Practice Changing Articles from the Past Year

Dr. Peter Bulova, Professor of Medicine

Dr. Kishore Vellody, Professor of Pediatrics

Disclosures

- Neither Dr. Vellody or Dr. Bulova have any financial interests or relationships that could potentially bias the presentation
- Both Dr. Vellody and Dr. Bulova wish that JAK inhibitors worked for male pattern baldness

Our Goals

- To bring our audience up to date on literature that is:
  - Relevant
  - Practice changing
  - Rigorous
  - Key take home points
  - Variety of ages/topics

Pitt | School of Medicine

Our Process

- Recommendations from colleagues
- Pub med review of articles with title or abstract MESH term “Down syndrome”, “Trisomy 21” or “Down’s syndrome” since 6/1/2024
  - 659 Articles
  - Reviewed for clinical applicability: 659 articles
- Presented as different chapters in the life of a person with Ds
  - Given time constraints, will be only a surface-level, high-yield overview of the studies discussed

Pitt | School of Medicine

Chapter 1

Malik and Jasmine are soon-to-be first-time parents who you have been asked to meet as their baby has been prenatally diagnosed with Down syndrome. During the course of your conversation, you discover them both emotionally upset about how they were initially informed about the diagnosis of Down syndrome. They ask if this is something other parents have experienced.

American Journal of Medical Genetics Part A

WILEY

AMERICAN JOURNAL OF medical genetics PART A

ORIGINAL ARTICLE

Prenatal Care of Parents Who Continued Pregnancies With Down Syndrome, 2003–2022

Jonathan M. Artal[1,2] | Lindsey Randall[3,4] | Sabina Rubeck[5] | Megan Allyse[6] | Marsha Michie[5] | Kirsten A. Riggan[6] | Stephanie Meredith[7] | Brian G. Skotko[2,8]

[1]Stanford Law School, Stanford University, Stanford, California, USA | [2]Down Syndrome Program, Division of Medical Genetics and Metabolism, Department of Pediatrics, Massachusetts General Hospital, Boston, Massachusetts, USA | [3]The Mayo Clinic Alix School of Medicine, Rochester, Minnesota, USA | [4]Mayo Clinic Graduate School of Biomedical Sciences, Rochester, Minnesota, USA | [5]Case Western Reserve University School of Medicine, Department of Bioethics, Cleveland, Ohio, USA | [6]Mayo Clinic, Biomedical Ethics Research Program, Rochester, Minnesota, USA | [7]Human Development Institute, University of Kentucky, Lexington, Kentucky, USA | [8]Department of Pediatrics, Harvard Medical School, Boston, Massachusetts, USA

Correspondence: Brian G. Skotko (bskotko@mgh.harvard.edu)

Received: 20 October 2024 | **Revised:** 20 December 2024 | **Accepted:** 25 December 2024

Background

- Same group studied postnatal diagnosis comparison to 20 years earlier (which we looked at last year)
  - Parent experiences in receiving a postnatal diagnosis of Ds continues to be poor, maybe even worse
- This study looked at prenatal diagnosis experience for families
  - Using a similar survey as was used in 2003 study

Pitt | School of Medicine

Methods

- RedCAP survey, 1-7 Likert scales (1=strong disagreement, 4=neutral, 7=strong agreement)
- Distributed by 4 Ds support organizations (CA, MA, NC, CO) to parents via e-mail
  - 60 respondents who received prenatal diagnoses (avg birth year 2014)
  - 93% mothers, 87.7% white, 41% Catholic faith

Pitt | School of Medicine

TABLE 5 | Comparison of parents who received amniocentesis, CVS, and/or prenatal cell-free DNA screening, 2003 and 2022.[a]

	2003 mean (SD) $N=137$	2022 mean (SD) $N=60$	p for change
Before the prenatal test, I already had a good idea about what DS was	4.2 (2.2)	3.9 (2.1)	0.36
After I received the results			
I felt encouraged by my physician to terminate the pregnancy	3.0 (2.2)	3.8 (2.4)	**0.02**
I felt encouraged by my physician to continue the pregnancy	3.6 (2.0)	3.2 (1.9)	0.18
I felt scared	6.3 (1.3)	5.7 (1.7)	**0.01**
I felt anxious	6.1 (1.5)	6.1 (1.3)	0.88
I experienced suicidal thoughts	1.5 (1.4)	1.3 (1.1)	0.28
I felt positive	3.0 (1.8)	3.2 (1.9)	0.49
I felt my physician gave me enough up-to-date printed or digital material on DS	2.7 (2.1)	2.6 (2.3)	0.77

[a]Pooling parents who received amniocentesis, CVS, and/or prenatal cell-free DNA screening. Parents indicated their agreement with statements on 1–7 Likert scales: "1" denoted strong disagreement, "4" a neutral response, and "7" strong agreement. Bolded p values denote statistical significance.

Conclusion

- Compared to 2003 survey, physicians continue to deliver diagnosis with pity, suggestion of termination, lack of accurate information on Ds
- Legislative changes and educational/trainee interventions are not mandatory nor adequately funded so impact is diminished
- The medical community continues to have a long way to go in how to deliver the diagnosis

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas / Hilton Anatole
Pitt School of Medicine

Received: 14 December 2023 | Revised: 13 June 2024 | Accepted: 28 June 2024

DOI: 10.1002/jgc4.1948

ORIGINAL ARTICLE

National Society of Genetic Counselors | WILEY

Parent-reported genetic counselor adherence to the NSGC practice resource for communicating a potential prenatal diagnosis: Impact on the Down syndrome diagnosis experience

Maryam R. Ijaz[1] | Angela M. Trepanier[1] | Harold L. Kleinert[2] | Sierra M. Weiss[2] | Stephanie H. Meredith[2]

Background

- 2011 - National Society of Genetic Counselors (NSGC) published practice resources on communicating prenatal and postnatal diagnosis of Ds
- Impact of practice resources had not yet been studied

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole

Pitt School of Medicine

Methods

- Survey distributed by 12 local DS organization and 1 national organization (DSDN)
  - Questions regarding whether the genetic counselor followed the published 2011 practice recommendations
- 242 participants (68% white)
- Open ended questions scored 1-5 (1=completely negative, 3=neutral, 5=completely positive)

Pitt School of Medicine

Conclusions

- Higher adherence to NSGC practice resources = enhanced prenatal Ds diagnostic experience for parents
- Conclusions
  - Standardize provision of care, including disability cultural competency training
  - Provide patient education resources to families
  - Update the NSGC recommendations (last in 2011) and broadly disseminate to practitioners

Pitt School of Medicine

Chapter 2

Baby Imani is born and is now 5 months old. During the clinic visit, you hear her having inspiratory squeaking sounds with substernal and subcostal retractions on inspiration with pectus excavatum (caved in chest appearance). You suspect laryngomalacia and possible sleep apnea. Her parents are aware that a sleep study is recommended between ages 3-4 years old, but they're worried if untreated sleep apnea may impact Imani's development.

