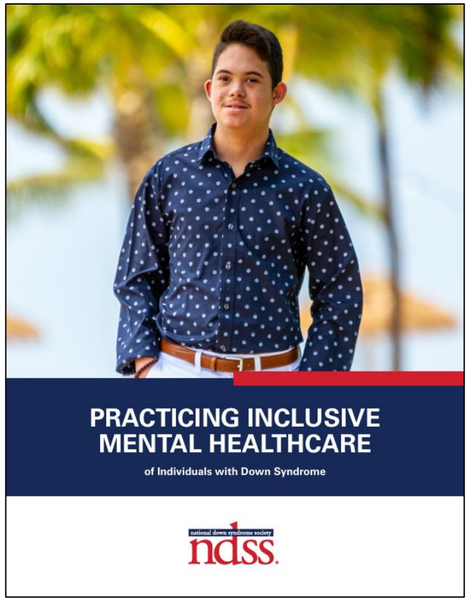

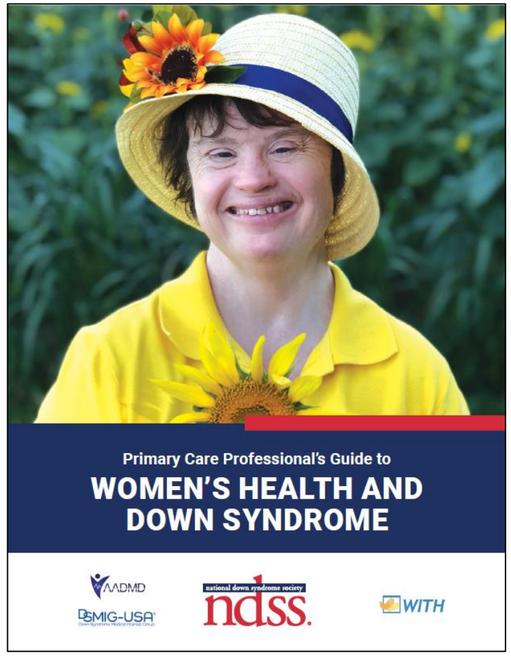

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Dana Sciullo, MOT, OTR/L

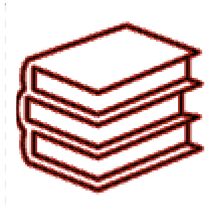
- Manager of Health and Wellness Programs at the National Down Syndrome Society

Mission

NDSS is creating a world where individuals with Down syndrome thrive.

NDSS Core Pillars

NDSS supports and advocates for the Down syndrome community by focusing on four key pillars:



Resources & Support

Research

Advocacy & Policy

Community Engagement

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NDSS Core Pillars

Resources & Support

- 1-800 helpline and info email
- Resources and information from birth to end of life
- Education, employment, health and wellness, aging, caregiving, and more

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Down Syndrome Regression Disorder

Down Syndrome Regression Disorder

Down syndrome regression disorder (DSRD), also referred to as regression, is a rare but serious disorder that occurs in some adolescents and young adults with Down syndrome. Regression is used to describe the loss of skills an individual has previously learned. These skills can be daily living, language, movement, or social skills. The loss is often sudden and occurs over a period of weeks to months. Since DSRD may be caused by a variety of factors, the first priority is to determine the most likely cause. Individuals who are younger than 10 years or older than 30 years are very rarely diagnosed with DSRD. If these individuals are experiencing regression-like symptoms, a close look at all other factors must be taken before giving the diagnosis of DSRD.

In 2022, a group of medical experts with experience treating regression in patients with Down syndrome gathered to create the first criteria for diagnosing DSRD. These criteria provide guidance to physicians and families who do not have access to specialists in DSRD. There are eight clusters of symptoms linked to regression, and some are broken down into specific examples. Individuals who are experiencing four or more of the eight symptom clusters are considered "possible" cases of DSRD, meaning further investigation is needed. Individuals who have seven or eight of the eight symptom clusters are considered "probable" cases, meaning they are likely to receive a diagnosis of DSRD if no other cause can be found.

