DS-Connect®

The Down Syndrome Registry

Get DS-Connected: Join, Participate, Discover!

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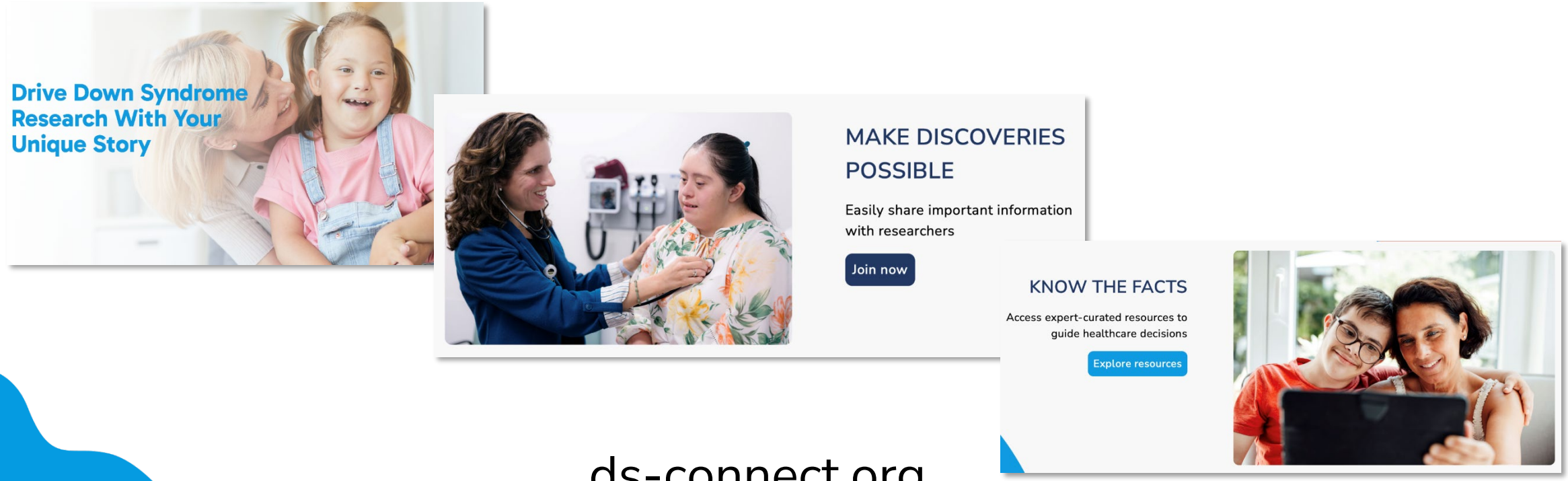
NIH *Eunice Kennedy Shriver* National Institute of Child Health and Human Development

THE INCLUDE PROJECT

What is DS-Connect?

DS-Connect®: The Down Syndrome Registry was developed to facilitate information-sharing and research participation among people with Down syndrome, their families, and investigators.



ds-connect.org

What is DS-Connect?

- A secure online survey-based tool developed for and by the Down syndrome community.
- Provides families with access to trusted research studies, events, and resources, while collecting privacy-protected demographic and health information.
- The information shared helps scientists identify patterns in related health conditions and advance research that informs care, services, and future scientific discovery.

History of DS-Connect

2006
NIH director asks NICHD to create a Down Syndrome Working Group (DSWG)

2007
The DSWG creates the NIH Research Plan on Down Syndrome and proposes the creation of a national registry

2013
The DS-Connect Registry is launched

2018
The INCLUDE Project is launched

2024
DS-Connect pauses operations for a refresh and new platform

2025
DS-Connect Is back!

What is a registry?

A registry is a resource to collect and store information about people affected by a specific medical condition.

Registries have been instrumental in advancing biomedical research and catalyzing discoveries for many medical conditions.

https://ds-connect.org/faq

Frequently Asked Questions

For the purpose of registering in DS-Connect®: The Down Syndrome Registry, "the person with Down syndrome" will refer to the person who has Down syndrome. "You" will refer to the person entering the information. This may be the person with Down syndrome or a family member or guardian of the person with Down syndrome (the person legally responsible for the care of the person with Down syndrome).

If you have questions about how to register, please visit our Help page.

- What is a registry?
- What is a research study?
- Why should I join DS-Connect?
- Why do we need DS-Connect?
- Who is sponsoring DS-Connect?

What is an epidemiological study?

An epidemiological study is a scientific investigation that examines the patterns, causes, and distribution of health-related conditions and events in a population.

People with Down syndrome have a different ‘clinical risk profile’

Less of:

Cancer

Atherosclerosis

Hypertension

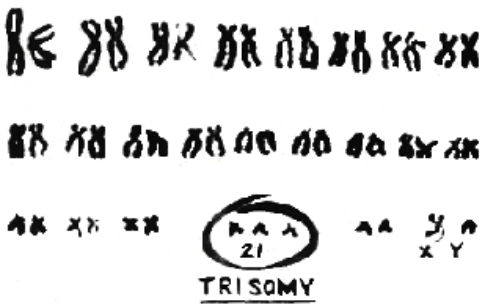
Allergies

Common (but variable) traits:

Stunted growth

Neurodevelopmental delays

Early ageing



More of:

Autoimmunity

Alzheimer’s

Lung infections

Vision and hearing issues

Congenital heart disease, autism spectrum disorders, seizures disorders, and more...

What are the other risks and resiliencies associated with Down syndrome?

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What are the **top two** causes of death for **children with** versus **without** Down syndrome **in the USA**?

Without Down syndrome:

- ❑ Respiratory illness
- ❑ Motor vehicle crash
- ❑ Firearm related injury
- ❑ Congenital anomalies

With Down syndrome:

- ❑ Respiratory illness
- ❑ Motor vehicle crash
- ❑ Firearm related injury
- ❑ Congenital anomalies

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- ❑ Congenital

With Down syndrome:

- ✓ **Respiratory illness**
- ❑ Motor vehicle crash
- ❑ Firearm related injury
- ✓ **Congenital anomalies**

https://pmc.ncbi.nlm.nih.gov/articles/PMC6637963/
https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.61924
https://www.thelancet.com/journals/lancet/article/PIIS0140-6736(02)08092-3/

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What are the **top two** causes of death for **adults with** versus **without** Down syndrome **in the USA**?

Without Down syndrome:

- ❑ Heart disease
- ❑ Pneumonia
- ❑ Cancer
- ❑ Alzheimer's disease

With Down syndrome:

- ❑ Heart disease
- ❑ Pneumonia
- ❑ Cancer
- ❑ Alzheimer's disease

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With Down syndrome:

- ❑ Heart disease
- ✓ **Pneumonia**
- ❑ Cancer
- ✓ **Alzheimer's disease**

https://www.cdc.gov/nchs/fastats/leading-causes-of-death.htm
https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.61924
https://pubmed.ncbi.nlm.nih.gov/35604690/

What is a natural history study?

A natural history study is a type of research that observes and documents the progression of a medical condition over time in a population of individuals, without any intervention or treatment.

Down syndrome is a lifelong condition with different manifestations across the lifespan

High risk of congenital heart defects

What happens in between?

High risk of Alzheimer's disease

What is the prevalence of **cataracts** in Down syndrome?

- ❑ 1%
- ❑ 4%
- ❑ 33.3 %
- ❑ 60%

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What is the prevalence of **cataracts** in Down syndrome?

- ❑ 1%
- ✓ **4% in the Human Trisome Project (all ages)**
- ✓ **33.3 % in the ABC-DS study (older adults)**
- ❑ 60%

When does the risk of cataracts increase?

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What happens in between?

High risk of Alzheimer's disease

What is the **mean age of diagnosis** for Alzheimer's disease in Down syndrome?

- ❑ 45 years old
- ❑ 54 years old
- ❑ 58 years old
- ❑ 64 years old

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- ✓ **54 years old**
- ❑ 58 years old
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Key registry functions

How it Works

Surveys

Fill out confidential health-related surveys written by researchers who are studying Down syndrome.

Research

Researchers use data from thousands of participants to make discoveries.

Participate

When researchers design new studies or clinical trials, you can opt-in to be notified and participate.

Resources

Find expert news, links and events, and see how the data from studies you've completed drives discoveries.

Top metrics

6350+ families registered

- Across the USA and some international participants
- 1000s of health surveys completed