Early detection and treatment of obstructive sleep apnoea in infants with Down syndrome: a prospective, non-randomised, controlled, interventional study

Brigitte Fauroux,[a,b,g,*] Silvia Sacco,[c] Vincent Couloigner,[d] Alessandro Amaddeo,[a,b,e] Aimé Ravel,[c] Emmanuelle Prioux,[c] Jeanne Toulas,[c] Cécile Cieuta-Walti,[c] Hervé Walti,[c] Romain Luscan,[d] Ségolène Falquero,[c] Manon Clert,[c] Marie-Anne Caillaud,[c] Livio De Sanctis,[a] Sonia Khirani,[a,b,f] Isabelle Marey,[c] and Clotilde Mircher[c,g]

www.thelancet.com Vol 45 October, 2024

Background

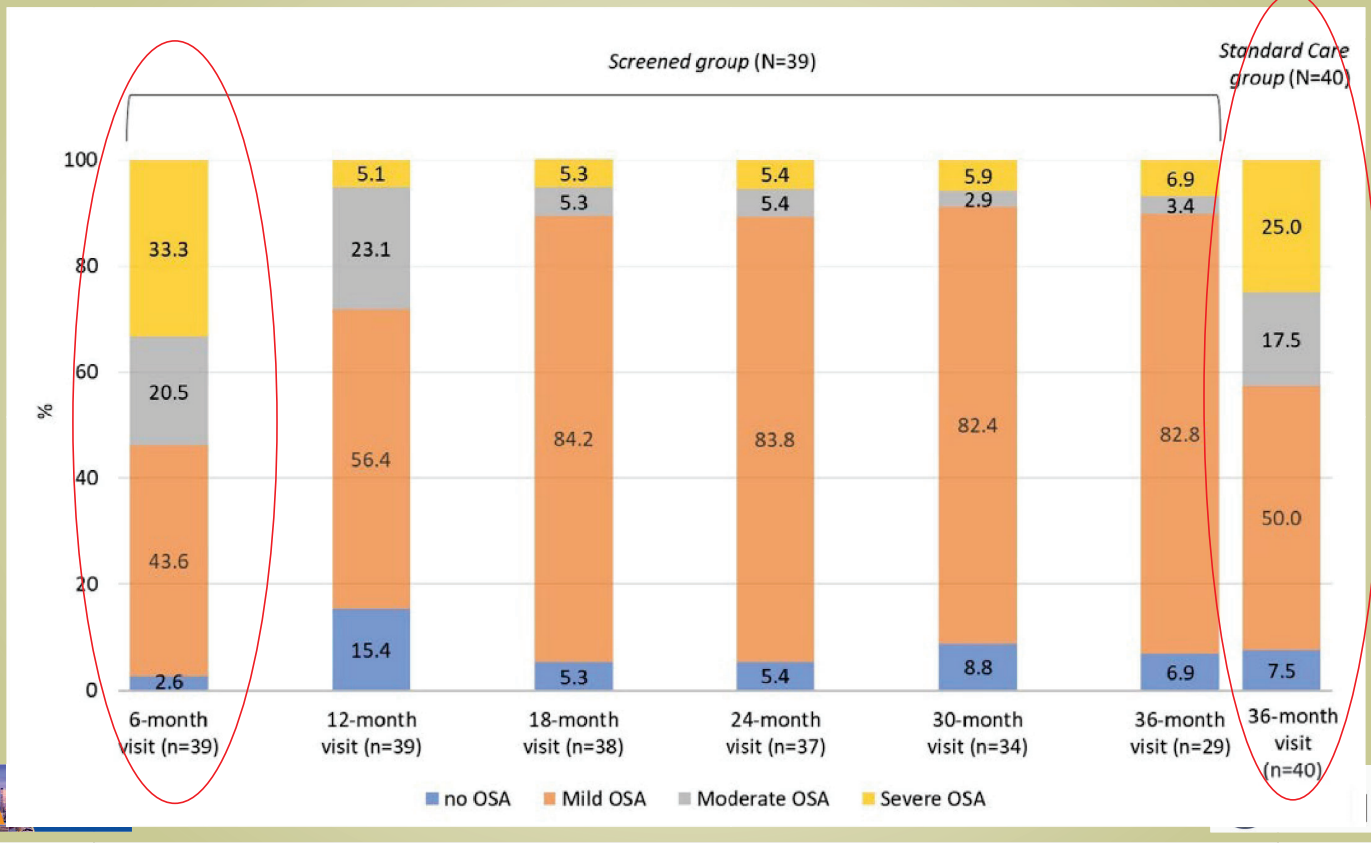
- Obstructive sleep apnea (OSA) is common in Ds
- OSA <1 year old is often severe OSA which can lead to neurocognitive dysfunction, behavioral issues
- Current guideline recommendation is for sleep study between 3-4 years old
  - Should sleep studies be done earlier in life?