Testing

If you or your loved one are experiencing symptoms in the checklist (see page 3), it is recommended that you make an appointment with your primary care doctor or neurologist as soon as possible. The diagnosis of DSRD is mostly based on symptoms, but your doctor may order some of the following medical tests as part of the evaluation:

- Imaging of the brain with an MRI
- Blood work
- Urine tests (if necessary)
- Lumbar puncture (also known as a spinal tap)
- Electroencephalogram (EEG)
- Genetic or metabolic testing (if necessary)
- Polysomnogram (sleep study)

For more detailed testing recommendations, you and your doctor can read the Assessment and Diagnosis of Regression in Down Syndrome (Santoro et al., Front Neurol, 2022) https://www.frontiersin.org/articles/10.3389/fneur.2022.940175/full

DOWN SYNDROME:
Guidelines for Inclusive Education
DECEMBER 2021
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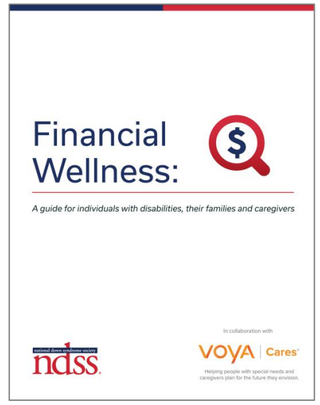
down syndrome education international
discovering potential · transforming lives


Financial Wellness:
A guide for individuals with disabilities, their families and caregivers
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In collaboration with
VOYA | Cares
Helping people with special needs and caregivers plan for the future they envision.

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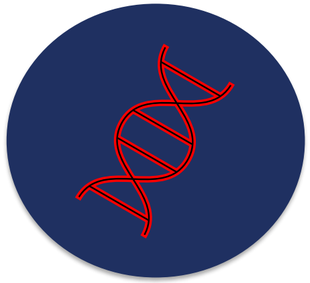
NDSS Core Pillars

Research

- Bridge between research and our community
- Coordinating research projects
- Preparing scientific industry companies for clinical trials
- Sharing research opportunities with families
- Expanding access to clinical trials, treatments, and diagnostics



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NDSS Core Pillars

Advocacy & Public Policy

- Federal, state, and local advocacy
- Develop and improve laws to positively impact people with Down syndrome across the country and affirm their human rights
- Legislative agenda spans the life experience of individuals with Down syndrome

NDSS Core Pillars

Community Engagement

- Scholarships, grants, and awards
- National Buddy Walk® Program
- Times Square Video Presentation
- Athlete Ambassador Program
- Virtual Racing for 3.21 for World Down Syndrome Day
- Gala, golf, and more!

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8-Fold Increase in Adult Population