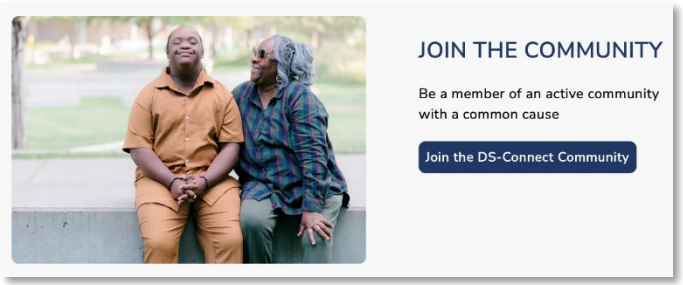
130+ research studies promoted

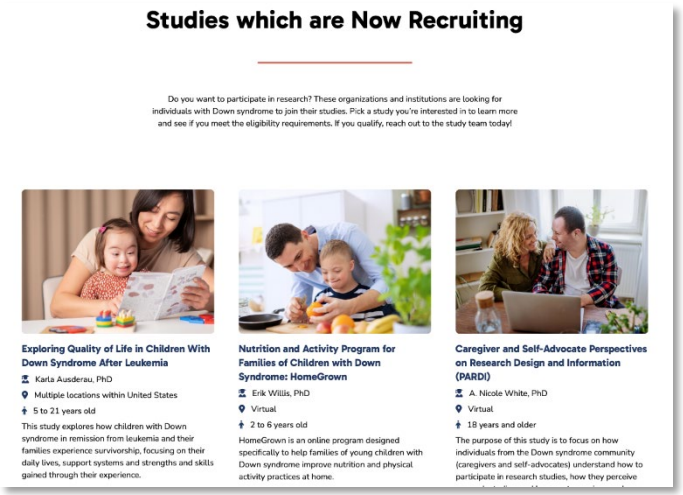
- 35 currently active

7200+ email subscribers

Countless outreach events organized

- Conferences, webinars, newsletters, and much more…

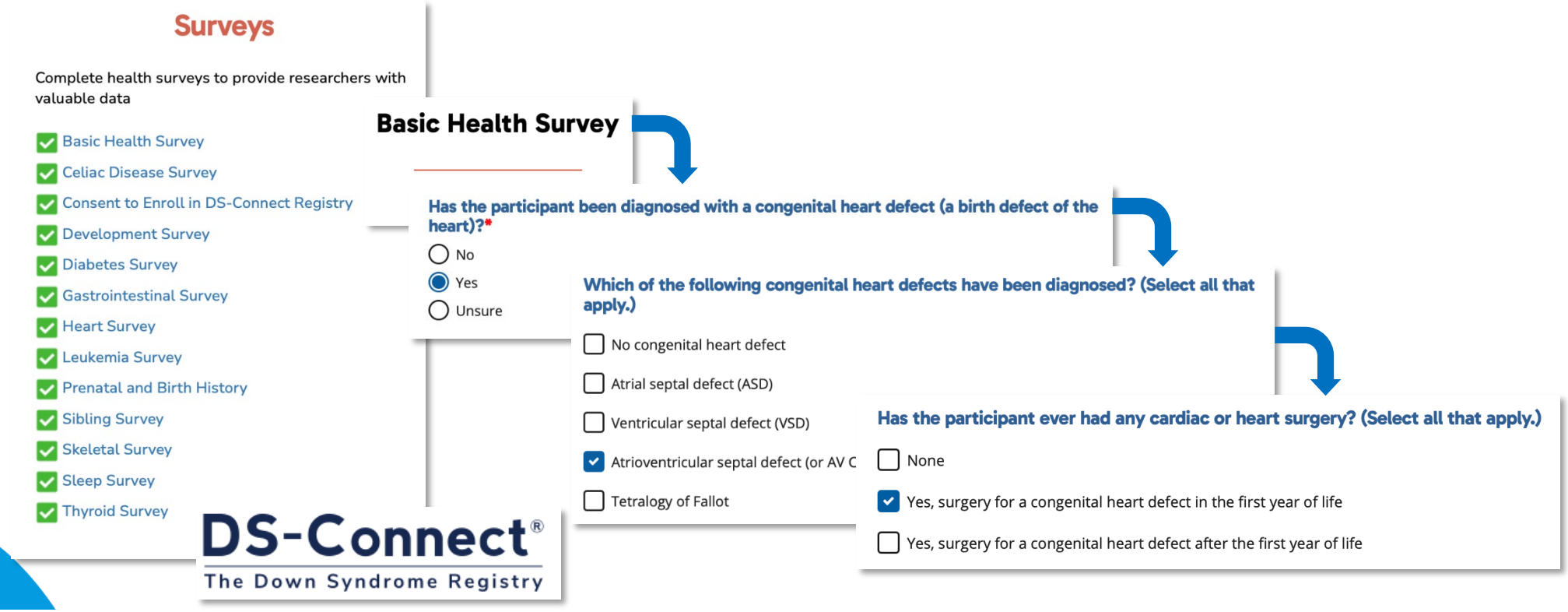




How is information collected?

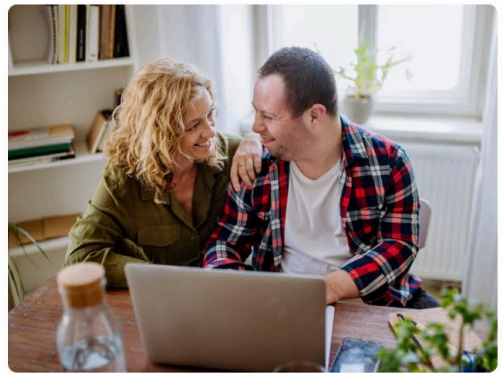
The information is collected through online surveys designed by experts in various disciplines.

Surveys can be completed in a ‘modular fashion’ over time.



The importance of self-reported data

Family-reported registry data provide a unique and indispensable source of information in Down syndrome research because they capture real-world experiences, health outcomes, and quality-of-life measures that are often unavailable in medical records or traditional research studies.



Families know best!

The importance of self-reported data

Self-advocates and families have important information that is not readily available to physicians and scientists.

Example: Down syndrome Regression Disorder (DSRD)

Story behind the first clinical trial for DSRD:

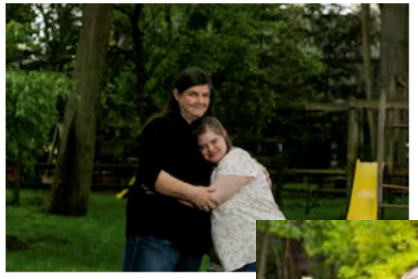
Well+Being

A mystery illness stole their kids' personalities. These moms fought for answers.

Their children's decline was dramatic, with patients losing function in days or weeks, including the ability to talk, move or take care of themselves.

May 12, 2024

The Washington Post



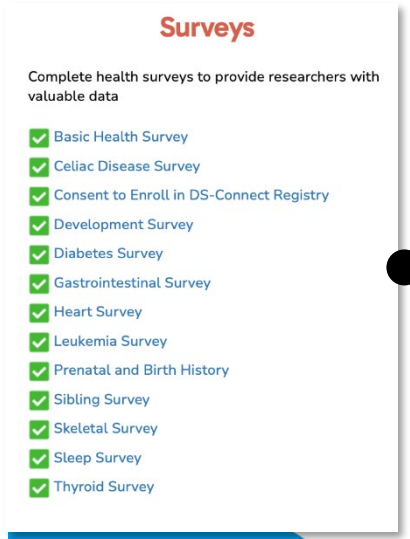
The importance of self-reported data

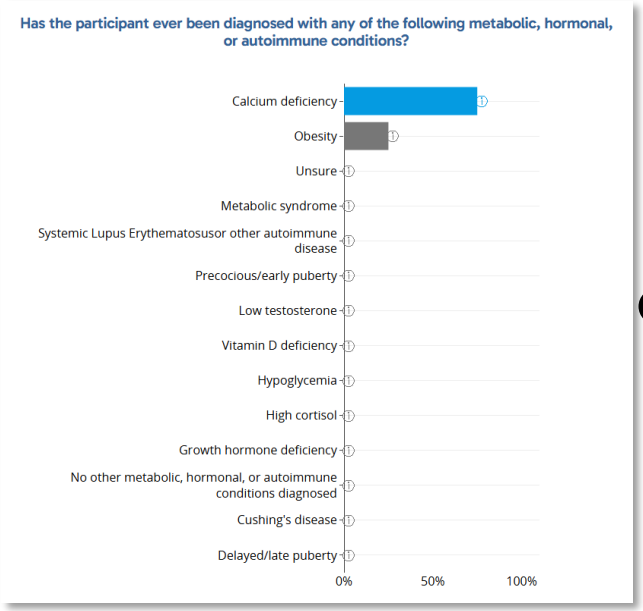
- ✓ **Capture** the full spectrum of the Down syndrome population
- ✓ **Enable** lifespan research
- ✓ **Identify** outcomes that matter to families
- ✓ **Discover** rare medical issues and sources of heterogeneity
- ✓ **Accelerate** clinical trial readiness
- ✓ **Empower** the Down syndrome community

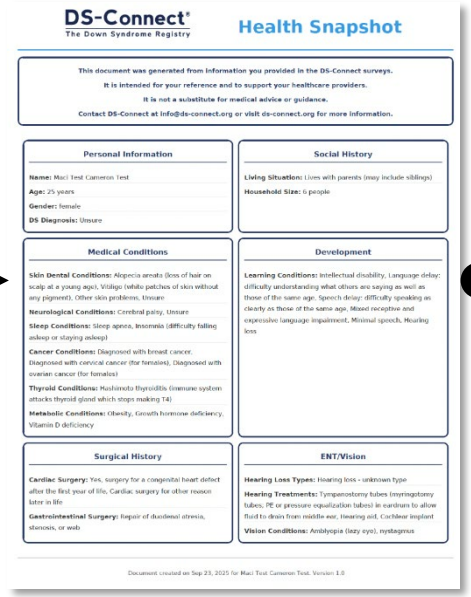
"Nothing for us without us"

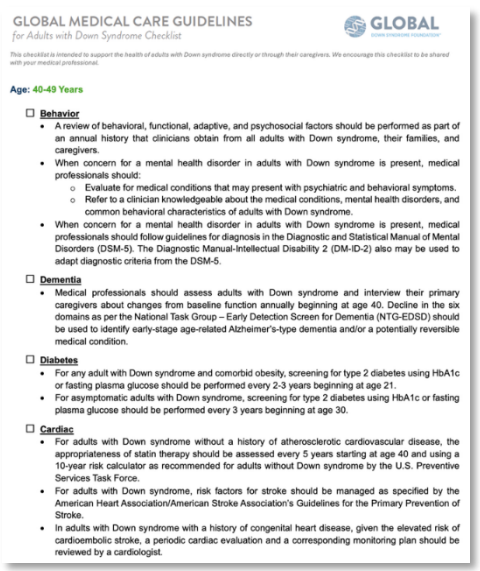
Empowering the families with their own data

1. Complete a survey
2. Visualize your data in comparison to community data
3. Download a friendly summary of your data
4. Download an individualized medical care checklist









Fueling the families' curiosity while rewarding survey completion

Data visualization is organized following the flow and logic of the surveys.

You open an 'accordion' to see data for that specific module or question...