Pitt School of Medicine

Methods

- Prospective interventional, non-randomized, single center study
  - 39 infants with Ds with home PSG every 6 months starting at 6 months old (29 completed all the studies, 34 completed 5 out of 6)
  - 40 infants with standard care of home PSG at 36 months old
    - Any PSGs or upper airway surgeries done before 36 months were retrospectively analyzed
- If OSA, ENT involved to consider
  - Drug induced sleep endoscopy (DISE) followed by upper airway surgery
  - Continuous positive airway pressure (CPAP)
- Endpoint was total score on Griffiths Scales of Child Development, 3rd Ed

Pitt School of Medicine



AHI at 36 months

Parameter [units]/Statistics	Screened Group N = 29	Standard Care Group N = 40
Age [months]		
Median	37.0	37.0
Q1; Q3	37.0; 39.0	36.0; 37.0
AHI [events/h]		
Median	1.0	4.0
Q1; Q3	1.0; 3.0	1.5; 9.0
<1 events/h, n (%) - no OSA	2 (6.9)	3 (7.5)
≥1 to ≤5 events/h, n (%) – mild OSA	24 (82.8)	20 (50.0)
>5 to ≤10 events/h, n (%) – moderate OSA	1 (3.4)	7 (17.5)
>10 events/h, n (%) – severe OSA	2 (6.9)	10 (25.0)
	14/29 had upper airway surgery before 36 mo (Avg 16 mo)	1/40 had upper airway surgery before 36 mo (no PSG)

Parameter/ Statistics[b]	Screened Group N = 34	Standard Care Group N = 40	p-value[a]
Global QD			
Median	55.4	50.7	
Q1; Q3	(52.6; 60.0)	(45.1; 56.3)	
Difference (SE)	4.1 (1.6)		
95% CI	1.3; 7.6		0.009

Conclusions

- OSA is highly prevalent in children with Ds <3 years old
- Surgical management of OSA earlier MAY improve neurodevelopmental outcomes
- Risks of anesthesia/surgery must be weighed against potential benefit
- Longitudinal controlled study with longer term neuropsychological follow-up would be helpful

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole
Pitt School of Medicine

Chapter 3

During your discussions about OSA with Imani's parents, they raised concerns that she was not developing as quickly as expected according to the "baby book" that they were given. Being the young doctor that you are, you wonder what she means by "book" as you've not seen one of those in a while. However, you look into some guidance on developmental expectations that you could provide.

Developmental Milestones for Children With Down Syndrome

Nicole Baumer, MD, MEd,[a,b,c] Rafael DePillis, BS,[a] Katherine Pawlowski, BA,[a] Bo Zhang, PhD,[b,c] Maitreyi Mazumdar, MD, MPH[b,c,d]

PEDIATRICS Volume 154, number 4, October 2024:e2023065402

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole

Pitt School of Medicine

Background

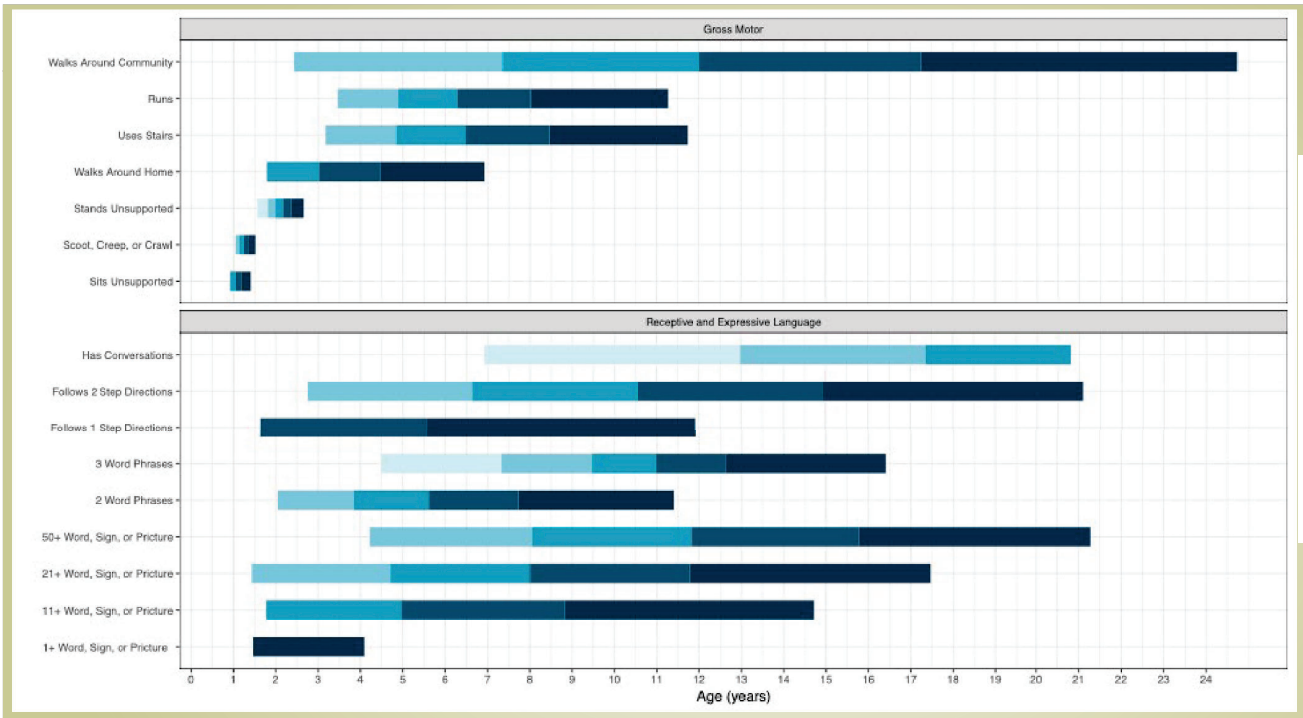
- Developmental delays are common, yet variable, in children with Ds
- Developmental milestones are available for the general population but not as readily available specifically to Ds

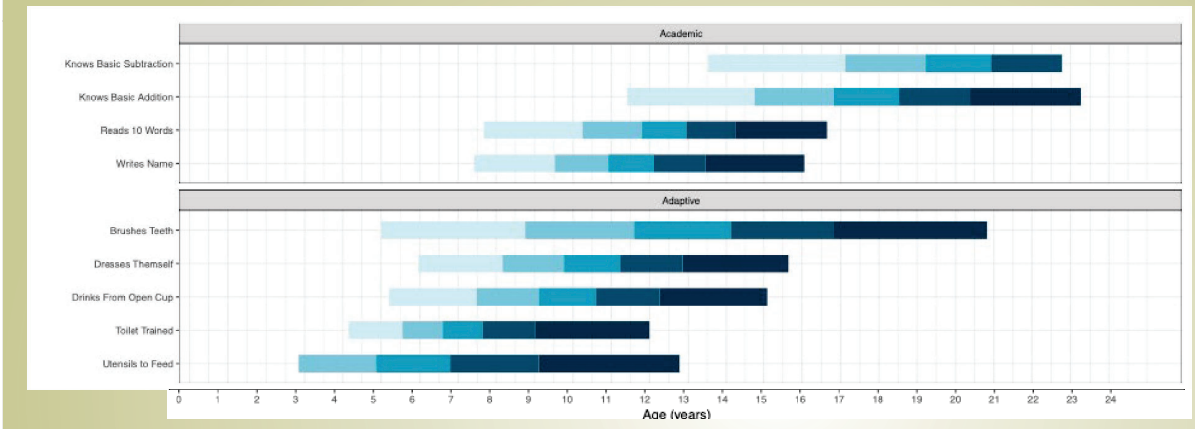
Methods

- Single center, 842 patients, 2 mo - 24 yrs, 63.7% White
- Caregiver and PT reports, but not standardized assessment