People with Down Syndrome in the United States

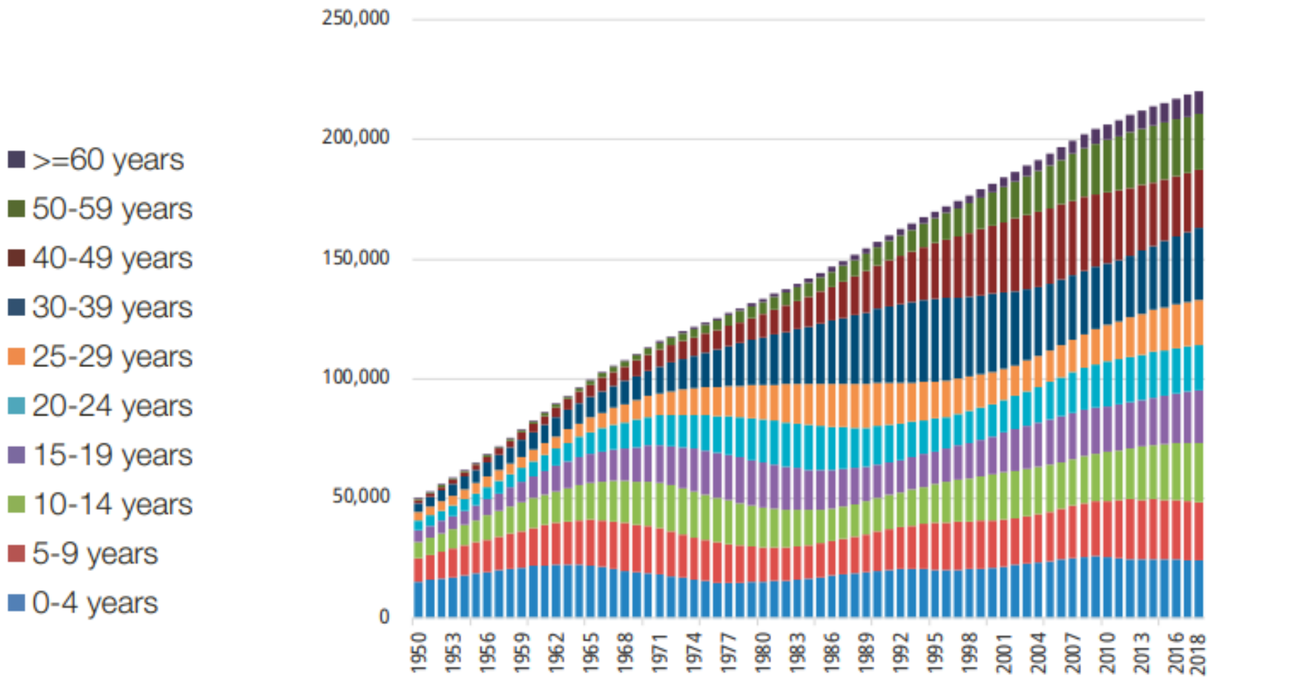

Figure 5. Population of people with Down syndrome in the USA, 1950-2018

Estimated number of people with Down syndrome 20 years of age or older

de Graaf G, Buckley F, Brian Skotko BG. People living with Down syndrome in the USA: births and population. *Down Syndrome Population*. Published May 2024. Accessed May 4, 2026. https://docs.downsyndromepopulation.org/factsheets/down-syndrome-population-usa-factsheet.pdf

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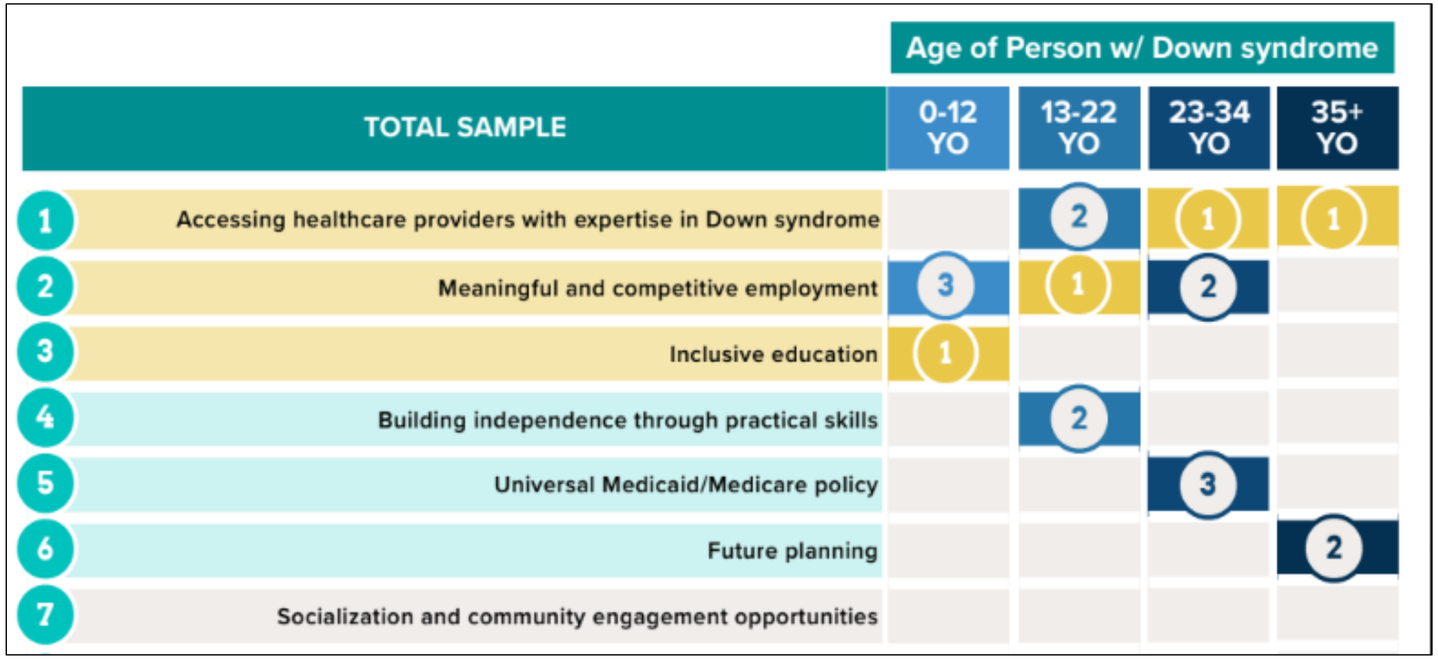
Only an estimated **5% of adults with Down syndrome** have access to specialty Down syndrome clinics. That means 95% rely on their primary care professional.