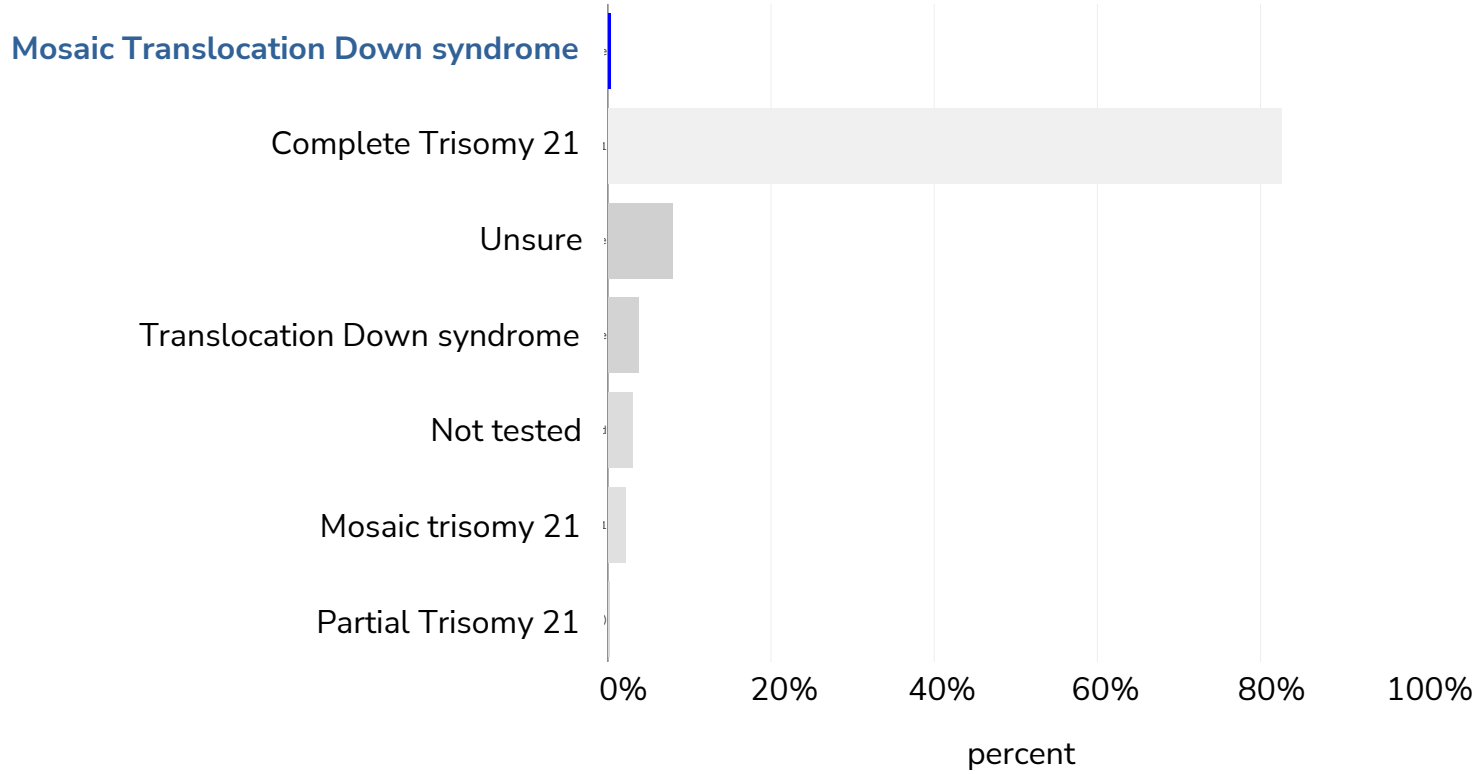
The more questions you answer, the more data you see...

Community Data

See how the data you've entered compares with the Down syndrome community data.

Diagnosis

What is the participant's Down syndrome diagnosis?



Basic Health Survey:

What is the participant's Down syndrome diagnosis?*

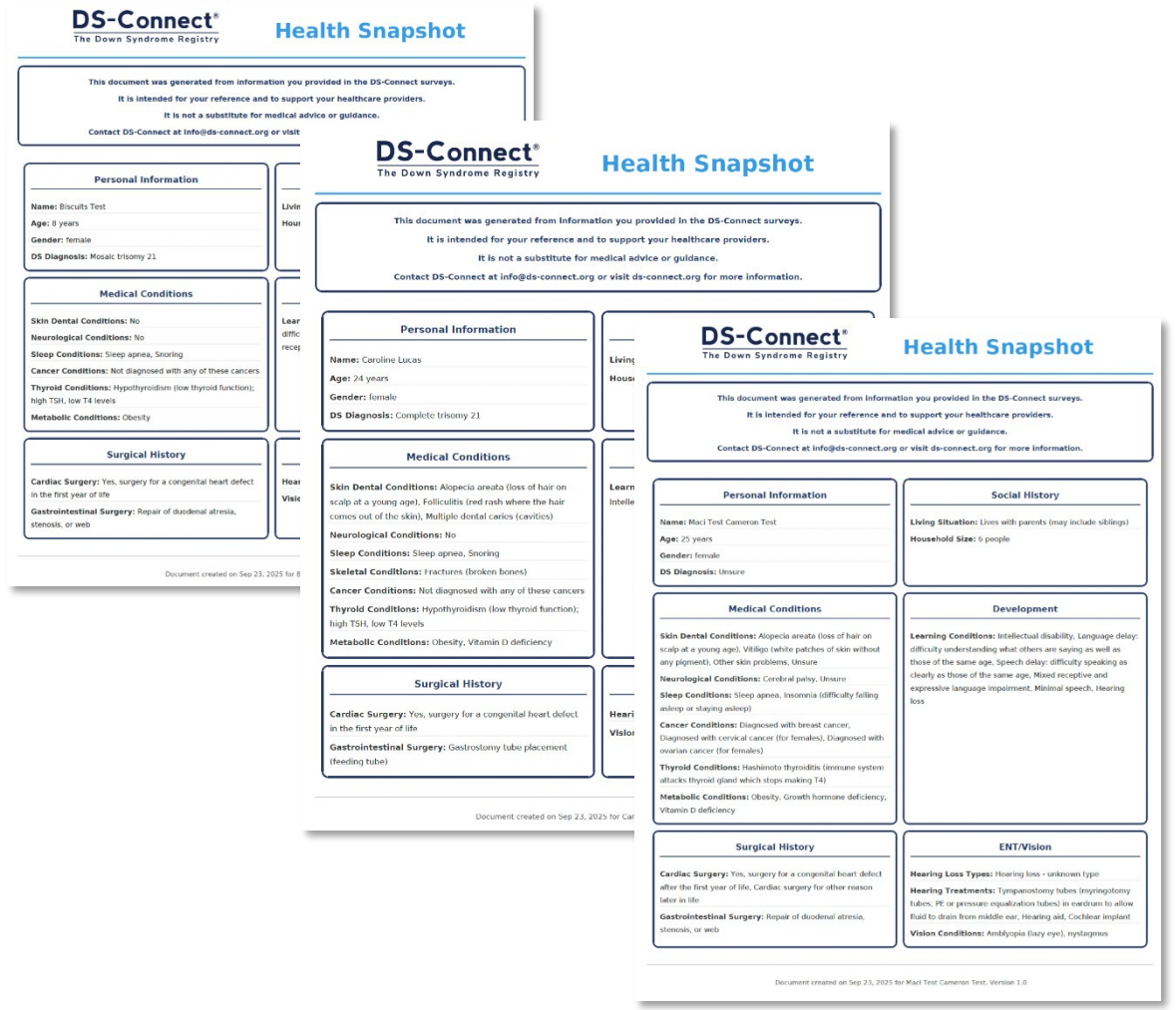
- ○ Complete trisomy 21
- ○ Mosaic trisomy 21
- ○ Translocation Down syndrome
- ◉ Mosaic Translocation Down syndrome
- ○ Partial Trisomy 21 (only a part of chromosome 21 is present in 3 copies)
- ○ Not tested
- ○ Unsure

The DS-Connect Health Snapshot

A friendly summary of the answers provided.

Can be downloaded as a pdf and shared with friends, caregivers, physicians, researchers.

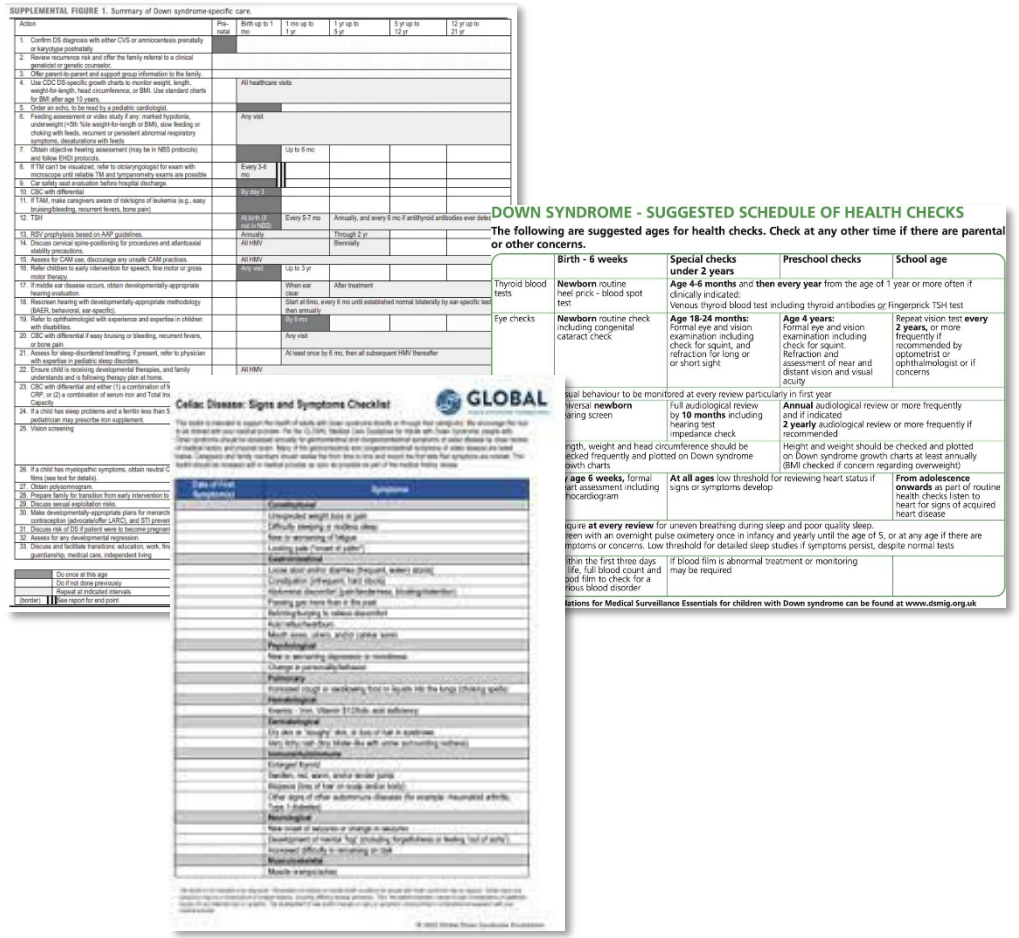
Updates with any new survey completed and revised answers.