TABLE 1 Data Sources and Example Items

Data Source	Source	Example Item	Item Responses	Responses Operationalized as Achievement	Collapsed Responses From Different Sources?
Developmental Intake	Caregiver	Reads 10 words	Never, sometimes, usually	Usually	No
		Is physically able to walk around the community independently	Never, sometimes, usually	Usually	Yes: responses from Developmental Intake and PT Clinical Form are both included
		Engages in pretend play	Never, sometimes, usually	Usually	Yes: responses from Developmental Intake and ND-PROM are both included
ND-PROM	Caregiver	Engages in pretend play	Never, rarely, sometimes, often, always	Often, always	Yes: responses from Developmental Intake and ND-PROM are both included
		Toilet trained during the day	Never, rarely, sometimes, often, always	Often, always	No
Physical Therapy (PT) Clinical Form	Clinician (physical therapist)	Sits unsupported for at least 10 min	Never, sometimes, usually, observed in clinic	Usually, observed in clinic	No
		Walks 2 or more blocks without rest or physical support (travel around community)	Never, sometimes, usually, observed in clinic	Usually, observed in clinic	Yes: responses from Developmental Intake and PT Clinical Form are both included


Gross Motor
Walks Around Community
Runs
Uses Stairs
Walks Around Home
Stands Unsupported
Scoot, Creep, or Crawl
Sits Unsupported
Receptive and Expressive Language
Has Conversations
Follows 2 Step Directions
Follows 1 Step Directions
3 Word Phrases
2 Word Phrases
50+ Word, Sign, or Picture
21+ Word, Sign, or Picture
11+ Word, Sign, or Picture
1+ Word, Sign, or Picture
0 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24
Age (years)
Probability
15% – 30%
30% – 45%
45% – 60%
60% – 75%
75% – 90%


Academic
Knows Basic Subtraction
Knows Basic Addition
Reads 10 Words
Writes Name
Adaptive
Brushes Teeth
Dresses Themself
Drinks From Open Cup
Toilet Trained
Utensils to Feed
0 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24
Age (years)
Probability
15% – 30%
30% – 45%
45% – 60%
60% – 75%
75% – 90%

Conclusions

- Wide variability in when children with Ds achieve milestones
- Milestones achieved in similar order as general population, just at a slower rate
- Several letters to editor about
  - Possibly skewed population with tertiary medical center
  - Including medically complex patients
  - Including co-occurring conditions like autism
  - Setting lower expectations for other children with Ds
  - May lead to less therapy being provided at appropriate times if felt to be in "normal range"

Pitt School of Medicine

Chapter 4

Imani's laryngomalacia was addressed in early childhood but a sleep study at 3 years old showed an AHI of 4.5 events/hr. She underwent adenotonsillectomy with clinical improvement. Unfortunately, snoring and restless sleep was again noted in her early teenage years. A repeat sleep study showed an AHI of 15 events/hr. She does not tolerate CPAP despite maximal efforts. You refer them to ENT for consideration of hypoglossal nerve stimulator placement. Her parents are impressed with the studies showing initial clinical improvement, but they wonder if the impact is longer lasting.

Pitt School of Medicine

Upper Airway Stimulation for Children and Adolescents with Down Syndrome: Long-Term Follow-Up

Danielle R. Larrow, MD; Kathryn S. Marcus, MD; Kevin Gipson, MD; Brian G. Skotko, MD; Cristina M. Baldassari, MD; David F. Smith, MD; Christine H. Heubi, MD; Allison Tobey, MD; Raol Nikhila, MD; Mark Vangel, PhD; Christopher J. Hartnick, MD

- Data for 33/42 adolescent patients followed at 5 centers
- No reported adverse effects from the device long-term
- 4/33 required HNS battery replacement (within average of 4 years – expected 8-12 years battery life)

TABLE II.
Polysomnogram Outcomes.

Characteristic	Mean (SD) [95% CI]	Range	% AHI Less Than 10	% AHI Less Than 5	% AHI Less Than 2
AHI					
Timepoint 1	23.8 (10.0)	10.0–48.8	0	0	0
Timepoint 2	11.1 (14.3)	0.6–61.1	72.7	36.4	9.1
Timepoint 3	8.1 (8.4)	0.0–45.0	78.8	39.4	18.2
Change in AHI					
Timepoint 1 to 2	−12.7 (13.4) [−8.1 to −17.3]	−41.4 to 18.3			
Timepoint 1 to 3	−15.7 (13.1) [−11.2 to −20.2]	−44.9 to 27.6			
% Reduction in AHI					
Timepoint 1 to 2	−51.1 (53.5) [−32.8 to −69.3]	−97.5 to 161.0			
Timepoint 1 to 3	−59.6 (51.8) [−42.0 to −77.3]	−100.0 to 158.6			

Timepoint 1 = baseline, timepoint 2 = 1 year post-implantation, timepoint 3 = long-term follow-up.

Avg 4y after Timepoint 1

Conclusions

- Pediatric hypoglossal nerve stimulator therapy appears to be effective, including several years after placement
- Increased need for earlier battery replacement than previously reported
  - ?Increased adherence rate for children, children with Ds in particular?

Pitt | School of Medicine

Chapter 5

Imani is 23 years old and her family has noticed a significant change in her desire to interact with friends and family. She has stopped speaking and seems agitated. She was previously fairly independent in taking care of herself, but she now requires complete help with tasks she used to be able to do on her own.