Santoro SL, Campbell A, Balasubramanian A, Haugen K, Schafer K, Mobley W. Specialty clinics for adults with Down syndrome: A clinic survey. *Am J Med Genet A*. 2021;185(6):1767-1775. doi:10.1002/ajmg.a.62169

CARE Down Syndrome

Community Needs: Market Research

- The Down syndrome community was asked to select the top three areas where they feel improvements are most needed.
- A top priority is access to medical professionals with expertise in Down syndrome.



	TOTAL SAMPLE	Age of Person w/ Down syndrome: 0-12 YO	13-22 YO	23-34 YO	35+ YO
1	Accessing healthcare providers with expertise in Down syndrome		2	1	1
2	Meaningful and competitive employment	3	1	2	
3	Inclusive education	1			
4	Building independence through practical skills		2		
5	Universal Medicaid/Medicare policy			3	
6	Future planning				2
7	Socialization and community engagement opportunities				

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Clinical and System Implications

- Diagnostic overshadowing
- Variability in care quality
- Challenges with transition to adult care

Where Education Falls Short

- Limited disability-focused training
- Primarily pediatric emphasis
- 36% of medical students felt adequately trained to care for patients with disabilities
- 97.6% felt they needed additional training

Marzolf BA, Plegue MA, Okanlami O, Meyer D, Harper DM. Are Medical Students Adequately Trained to Care for Persons With Disabilities?. *PRiMER*. 2022;6:34. Published 2022 Aug 30. doi:10.22454/PRiMER.2022.878147

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Elevating Primary Care for Adults with Down Syndrome