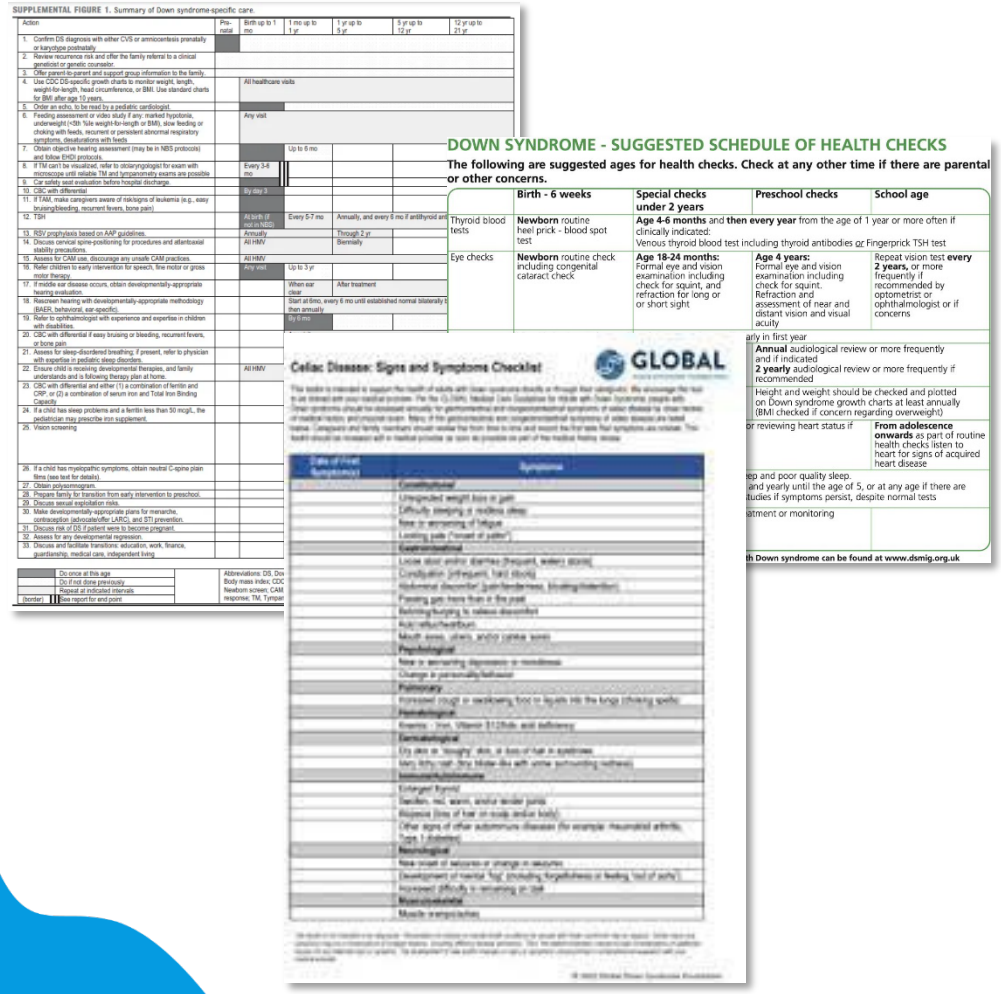
Facilitating access to medical care guidelines

Families often struggle to find the latest versions of the medical care guidelines that apply to the age and characteristics of the participant.

Using answers from the Basic Health Survey, we can provide the guidelines that apply to them.



Facilitating access to medical care guidelines



SUPPLEMENTAL FIGURE 1. Summary of Down syndrome-specific care.

Action	Pre-natal	Birth up to 1 mo	1 mo up to 1 yr	1 yr up to 5 yr	5 yr up to 12 yr	12 yr up to 21 yr
1. Confirm DS diagnosis with either CVS or amniocentesis prenatally or karyotype postnatally						
2. Review recurrence risk and offer the family referral to a clinical geneticist or genetic counselor.						
3. Offer parent-to-parent and support group information to the family.						
4. Use CDC DS-specific growth charts to monitor weight, length, weight-for-length, head circumference, or BMI. Use standard charts for BMI after age 10 years.		All healthcare visits				
5. Order an echo, to be read by a pediatric cardiologist.						
6. Feeding assessment or video study if any: marked hypotonia, underweight (<5th %ile weight-for-length or BMI), slow feeding or choking with feeds, recurrent or persistent abnormal respiratory symptoms, desaturations with feeds		Any visit				
7. Obtain objective hearing assessment (may be in NBS protocols) and follow EHDI protocols.			Up to 6 mo			
8. If TM can't be visualized, refer to otolaryngologist for exam with microscope until reliable TM and tympanometry exams are possible		Every 3-6 mo				
9. Car safety seat evaluation before hospital discharge.						
10. CBC with differential		By day 3				
11. If TAM, make caregivers aware of risk/signs of leukemia (e.g., easy bruising/bleeding, recurrent fevers, bone pain)						
12. TSH		At birth (if done in NICU)	Every 5-7 mo	Annually, and every 6 mo if antithyroid ab		
13. RSV prophylaxis based on AAP guidelines.				Annually	Through 2 yr	
14. Discuss cervical spine-positioning for procedures and atlantoaxial stability precautions.				All HMV	Biennially	
15. Assess for CAM use, discourage any unsafe CAM practices.				All HMV		
16. Refer children to early intervention for speech, fine motor or gross motor therapy.				Any visit	Up to 3 yr	
17. If middle ear disease occurs, obtain developmentally-appropriate hearing evaluation.					When ear clear	After treatment
18. Reconvene hearing with developmentally-appropriate methodology (BAER, behavioral, ear-specific).					Start at 6 mo, every 6 mo until established normal bilaterally, then annually	
19. Refer to ophthalmologist with experience and expertise in children with disabilities.					By 6 mo	
20. CBC with differential if easy bruising or bleeding, recurrent fevers, or bone pain						
21. Assess for sleep-disordered breathing; if present, refer to physician with expertise in pediatric sleep disorders.						
22. Ensure child is receiving developmental therapies, and family understands and is following therapy plan at home.					All HMV	
23. CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and Total Iron Binding Capacity						
24. If a child has sleep problems and a ferritin less than 50 mcg/L, the pediatrician may prescribe iron supplement.						
25. Vision screening						
26. If a child has myelopathic symptoms, obtain neutral C-spine plain films (see text for details).						
27. Obtain polysomnogram.						
28. Prepare family for transition from early intervention to preschool.						
29. Discuss sexual exploitation risks.						
30. Make developmentally-appropriate plans for menarche, contraception (advocate/offer LARC), and STI prevention.						
31. Discuss risk of DS if patient were to become pregnant.						
32. Assess for any developmental regression.						
33. Discuss and facilitate transitions: education, work, finance, guardianship, medical care, independent living						

Do once at this age
Do if not done previously
Repeat at indicated intervals
(border) See report for end point

Abbreviations: DS, Down syndrome; BMI, Body mass index; CDC, Centers for Disease Control and Prevention; NBS, Newborn screen; CAM, complementary and alternative medicine; HMV, health maintenance visit; TM, Tympanic membrane; TSH, Thyroid stimulating hormone.

DOWN SYNDROME - SUGGESTED SCHEDULE OF HEALTH CHECKS

The following are suggested ages for health checks. Check at any other time if there are parental or other concerns.

	Birth - 6 weeks	Special checks under 2 years	Preschool checks	School age
Thyroid blood tests	**Newborn** routine heel prick - blood spot test	**Age 4-6 months** and **then every year** from the age of 1 year or more often if clinically indicated: Venous thyroid blood test including thyroid antibodies or Fingerprick TSH test		
Eye checks	**Newborn** routine check including congenital cataract check	**Age 18-24 months:** Formal eye and vision examination including check for squint, and refraction for long or or short sight	**Age 4 years:** Formal eye and vision examination including check for squint. Refraction and assessment of near and distant vision and visual acuity	Repeat vision test **every 2 years,** or more frequently if recommended by optometrist or ophthalmologist or if concerns
			arly in first year	**Annual** audiological review or more frequently and if indicated **2 yearly** audiological review or more frequently if recommended
				Height and weight should be checked and plotted on Down syndrome growth charts at least annually (BMI checked if concern regarding overweight)
			or reviewing heart status if	**From adolescence onwards** as part of routine health checks listen to heart for signs of acquired heart disease
			ep and poor quality sleep. and yearly until the age of 5, or at any age if there are tudies if symptoms persist, despite normal tests	
			atment or monitoring	

th Down syndrome can be found at www.dsmig.org.uk

Celiac Disease: Signs and Symptoms Checklist GLOBAL

Date of Visit: Symptom(s)	Symptoms
	Constitutional
	Unexplained weight loss or gain
	Difficulty sleeping or restless sleep
	New or worsening of fatigue
	Loosing pain ("onset of pain")
	Gastrointestinal
	Loose stool and/or diarrhea (frequent, watery stools)
	Constipation (infrequent, hard stools)
	Abdominal discomfort (pain/tenderness, bloating/distention)
	Passing gas more than in the past
	Belching/burping to relieve discomfort
	Acid reflux/heartburn
	Mouth sores, ulcers, and/or canker sores
	Psychological
	New or worsening depression or moodiness
	Change in personality/behavior
	Pulmonary
	Increased cough or wheezing, fluid in lungs, trouble breathing (choking spells)
	Hematological
	Anemia - Iron, Vitamin B12/folic acid deficiency
	Dermatological
	Dry skin or "scaling" skin, or loss of hair in eyebrows
	Very itchy rash (tiny blister-like with white surrounding redness)
	Immune/Autoimmune
	Enlarged thyroid
	Swollen, red, warm, and/or tender joints
	Alopecia (loss of hair on scalp and/or body)
	Other signs of other autoimmune diseases (for example: rheumatoid arthritis, Type 1 diabetes)
	Neurological
	New onset of seizures or change in seizures
	Development of mental "fog" (including forgetfulness or feeling "out of sorts")
	Increased difficulty in remaining in one
	Musculoskeletal
	Muscle cramps/aches