Pitt | School of Medicine

www.nature.com/scientificreports

scientific reports

OPEN

Diagnostic abnormalities, disease severity and immunotherapy responsiveness in individuals with Down syndrome regression disorder

Jonathan D. Santoro[1,2]✉, Saba Jafarpour[1], Laura Keehan[3], Mellad M. Khoshnood[1,2], Lilia Kazerooni[1], Natalie K. Boyd[1], Benjamin N. Vogel[1], Lina Nguyen[1], Melanie Manning[3,4], Deepti Nagesh[1,2], Noemi A. Spinazzi[5], Aaron D. Besterman[6], Eileen A. Quinn[7] & Michael S. Rafii[2,8]

Pitt School of Medicine

What is Down Syndrome Regression Disorder (DSRD)?

- Acute/subacute decline in functioning in youth with Down syndrome
- Key features: Catatonia, mutism, loss of ADLs, agitation, OCD behavior
- Typical onset: age 10–30
- Often triggered by infection or life changes

Pitt School of Medicine

Study Overview

Retrospective, multi-center study

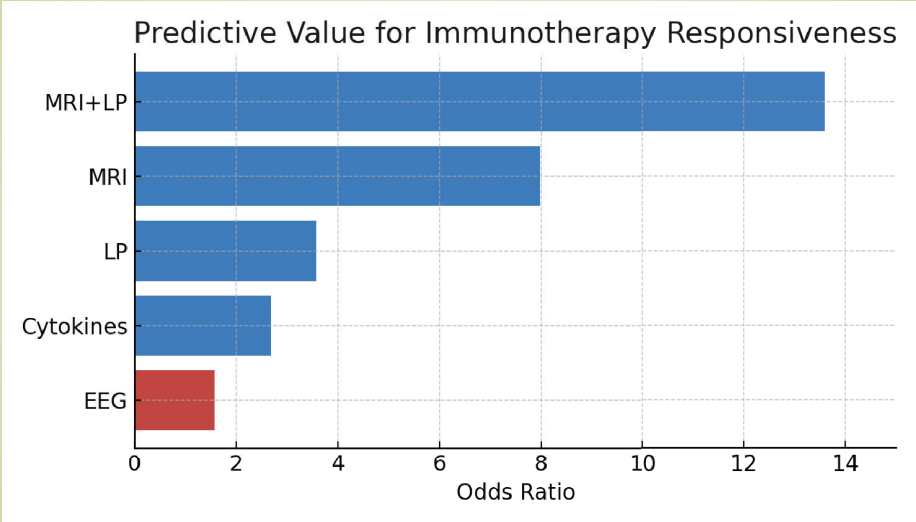
164 patients with confirmed DSRD (age 10–30)

Evaluated labs, MRI, EEG, CSF for abnormalities

Tracked disease severity and immunotherapy response

Who will benefit most from treatment?

Diagnostic Predictors of Treatment Response



Key Clinical Takeaways

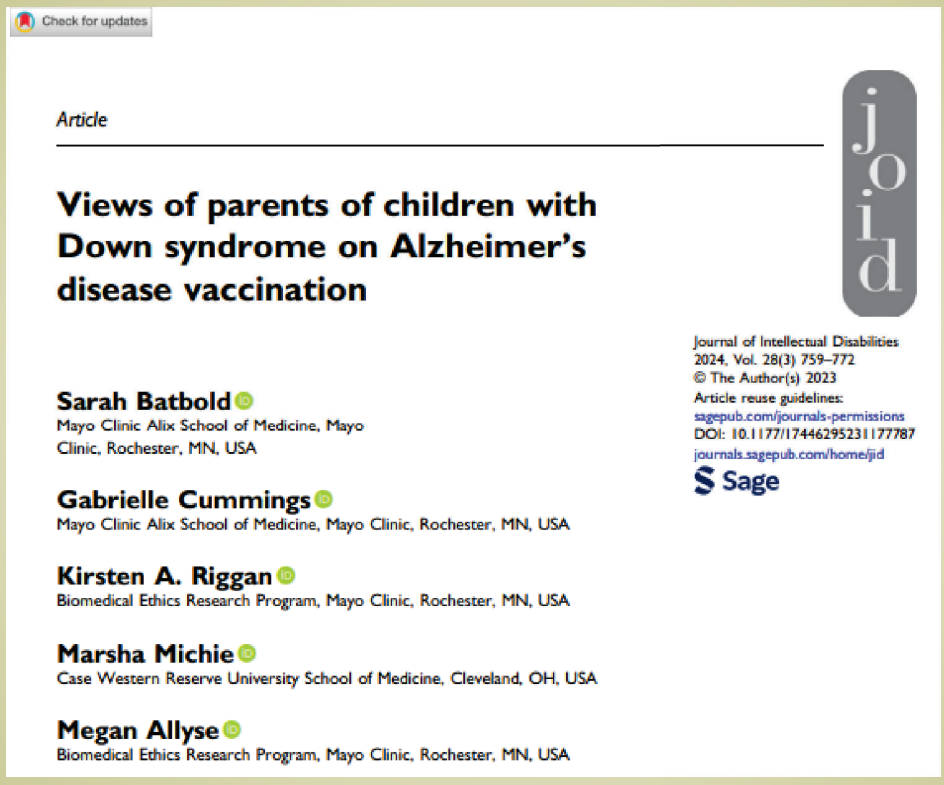
- MRI and LP are the most predictive tests for response to treatment
- EEG, ANA, and routine labs not strongly predictive
- Early testing (within 6 months of symptom onset) increases diagnostic yield
- Cytokines may offer additional clues, especially early in the disease

Pitt | School of Medicine

Chapter 6

Imani recovers many of her prior abilities following the treatment for DSRD. Her family is understandably shaken by what looked like "early onset Alzheimer disease." They wonder if there is any research towards prevention of Alzheimer disease, perhaps through a vaccine. You tell them that the answer is: Yes! But the researchers ask: even if one were available, who would be interested in getting it?