A Clinical Education Hub for Healthcare Professionals

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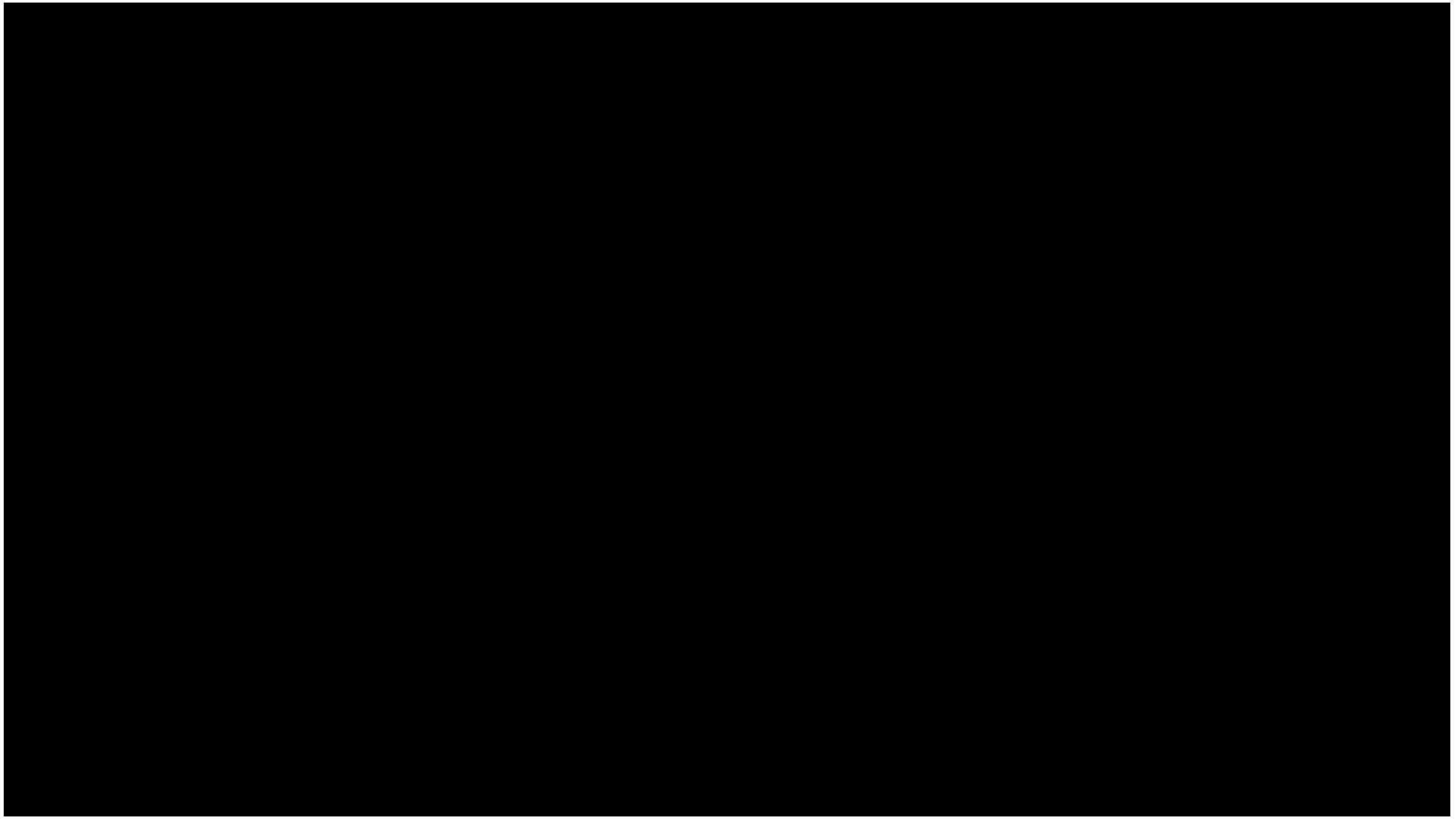

Informing the Content

- Listening sessions with adults with Down syndrome and families
- Insights gathered before development
- Data from 300+ NDSS calls and emails
- Topics based on real-world needs including Down Syndrome Regression Disorder, mental health, weight management, Alzheimer’s disease
- Emphasis both on knowledge and respectful care

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Authors and Reviewers

CARE Down Syndrome was made possible thanks to the collaboration of more than 40 healthcare professionals. From knowledgeable authors and expert peer reviewers to thoughtful pilot testers, each played a key role in shaping this resource. The Down Syndrome Medical Interest Group and the American Academy of Developmental Medicine and Dentistry have endorsed the eLearning course and articles. We're grateful for their commitment to improving healthcare for individuals with Down syndrome.

What You'll Find...

CARE DOWN SYNDROME
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Self-Paced eLearning Course

Reference Articles

Resource Library

CARE DOWN SYNDROME
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EARN

2-Hour Self-Paced eLearning Course

Earn continuing medical education (CME) credits while advancing your expertise in caring for adults with Down syndrome. This accredited course offers practical, evidence-based strategies for managing health conditions and was developed and peer-reviewed by leading national experts, including Brian Chicoine, MD.

EXPLORE

Clinical Reference Articles

Explore clinically relevant articles and resources covering key topics including Alzheimer's disease, Down Syndrome Regression Disorder, and mental health. Medical professionals can earn CME credits for each reference article reviewed by validating their participation within NDSS' learning platform.