Basic Health Survey

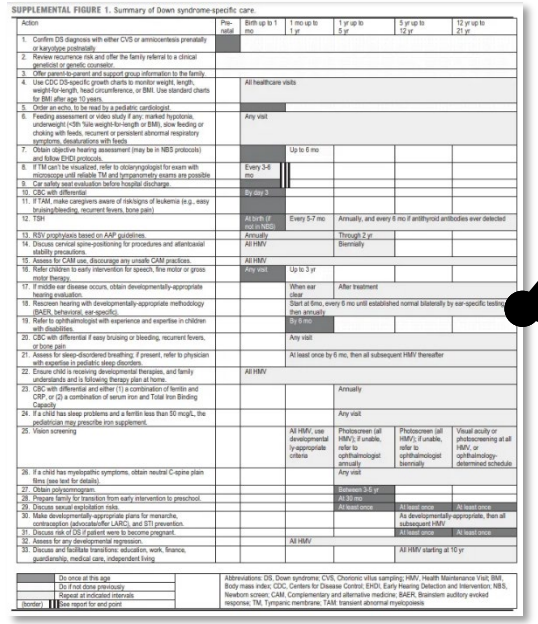
Age, sex,
major diagnoses

The medical care guidelines that apply to you!

Facilitating access to medical care guidelines

American Academy of Pediatrics (AAP):

- Prenatal period
- Birth to 1 month
- 1 month to 1 year
- 1 year to 5 years
- 5 years to 12 years
- 12 years to 21 years or older



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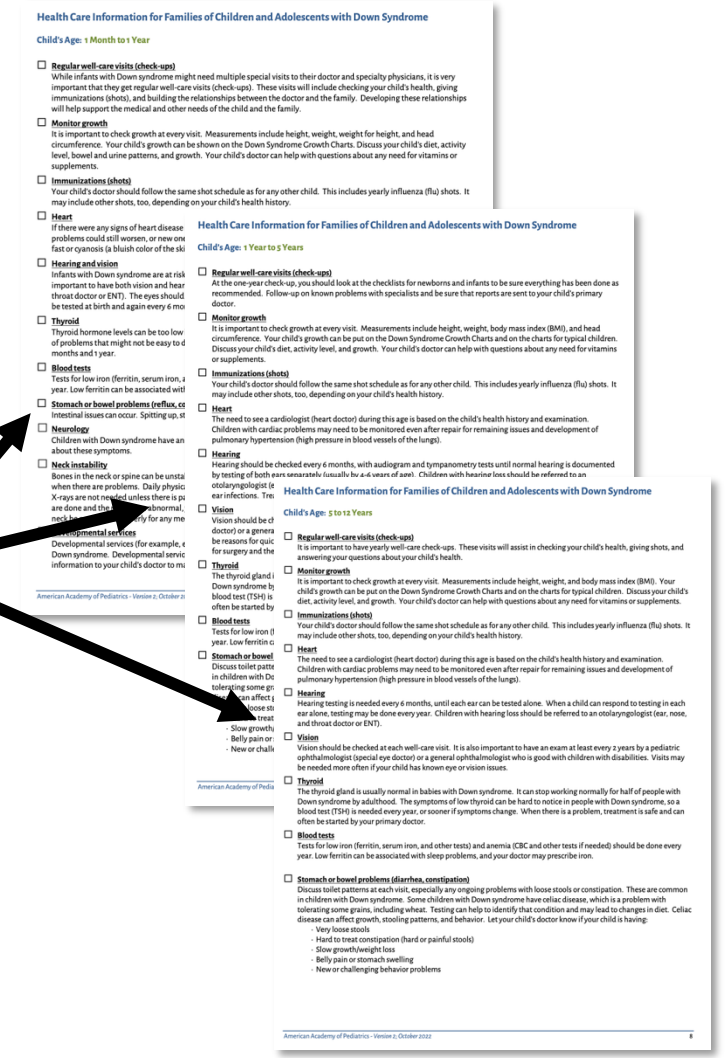
3. Offer parent-to-parent and support group information to the family.						
4. Use CDC DS-specific growth charts to monitor weight, length, weight-for-length, head circumference, or BMI. Use standard charts for BMI after age 10 years.		All healthcare visits				
5. Order an echo, to be read by a pediatric cardiologist.						
6. Feeding assessment or video study if any: marked hypotonia, underweight (<5th %ile weight-for-length or BMI), slow feeding or choking with feeds, recurrent or persistent abnormal respiratory symptoms, desaturations with feeds		Any visit				
7. Obtain objective hearing assessment (may be in NBS protocols) and follow EHDI protocols.			Up to 6 mo			
8. If TM can't be visualized, refer to otolaryngologist for exam with microscope until reliable TM and tympanometry exams are possible.		Every 3-6 mo				
9. Car safety seat evaluation before hospital discharge.						
10. CBC with differential		By day 3				
11. If TAM, make caregivers aware of risk/signs of leukemia (e.g., easy bruising/bleeding, recurrent fevers, bone pain)						
12. TSH		At birth (if not in NBS)	Every 5-7 mo	Annually, and every 6 mo if antithyroid antibodies ever detected		
13. RSV prophylaxis based on AAP guidelines.		Annually		Through 2 yr		
14. Discuss cervical spine-positioning for procedures and atlantoaxial stability precautions.		All HMV		Biennially		
15. Assess for CAM use, discourage any unsafe CAM practices.		All HMV				
16. Refer children to early intervention for speech, fine motor or gross motor therapy.		Any visit	Up to 3 yr			
17. If middle ear disease occurs, obtain developmentally-appropriate hearing evaluation.			When ear clear	After treatment		
18. Rescreen hearing with developmentally-appropriate methodology (BAER, behavioral, ear-specific).			Start at 6mo, every 6 mo until established normal bilaterally by ear-specific testing, then annually			
19. Refer to ophthalmologist with experience and expertise in children with disabilities.			By 6 mo			
20. CBC with differential if easy bruising or bleeding, recurrent fevers, or bone pain.			Any visit			
21. Assess for sleep-disordered breathing; if present, refer to physician with expertise in pediatric sleep disorders.			At least once by 6 mo, then all subsequent HMV thereafter			
22. Ensure child is receiving developmental therapies, and family understands and is following therapy plan at home.		All HMV				
23. CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and Total Iron Binding Capacity				Annually		
24. If a child has sleep problems and a ferritin less than 50 mcg/L, the pediatrician may prescribe iron supplement.				Any visit		
25. Vision screening			At HMV, use developmentally-appropriate criteria	Photoscreen (all HMV); if unable, refer to ophthalmologist	Photoscreen (all HMV); if unable, refer to ophthalmologist biennially	Visual acuity or photoscreening at all HMV, or ophthalmology-determined schedule
26. If a child has myelopathic symptoms, obtain neutral C-spine plain films (see text for details).				Any visit		
27. Obtain polysomnogram.				Between 3-5 yr		
28. Prepare family for transition from early intervention to preschool.				At 30 mo		
29. Discuss sexual exploitation risks.				At least once	At least once	At least once
30. Make developmentally-appropriate plans for menarche, contraception (advocate/offer LARC), and STI prevention.					As developmentally-appropriate, then all subsequent HMV	
31. Discuss risk of DS if patient were to become pregnant.					At least once	At least once
32. Assess for any developmental regression.			At HMV			
33. Discuss and facilitate transitions: education, work, finance, guardianship, medical care, independent living					At HMV starting at 16 yr	

Do once at this age
Do if not done previously
Repeat at indicated intervals
(border) See report for end point

Abbreviations: DS, Down syndrome; CVS, Chorionic villus sampling; HMV, Health Maintenance Visit; BMI, Body mass index; CDC, Centers for Disease Control; EHDI, Early Hearing Detection and Intervention; NBS, Newborn screen; CAM, Complementary and alternative medicine; BAER, Brainstem auditory evoked response; TM, Tympanic membrane; TAM, transient abnormal myelopoiesis



Health Care Information for Families of Children and Adolescents with Down Syndrome

Child's Age: 1 Month to 1 Year

- **Regular well-care visits (check-ups)**
  While infants with Down syndrome might need multiple special visits to their doctor and specialty physicians, it is very important that they get regular well-care visits (check-ups). These visits will include checking your child's health, giving immunizations (shots), and building the relationships between the doctor and the family. Developing these relationships will help support the medical and other needs of the child and the family.
- **Monitor growth**
  It is important to check growth at every visit. Measurements include height, weight, weight for height, and head circumference. Your child's growth can be shown on the Down Syndrome Growth Charts. Discuss your child's diet, activity level, bowel and urine patterns, and growth. Your child's doctor can help with questions about any need for vitamins or supplements.
- **Immunizations (shots)**
  Your child's doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child's health history.
- **Heart**
  If there were any signs of heart disease
- **Hearing and vision**
  Infants with Down syndrome are at risk
- **Thyroid**
  Thyroid hormone levels can be too low
- **Blood tests**
  Tests for low iron (ferritin, serum iron,
- **Stomach or bowel problems (reflux, co**
- **Neurology**
  Children with Down syndrome have an
- **Neck instability**
  Bones in the neck or spine can be unsta