Pitt | School of Medicine



Check for updates

Article

Views of parents of children with Down syndrome on Alzheimer's disease vaccination

Journal of Intellectual Disabilities
2024, Vol. 28(3) 759–772

DOI: 10.1177/17446295231177787
journals.sagepub.com/home/jid
Sage

Sarah Batbold
Mayo Clinic Alix School of Medicine, Mayo Clinic, Rochester, MN, USA

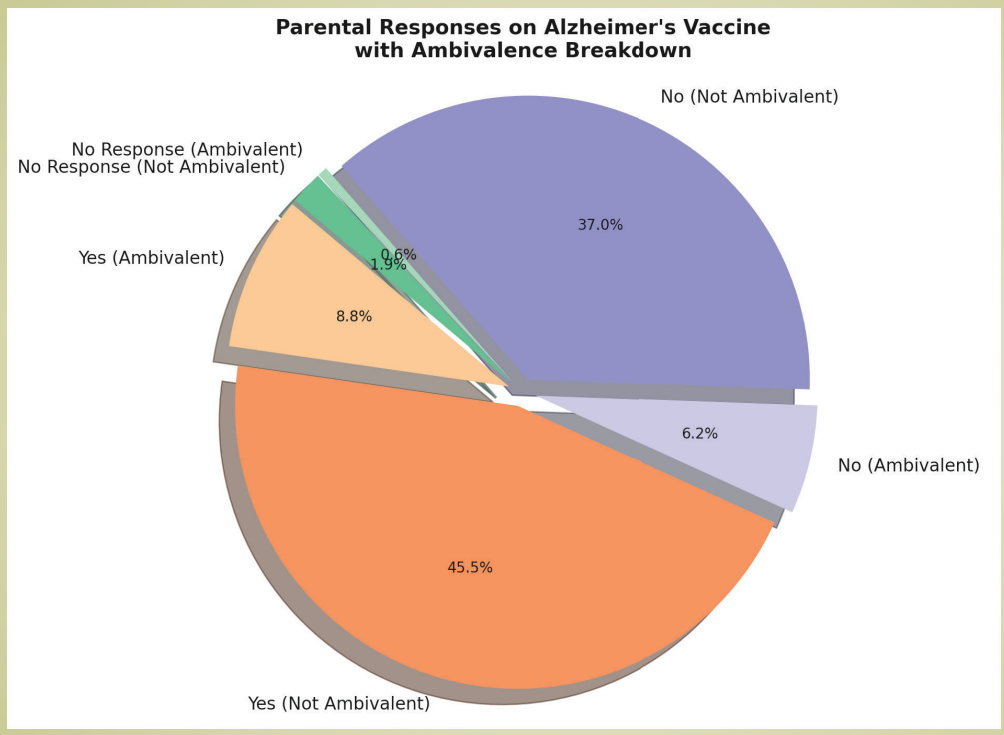
Gabrielle Cummings
Mayo Clinic Alix School of Medicine, Mayo Clinic, Rochester, MN, USA

Kirsten A. Riggan
Biomedical Ethics Research Program, Mayo Clinic, Rochester, MN, USA

Marsha Michie
Case Western Reserve University School of Medicine, Cleveland, OH, USA

Megan Allyse
Biomedical Ethics Research Program, Mayo Clinic, Rochester, MN, USA

Scenario	Benefits & Risks	Survey Questions
Gavin is 35 years old and has Down syndrome. Gavin and his family know that people with Down syndrome often develop Alzheimer's disease as young as 40 years old. Gavin's doctors say a new vaccine could help stop him from getting Alzheimer's.	Gavin would have fewer or milder symptoms of Alzheimer's disease. Gavin would have better memory and better ability to live on his own. The vaccine has a chance of causing seizures (epilepsy), and the long-term effects are unknown.	1. If you were Gavin's family, would you want him to choose to have the vaccine? (*yes/no*) 2. Why? (*open*) 3. What do you think are the most important risks and benefits? (*open*) 4. What other thoughts do you want to share about this? (*open*)



Take home points

Emphasis on Education and Evidence:

- Parents consistently emphasize the need for detailed, clear, and robust safety and efficacy data.
- Clinicians should be prepared to discuss extensively researched data, potential risks, and clear benefits during consultations.

High Importance of Autonomy:

- Parents strongly value including their adult child with Down syndrome in decision-making processes

Recognition of Emotional Context:

- Personal experiences with Alzheimer's disease heavily influence parental acceptance of preventive interventions.

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole
Pitt School of Medicine

Chapter 7

You find yourself at IKEA looking for more bookshelves to place all your awards and get a notification from the EMR that Imani's routine screening showed elevated levels of total cholesterol, LDL, and triglycerides. As you finish your Swedish meatballs, you prepare to reassure Imani and her family that cardiovascular events are less common in adults with Down syndrome. But in the IKEA cafeteria, you come across this article …

Journal of Internal Medicine June 2025

JIM Original Article

doi: 10.1111/joim.20093

Age-related cardiovascular disease in Down syndrome: A population-based matched cohort study

Annie Pedersen[1,2], Anna Skarin Nordenvall[3,4], Giorgio Tettamanti[4,5] & Ann Nordgren[1,2,4,6]

From the [1]Department of Laboratory Medicine, Institute of Biomedicine, University of Gothenburg, Gothenburg, Sweden; [2]Department of Clinical Genetics and Genomics, Sahlgrenska University Hospital, Gothenburg, Sweden; [3]Department of Radiology, Karolinska University Hospital, Stockholm, Sweden; [4]Department of Molecular Medicine and Surgery, Center for Molecular Medicine, Karolinska Institutet, Stockholm, Sweden; [5]Institute of Environmental Medicine, Karolinska Institutet, Unit of Epidemiology, Stockholm, Sweden; and [6]Department of Clinical Genetics, Karolinska University Hospital, Stockholm, Sweden

Cardiovascular Disease in Down Syndrome

- Study: Population-based matched cohort in Sweden
- 5,155 individuals with DS matched to 257,750 non-DS comparators
- Aim: Evaluate age-related cardiovascular risk in DS population

Pitt | School of Medicine

Methods

- Data from Swedish national registers (NPR, MBR)
- Outcomes: ischemic stroke, hemorrhagic stroke, Acute myocardial infarction