ACCESS

Resource Library

Access our resource library for downloadable patient education materials and tools designed to help healthcare professionals deliver exceptional care to adults with Down syndrome.

- Down Syndrome Regression Disorder Checklist
- Alzheimer's Disease and Down Syndrome Guidebook
- Primary Care Provider's Guide to Women's Health and Down Syndrome

Case Example: Alzheimer's Disease

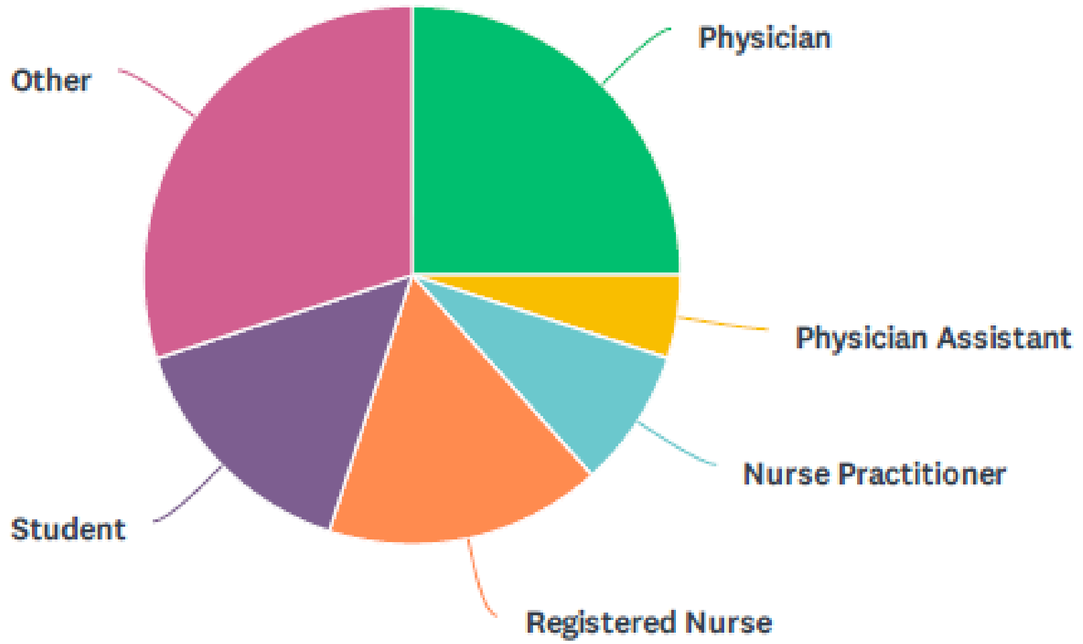
Example: Alzheimer's Disease in Down Syndrome

- Significantly higher risk due to third copy of the 21st chromosome
- Earlier onset than those without Down syndrome
- Different presentation

Feedback

Evaluation survey respondents identified as:

- 25% physician
- 5% physician assistant
- 8.33% nurse practitioner
- 16.67% registered nurse
- 15% student
- 30% “Other”



Feedback

Feedback we have received on the foundational eLearning course

- *"This was an excellent course. I gained knowledge in regard to clinical and social concepts for patients with Down syndrome. Thank you!"*
- *"The patient vignette format was very helpful to internalize the material presented. I also greatly appreciate that this content was developed with and informed by persons with Down syndrome."*
- 100% of eLearning course evaluation survey respondents said they would recommend it to a colleague

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Personal Perspective

Bridging the Gap

- Family experience
- Clinician partnership
- Shared responsibility

Share CARE Down Syndrome

Share the initiative with your local medical community.

- Primary Care Clinicians
- Medical Schools
- Hospital Systems

Together,

we can raise awareness and promote informed, compassionate care.

Tell Your Story

Personal stories are powerful.

Whether you're a caregiver or self-advocate, sharing your experience can inspire others, raise awareness, and help shape the future of care. Has your clinician participated in CARE Down Syndrome? Tell us about the difference it has made in your life.

Let others know what compassionate, informed care looks like

—and why it matters.

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Join us in elevating primary care for **adults with Down syndrome**

Visit **careds.org** to learn more