American Academy of Pediatrics - Version 2, October 2

Health Care Information for Families of Children and Adolescents with Down Syndrome

Child's Age: 1 Year to 5 Years

- **Regular well-care visits (check-ups)**
  At the one-year check-up, you should look at the checklists for newborns and infants to be sure everything has been done as recommended. Follow-up on known problems with specialists and be sure that reports are sent to your child's primary doctor.
- **Monitor growth**
  It is important to check growth at every visit. Measurements include height, weight, body mass index (BMI), and head circumference. Your child's growth can be put on the Down Syndrome Growth Charts and on the charts for typical children. Discuss your child's diet, activity level, and growth. Your child's doctor can help with questions about any need for vitamins or supplements.
- **Blood tests**
  Tests for low iron (ferritin, serum iron,
- **Immunizations (shots)**
  Your child's doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child's health history.
- **Heart**
  The need to see a cardiologist (heart doctor) during this age is based on the child's health history and examination. Children with cardiac problems may need to be monitored even after repair for remaining issues and development of pulmonary hypertension (high pressure in blood vessels of the lungs).
- **Hearing**
  Hearing should be checked every 6 months, with audiogram and tympanometry tests until normal hearing is documented by testing of both ears separately (usually by 4-6 years of age). Children with hearing loss should be referred to an otolaryngologist (e
  ear infections. Tre
- **Vision**
  Vision should be c
  doctor) or a genera
- **Developmental services**
  Developmental services (for example,
  Down syndrome. Developmental servic
  information to your child's doctor to m

Health Care Information for Families of Children and Adolescents with Down Syndrome

Child's Age: 5 to 12 Years

- **Regular well-care visits (check-ups)**
  It is important to have yearly well-care check-ups. These visits will assist in checking your child's health, giving shots, and answering your questions about your child's health.
- **Monitor growth**
  It is important to check growth at every visit. Measurements include height, weight, and body mass index (BMI). Your child's growth can be put on the Down Syndrome Growth Charts and on the charts for typical children. Discuss your child's diet, activity level, and growth. Your child's doctor can help with questions about any need for vitamins or supplements.
- **Immunizations (shots)**
  Your child's doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child's health history.
- **Heart**
  The need to see a cardiologist (heart doctor) during this age is based on the child's health history and examination. Children with cardiac problems may need to be monitored even after repair for remaining issues and development of pulmonary hypertension (high pressure in blood vessels of the lungs).
- **Hearing**
  Hearing testing is needed every 6 months, until each ear can be tested alone. When a child can respond to testing in each ear alone, testing may be done every year. Children with hearing loss should be referred to an otolaryngologist (ear, nose, and throat doctor or ENT).
- **Vision**
  Vision should be checked at each well-care visit. It is also important to have an exam at least every 2 years by a pediatric ophthalmologist (special eye doctor) or a general ophthalmologist who is good with children with disabilities. Visits may be needed more often if your child has known eye or vision issues.
- **Thyroid**
  The thyroid gland is usually normal in babies with Down syndrome. It can stop working normally for half of people with Down syndrome by adulthood. The symptoms of low thyroid can be hard to notice in people with Down syndrome, so a blood test (TSH) is needed every year, or sooner if symptoms change. When there is a problem, treatment is safe and can often be started by your primary doctor.
- **Blood tests**
  Tests for low iron (ferritin, serum iron, and other tests) and anemia (CBC and other tests if needed) should be done every year. Low ferritin can be associated with sleep problems, and your doctor may prescribe iron.
- **Stomach or bowel problems (diarrhea, constipation)**
  Discuss toilet patterns at each visit, especially any ongoing problems with loose stools or constipation. These are common in children with Down syndrome. Some children with Down syndrome have celiac disease, which is a problem with tolerating some grains, including wheat. Testing can help to identify that condition and may lead to changes in diet. Celiac disease can affect growth, stooling patterns, and behavior. Let your child's doctor know if your child is having:
  - Very loose stools
  - Hard to treat constipation (hard or painful stools)
  - Slow growth/weight loss
  - Belly pain or stomach swelling
  - New or challenging behavior problems

American Academy of Pediatrics - Version 2, October 2022 8

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN®

Facilitating access to medical care guidelines

GLOBAL Adult Medical Care Guidelines:

- 21-29 years
- 30-39 years
- 40-49 years
- 50-59 years
- 60+ years

GLOBAL
DOWN SYNDROME FOUNDATION®

One family dashboard, multiple benefits

Surveys

Complete health surveys to provide researchers with valuable data

- [ ] Adolescence to Adult Transition Services
  Last Completed: Never
- [ ] Adulthood Survey
  Last Completed: Never
- [x] Basic Health Survey
  Last Completed: Sep 23, 2025
- [ ] Caregiver Depression Survey
  Last Completed: Never
- [ ] Caregiver Healthy Days
  Last Completed: Never
- [ ] Caregiver Stress Scale
  Last Completed: Never
- [ ] Celiac Disease Survey
  Last Completed: Never
- [ ] Confusion Hubbub and Order Scale Survey
  Last Completed: Never
- [x] Consent to Enroll in DS-Connect Registry
  Last Completed: Sep 8, 2025
- [ ] Development Survey
  Last Completed: Never

Community Data

Explore how your health history compares to that of the rest of the DS-Connect community

- Prenatal and Birth History
- Basic Health Survey

Healthcare Checklist

Use these checklists to better inform your health care provider discussions. These checklists are not intended to replace professional medical advice or services.

- Current - Checklist - Year 12 to Year 21
- Upcoming - Checklist - Year 21 to Year 29

Health History Snapshot

Create your own personal health history snaphot and further discussions with your health care provider.

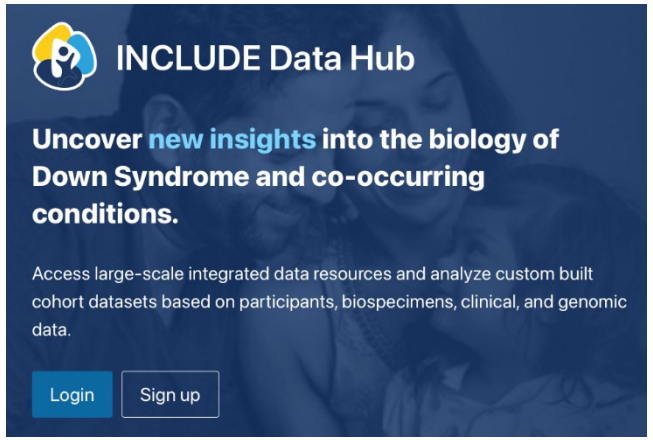
- Create Health History Snapshot

How is DS-Connect data shared?

Only **de-identified information** is shared with scientists.

De-identified information does not contain any of the so called ‘HIPAA identifiers’: no names, no dates, no account numbers, nothing that could be tied to a given individual.

De-identified data is shared through the INCLUDE Data Hub:



https://portal.includedcc.org/

A researcher portal supported by NIH

Other resources for families

Building community and supporting exchange of information

Upcoming Events

Empowerment Conference
Lafayette, CA
Click here for more information.

2026 New York City Buddy Walk
New York, NY
Click here for more information.

Down Syndrome Diagnosis Network Rockin' Mom™ Retreat
Phoenix, AZ
Click here for more information.

2026 St. Paul Step Up for Down Syndrome
St. Paul, MN
Click here for more information.

RMDSA Step Up for Down Syndrome Walk
Denver, CO
Click here for more information.

5th Annual Connect26 Las Vegas Down Syndrome Conference
Las Vegas, NV
Click here for more information.

Latest DS-Connect® News

Recording Available: May 2026 Webinar Hosted by DS-Connect

June 11, 2026

We appreciate your interest in the recent DS-Connect webinar featuring Dr. Meghan O'Neill, and members of the DS-...

Recording Available: March Webinar Hosted by DS-Connect

March 31, 2026

We appreciate your interest in the recent DS-Connect webinar featuring Dr. Ruth Brown, and members of the DS-Connect...

DS-Connect Team Joins the 2026 Brighter Futures Conference

March 30, 2026

The DS-Connect®: The Down Syndrome Registry team participated in the 2026 Brighter Futures: A National Conference on...

Recording Available: December Webinar Hosted by DS-Connect

December 22, 2025

We appreciate your interest in the recent DS-Connect webinar featuring Dr. Daniel Combs, and members of the DS-Connect...

DS-Connect Team Attends 2025 Atlanta Buddy Walk®

October 6, 2025

The DS-Connect®: The Down Syndrome Registry team participated in the 2025 Atlanta Buddy Walk® in Marietta, GA....

Recording Available: September Webinar Hosted by DS-Connect

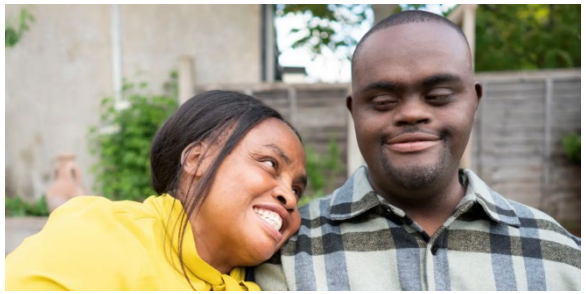
September 25, 2025

We appreciate your interest in the recent DS-Connect webinar featuring Dr. Anna Esbensen, Dr. Mike Raffi, and members...