Pitt | School of Medicine

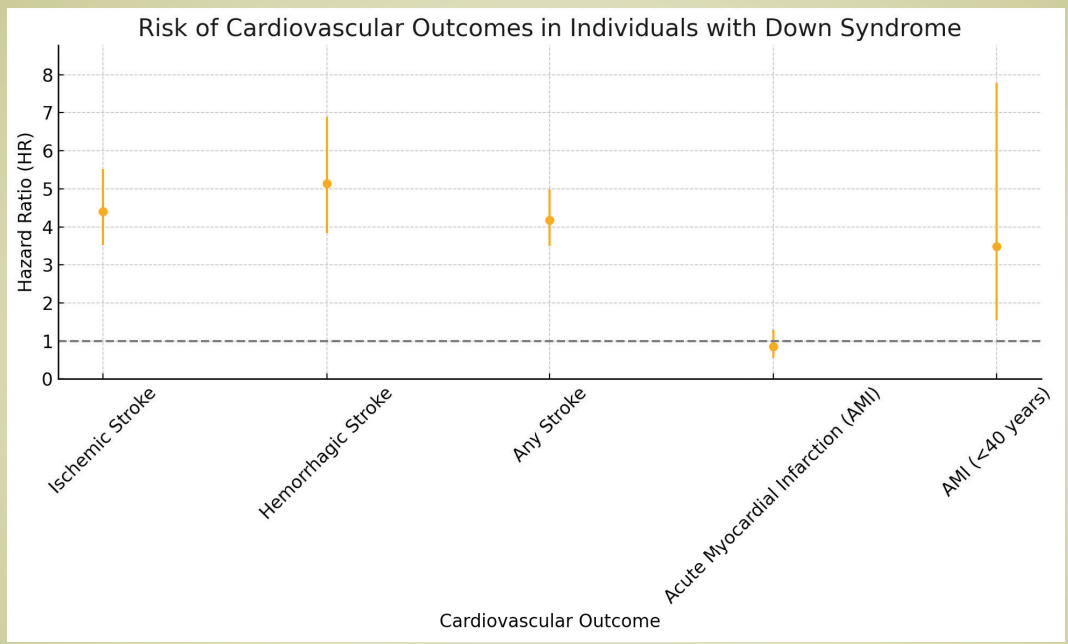
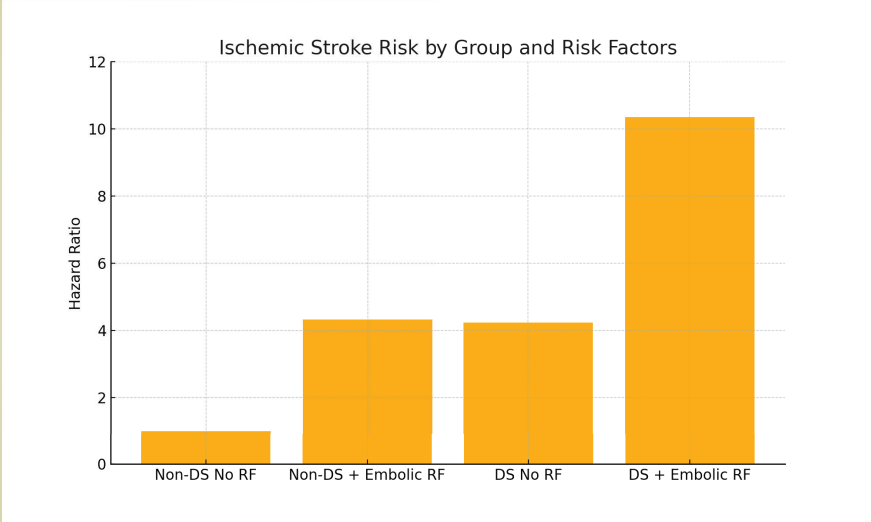


Chart: Ischemic Stroke by Risk Factors



DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole
Pitt School of Medicine

Key Findings: AMI Risk

- Overall AMI risk similar: HR 0.85 (95% CI 0.56–1.29)
- Increased AMI risk in DS < 40 years: HR 3.48 (95% CI 1.55–7.78)
- DS individuals less likely to receive coronary intervention: HR 0.13

Pitt | School of Medicine

Key Findings: Stroke Risk

- Ischemic stroke risk: HR 4.41 (95% CI 3.53–5.52)
- Hemorrhagic stroke risk: HR 5.14 (95% CI 3.84–6.89)
- Stroke risk highest with additional embolic or atherosclerotic risk factors

Pitt | School of Medicine

Risk Factor Interaction

- DS with embolic risk: HR 10.35 for ischemic stroke
- DS with atherosclerotic risk: HR 12.67 for ischemic stroke
- No clear impact of these risk factors on hemorrhagic stroke

Implications for Screening

- Traditional risk models may not apply to DS
- Monitoring in patients with congenital heart disease or diabetes is essential

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole
Pitt School of Medicine

Limitations

- Median follow-up age was ~40 years—may underestimate lifetime risk
- Lack of data on hypertension, lipids, smoking, obesity
- Potential surveillance bias due to frequent specialist visits in DS

Pitt | School of Medicine

Chapter 8

While living in her group home, 50-year-old Imani slips while running down the stairs in excitement for her upcoming clinic visit with you. She goes to the ED and is found to have suffered a tibial fracture. Her caretakers ask whether fractures are more common in adults with Down syndrome.

Pitt | School of Medicine

JIDR Journal of Intellectual Disability Research
Published on behalf of mencap and in association with IASSID

Journal of Intellectual Disability Research doi: 10.1111/jir.13183

VOLUME 68 PART 12 pp 1374–1385 DECEMBER 2024

Age and sex-specific risk in fractures with Down syndrome in a retrospective case–control study from Germany