Other resources for families

Large collection of curated resources in partnership with other organizations

Resources



- **About Down Syndrome**
- **Down Syndrome Clinics in the United States**
- **New and Expectant Parents**
- **Newborn to School Age**
- **Teenagers to Adults**
- **Siblings**
- **Learn About Down Syndrome Research**
- **Resources for Health Care Providers**

About Down Syndrome

- INCLUDE Project Down Syndrome Research Plan (NIH)
- Language Guidelines (NDSS)
- Medical Resources (NDSC)
- Facts and FAQ About Down Syndrome (GLOBAL)
- GLOBAL Webinars (GLOBAL)
- Centers for Disease Control and Prevention (CDC) Down syndrome page
- What is Down syndrome? (NDSC)
- Words Can Hurt (GLOBAL)
- An Overview of Down Syndrome (NDSS)
- Ages and Stages (NDSC)
- Mosaic Down Syndrome (IMDSA)
- Down Syndrome Misconceptions vs. Reality (GLOBAL)

A central hub for approved research studies

Studies are approved by a Research Review Committee (RRC) that includes scientists, physicians, and community members

Connect Your Research with the Down Syndrome Community

Promote recruitment for your study through DS-Connect

Did you know that 75% of families with a loved one with Down syndrome are interested in participating in research? A main goal of DS-Connect is to connect families who want to participate in research with the research community. You can request to share your study information with DS-Connect participants here.

Get started

RRC review and approval

Studies which are Now Recruiting

Down Syndrome Health Measure (DSHM) Survey

Stephanie Santoro, MD
Virtual
0 to 21 years old

The Down Syndrome Health Measure (DSHM) is a new, short survey that was created specifically with people with Down syndrome in mind, and asks questions about aspects of overall health. Now that this final measure is available, we're interested in collecting feedback on the survey from caregivers of individuals with Down syndrome age 0 to 21.

Workshop to Help Reduce Stress for Caregivers: CarePRO Virtual-DS/ID

David Coon, PhD
Virtual
18 years and older

Family caregivers of adults living with Down syndrome or another intellectual disability who are experiencing memory changes are invited to participate in CarePRO Virtual-DS/ID, a free 5-week online workshop designed to reduce distress and improve quality of life.

Treatment of Obstructive Sleep Apnea with Personalized Surgery in Children with Down Syndrome (TOPS-DS)

Derek Lam, MD, MPH
Multiple locations in the United States
2 to 17 years old

The purpose of this study is to test how well personalized surgical treatment helps children with Down syndrome (DS) who have obstructive sleep apnea (OSA).

Spotlight Study

Scan for recruiting studies:

Healthy Aging and Neurodevelopment in Down Syndrome (HANDS)

Jennifer Bruno, PhD

Stanford, CA

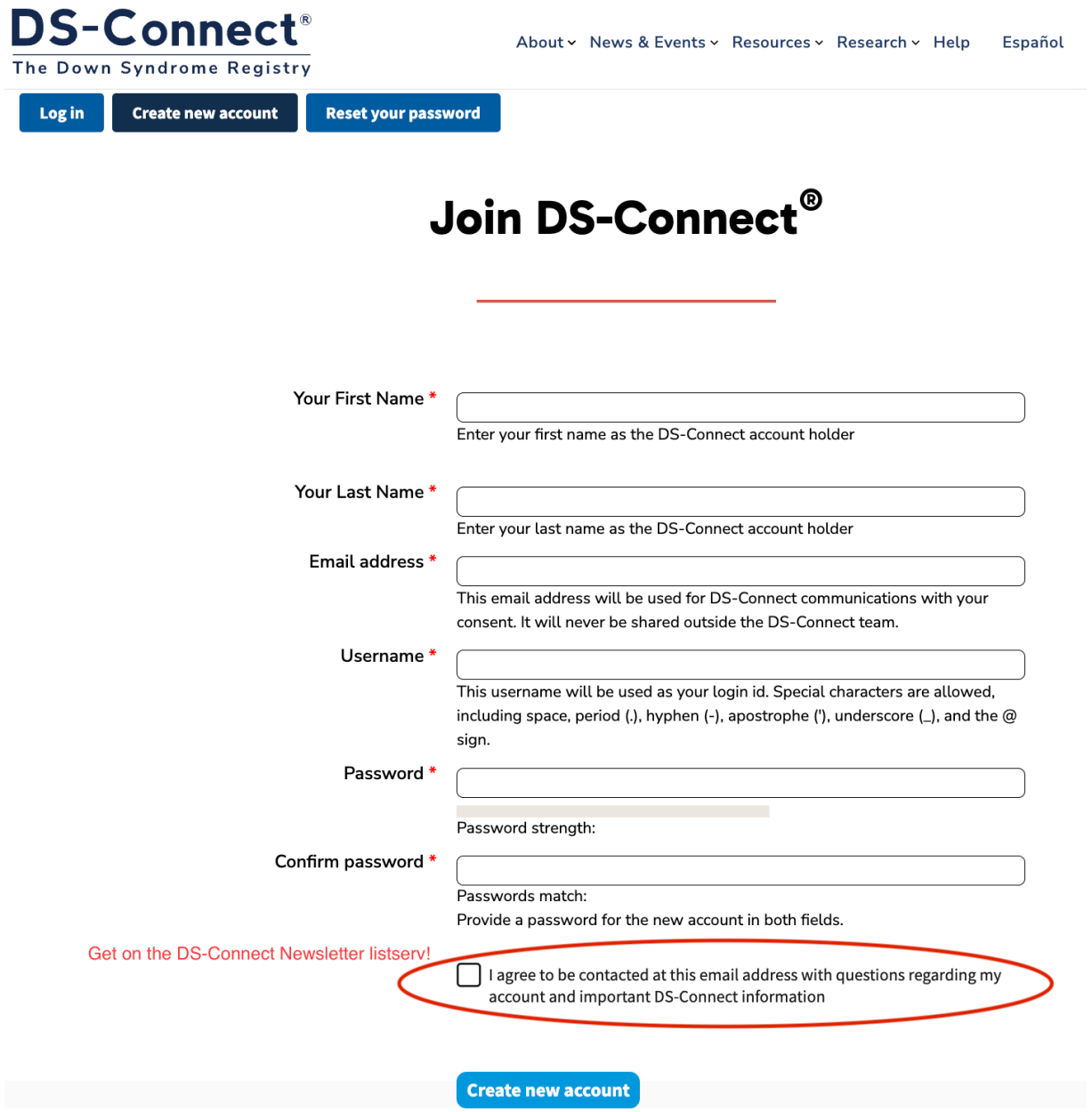
8 to 65 years old

This study investigates brain changes related to development in children and adults with Down syndrome.

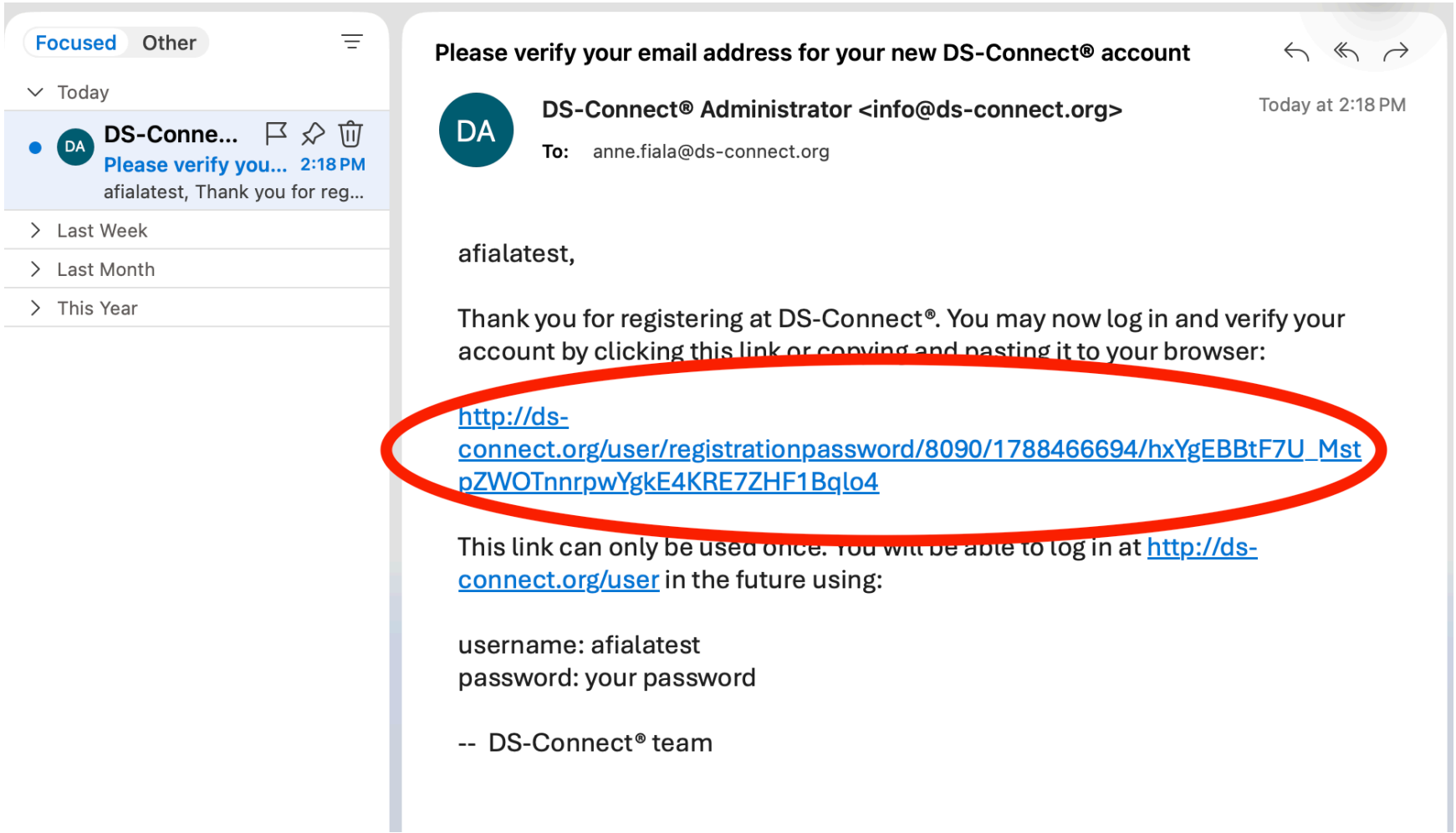
Ready to Get DS-Connected?