S. Krieg,[1] A. Krieg[2] & K. Kostev[3]

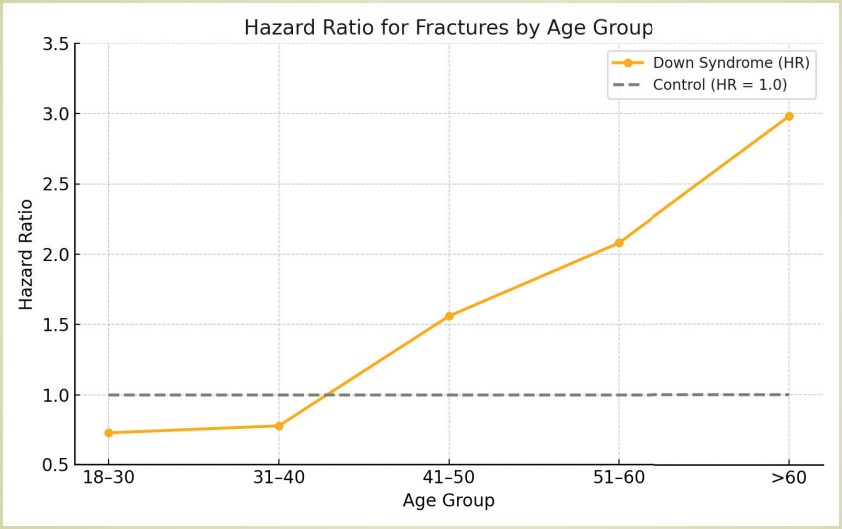
Study at a Glance

- 2547 adults with DS vs. 12,735 matched controls in Germany
- Tracked first fracture diagnosis over 5 years
- Focus: Age- and sex-specific fracture risk

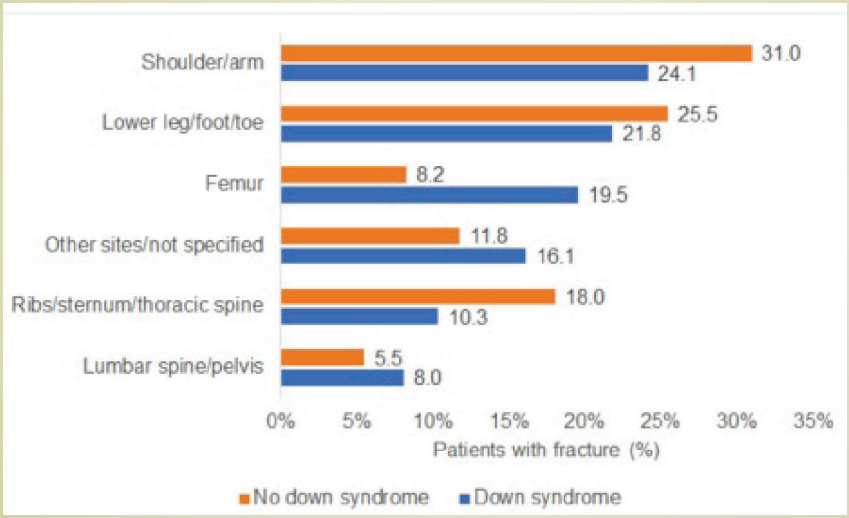
DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole

Pitt School of Medicine

Fracture Risk by Age in Down Syndrome



Body regions of fractures in patients with and without Down syndrome



DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole

Pitt School of Medicine

Key Findings – Fracture Risk

- Fracture risk increases significantly after age 50
- Women with DS are most at risk:
    - Age 51–60: 2x higher risk
    - Age >60: nearly 3x higher risk
- Femur fractures especially common in DS group

Pitt | School of Medicine

What Clinicians Should Know

- Begin risk assessments at age 50
- Review medications that may affect bone health
- Encourage safe, weight-bearing physical activity
- Evaluate home/living environment for fall risks

Pitt | School of Medicine

Chapter 9

Imani's caretakers discover that you were one of the authors of the 2020 Adult Healthcare Guidelines published in JAMA. They mention that it must feel so good that everyone who cares for adults with Down syndrome can now follow guidelines. And just when you start to feel good about yourself, you find this article…

Pitt School of Medicine

Received: 11 June 2024 | Revised: 25 July 2024 | Accepted: 5 August 2024

DOI: 10.1002/ajmg.a.63850

ORIGINAL ARTICLE

AMERICAN JOURNAL OF medical genetics PART A WILEY

Adherence to adult clinical practice guidelines for Down syndrome

Jordan C. Wood[1] | Perman Gochyyev[1] | Stephanie L. Santoro[2,3]

Pitt School of Medicine

Study Overview

- Reviewed charts of 327 adults with Down syndrome at MGH DSP.
- Evaluated adherence to 15 clinical guidelines (adult, pediatric, general).
- Mean adherence: 67.3%; only 14.7% fully up-to-date.

Pitt School of Medicine

Clinical Guidelines Adherence

- High Adherence (>80%)
  Thyroid (95%), BMI (92%), Weight Loss Counseling (90%)
  Dementia Screening (88%), Diabetes Screening (84%)
- Moderate Adherence (69%-81%)
  Ophthalmology (70%), Mammograms (73%)
  Celiac screening (81%), Vitamin D (69%)
- Low Adherence (<50%) -
  Iron screening (42%), Audiology (~35%)
  Colonoscopy (43%), Bone Density Scan (26%)
  Parathyroid hormone (22%)

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole
Pitt School of Medicine

Chapter 10

You are giving your favorite lecture of the year to over 150 of your colleagues, and you are contemplating the best way to wrap a bow on an amazing conference. You decide to share an article written by an adult with Down syndrome on their experience with Alzheimer's disease in their grandparent so that she can remind us all of why we do what we do.

Received: 10 December 2024 | Accepted: 23 February 2025

DOI: 10.1002/alz.70204

Alzheimer's & Dementia®
THE JOURNAL OF THE ALZHEIMER'S ASSOCIATION

PERSPECTIVE

Down syndrome versus dementia

Eden Rapp

School of Public Health, Boston University, Boston, USA

Correspondence
Eden Rapp, School of Public Health, Boston University, Boston 02118, United States.
Email: integrityrocks6@gmail.com

- Eden Rapp is a 28-year-old with Down syndrome whose grandmother developed Alzheimer's dementia. Here are her insights she learned from the experience:

- Issues her grandmother experienced:
  - Memory Loss
  - Being Territorial
  - Being Disoriented

- Eden's take home points:

There are still important things that young adults with Down syndrome can do to prevent and delay dementia.

1. Not being isolated and having a support network
2. Not feeling afraid to do things on your own
3. Taking care of yourself physically, emotionally, and spiritually
4. Eating healthy
5. Having an active mind

DSMIG-USA ANNUAL Symposium JULY 24-26, 2025 Dallas, Texas | Hilton Anatole

Pitt School of Medicine

Epilogue

You are attending the world's greatest Down Syndrome Medical Conference. You have a question or comment about something you've just heard, and you boldly raise your hand for the microphone to ask …