Register Now!

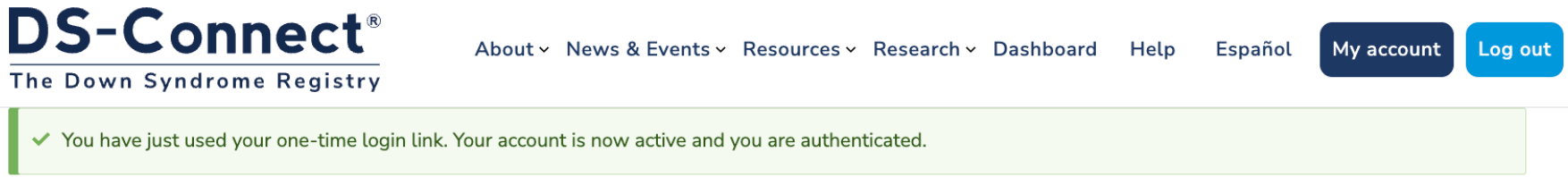
Create an account



Verify your email address



First few steps...



1

Consent

DS-Connect®
The Down Syndrome Registry

About News & Events Resources Research Dashboard Help Español My account Log out

Consent to Enroll in DS-Connect Registry

0%

PI Name: Dr. Joaquin Espinosa

COMIRB#: 24-0396

In this form, we use the words "you" and "your." If you are reading this form and deciding for someone else, the words "you" and "your" refer to that person, not to you.

You are being asked to be in this research study because you have Down syndrome.

If you join the study, you will be asked to enter an online registry and complete a series of questions about yourself and about your health/diagnosis. We will reach out once a year and ask you to update your health information in the registry.

This study is designed to learn more about the demographics, health, and other medical information of people living with Down syndrome.

Possible discomforts or risks include anxiety, fatigue, boredom or frustration during the completion of the questionnaires. There may be risks the researchers have not thought of.

This study is not designed to benefit you directly.

Every effort will be made to protect your privacy and confidentiality. Your identifiable information will be securely stored and only accessible to approved research personnel.

Data sharing is crucial to increasing the speed of Down syndrome research.

We share your de-identified data following National Institutes of Health (NIH) requirements.

2

Demographic Information

DS-Connect®
The Down Syndrome Registry

About News & Events Resources Research Dashboard Help Español My account Log out

Participant Information

Your Relationship to Person with Down Syndrome *

- Select a value -

What is your relationship to the DS-Connect participant?

Participant First Name *

The first name of the person with Down syndrome aka the "DS-Connect participant"

Participant Middle Name

The middle name of the person with Down syndrome aka the "DS-Connect participant."

Participant Last Name *

The last name of the person with Down syndrome aka the "DS-Connect participant"

Participant Sex at Birth *

- Select a value -

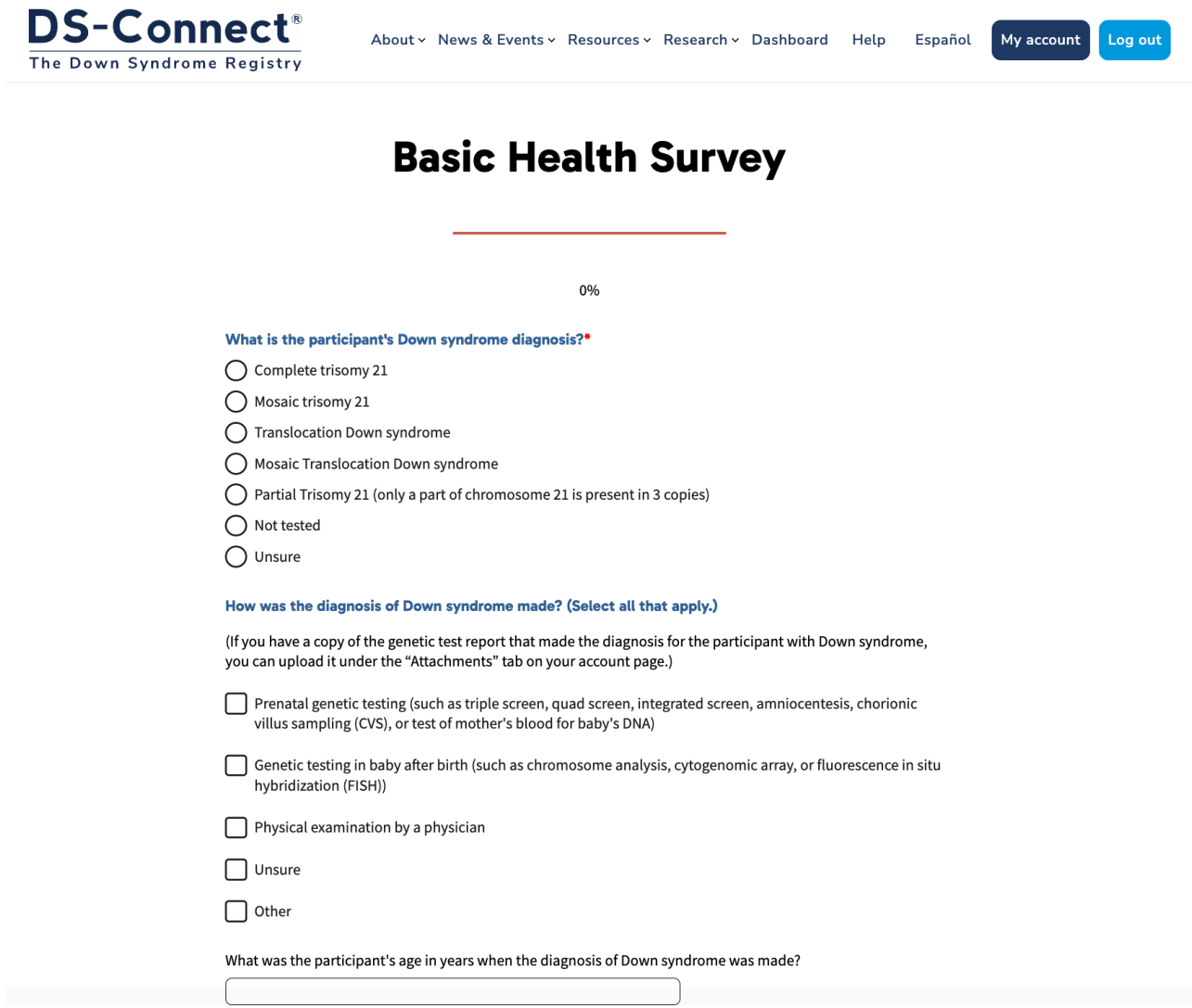
Participant Birthdate *

mm/dd/yyyy

Date of birth of the participant

3

Basic Health Survey



4

Dashboard!

Welcome, TestAnne

Thank you for participating in the DS-Connect® community!

Complete your Health surveys shown on the list below. The ability to see your previously entered information will become available again in 2025. You will receive updates on the new DS-Connect® via email.

Surveys

Complete health surveys to provide researchers with valuable data

- [ ] Adulthood Survey
  Last Completed: Never
- [x] Basic Health Survey
  Last Completed: Sep 4, 2025
- [x] Consent to Enroll in DS-Connect Registry
  Last Completed: Feb 10, 2025
- [ ] Development Survey
  Last Completed: Never
- [ ] Gastrointestinal Survey
  Last Completed: Never
- [ ] Leukemia Survey
  Last Completed: Never
- [ ] Prenatal and Birth History
  Last Completed: Never
- [ ] Sibling Survey
  Last Completed: Never
- [ ] Sleep Survey
  Last Completed: Never

Community Data

Explore how your health history compares to that of the rest of the DS-Connect community

- Basic Health Survey

Healthcare Checklist

Use these checklists to better inform your health care provider discussions. These checklists are not intended to replace professional medical advice or services.

- Current - Checklist - Year 30 to Year 39

Health History Snapshot

Create your own personal health history snaphot and further discussions with your health care provider.

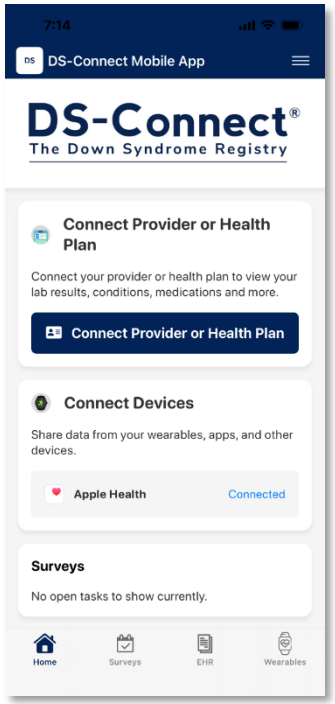
- Create Health History Snapshot

Upcoming activities

Social media platforms

Coming soon,
be ready to follow us!

DS-Connect mobile app



Stay tuned,
DS-Connect at your thumbtips...

DS-Connect podcast

The Science of
Down Syndrome

Renowned researchers
explaining scientific discoveries
in lay terms

A team effort!

Team at the Crnic Institute / University of Colorado:
Joaquin Espinosa
Angela Rachubinski
Matthew Galbraith
Anne Fiala
Lyndy Bush
Chelsea Donohoe
Charlotte Plotzke
Chad McGimpsey
Kyle Bartsch
Daniela Leon-Alvarez
Samantha Baker
Maci Cameron
Caroline Lucas

Team at NIH:
Sujata Bardhan
Melissa Parisi
Linda Garcia
Valerie Cotton
Rebecca Rosen
Artisha Wright
Renee Goodman

Research Review Committee:
Niki Baumer
Emily Chesnut
Anna Esbensen
Debbie Fidler
Sharon Krinsky-McHale
Linda Roan
Traci Rosser
Stephanie Sherman

DS-Connect: The Down Syndrome Registry ("DS-Connect") is supported by the National Institutes of Health under Project Number OT2HD117033 administered by the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development.

Thank you to the participants and their families!