Evaluation, treatment, and support for patients with Down syndrome and Alzheimer's disease

Massachusetts General Hospital Down Syndrome Program

Massachusetts General Hospital
Founding Member, Mass General Brigham

HARVARD MEDICAL SCHOOL

Disclosures

BRIAN SKOTKO, MD, MPP

- I occasionally consult on the topic of Down syndrome through Gerson Lehrman Group.
- I receive remuneration from Down syndrome non-profit organizations for speaking engagements and associated travel expenses.
- I receive annual royalties from Woodbine House, Inc., for the publication of my book, *Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters*.
- Within the past 3 years, I have received research funding from F. Hoffmann-La Roche, Inc., AC Immune, and LuMind IDSC Down Syndrome Foundation to conduct clinical trials for people with Down syndrome.
- I am occasionally asked to serve as an expert witness for legal cases where Down syndrome is discussed.
- I serves in a non-paid capacity on the Honorary Board of Directors for the Massachusetts Down Syndrome Congress and the Professional Advisory Committee for the National Center for Prenatal and Postnatal Down Syndrome Resources.
- I have a sister with Down syndrome.

Disclosures

DOMINICA NICHOLS, PhD, RD, LDN

- Stock in Abbott (a company that makes nutritional products); I do not consult with them.

Disclosures

AMANDA LAEZZA, RD, LDN

No disclosures

Disclosures

NICOLAS ORESKOVIC, MD, MPH

- I have served as an expert witness for a legal case where Down syndrome was discussed.
- I have received remuneration from a Down syndrome non-profit organization for a speaking engagement and associated travel expenses.
- I serve in a non-paid capacity as a member on the Medical and Scientific Advisory Council for the Massachusetts Down Syndrome Congress.

Disclosures

CAROLINE BREGMAN, LICSW

No disclosures

Disclosures

STEPHANIE L. SANTORO, MD

Dr. Santoro is an unpaid volunteer on the Medical and Scientific Advisory Committee and the Board of the Massachusetts Down Syndrome Congress, a fellow and member of Executive Committee of the Council on Genetics of the American Academy of Pediatrics, a member of the Board of the Down Syndrome Medical Interest Group (DSMIG-USA) and provides clinical care in the MassGeneral Hospital Down Syndrome Program.

Learning objectives

1. Learners will understand how best to **screen for, evaluate, and treat** Alzheimer's disease in adults with Down syndrome.
2. Learners will gain knowledge about the **national resources and supports** that are available for family members who have a loved one with Down syndrome and Alzheimer’s disease.
3. Learners will understand how best to support the **co-occurring nutritional needs** of patients with Down syndrome who have dementia.

Outline of Talk

Speaker 1: **Stephanie Santoro**, MD (10 min)
How do we screen our patients with Down syndrome for Alzheimer's disease?

Speaker 2: **Nicolas Oreskovic,** MD, MPH (10 min)
How do we care for our patients with Down syndrome diagnosed with Alzheimer's disease?

Speaker 3: **Dominica Nichols**, PhD, RD, LDN, and **Amanda Laezza**, RD, LDN (10 min)
What are the nutritional considerations?

Speaker 4: **Caroline Bregman**, LICSW (10 min)
What are the psychosocial supports for families?

Speaker 5: **Brian Skotko**, MD, MPP (10 min)
How do we implement a Brain Donation Program?

Clinical Screeners: MGH DSP Dementia Screening Protocol

Stephanie L. Santoro, MD

Massachusetts General Hospital
Associate Professor of Pediatrics, Harvard Medical School

Massachusetts General Hospital
Founding Member, Mass General Brigham

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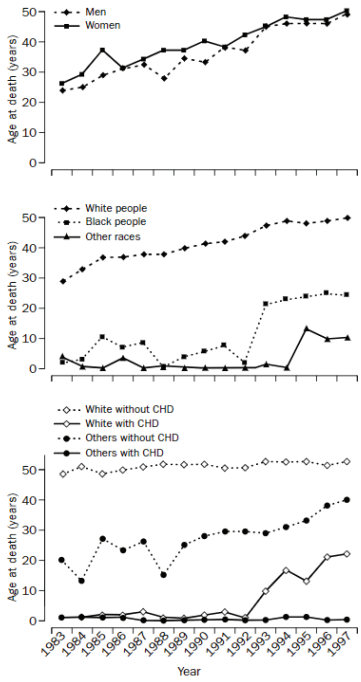
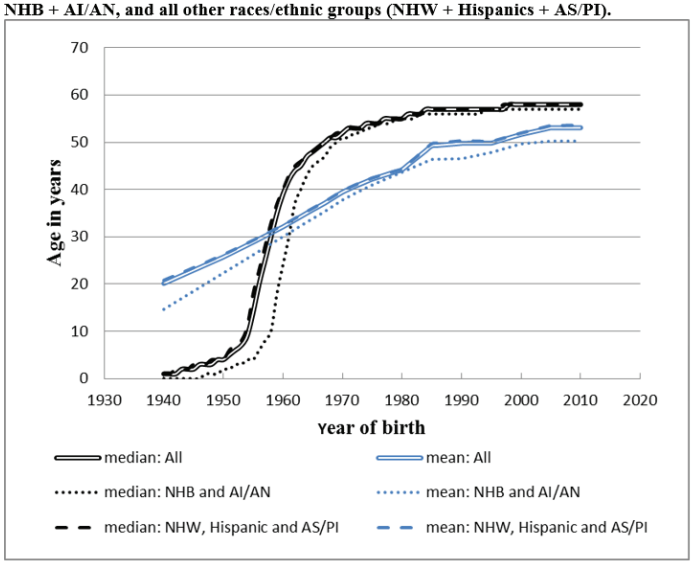


Figure 2: **Median age at death of people with Down's syndrome by sex (upper), racial group (middle), and with or without congenital heart defects (CHD) by racial group (lower)**

Life Expectancy in Down Syndrome

Figure S3B. Estimated mean and median life expectancy for different years of birth as constructed on basis of the survival curves used in the current model; separately for all, NHB + AI/AN, and all other races/ethnic groups (NHW + Hispanics + AS/PI).



Alzheimer's Disease in Down Syndrome

Alzheimers Dement. 2015 June ; 11(6): 700–709. doi:10.1016/j.jalz.2014.10.007.

Down syndrome and Alzheimer's disease: Common pathways, common goals

Dean Hartley[a,*], Thomas Blumenthal[b,c], Maria Carrillo[a], Gilbert DiPaolo[d], Lucille Esralew[e], Katheleen Gardiner[b,f], Ann-Charlotte Granholm[g], Khalid Iqbal[h], Michael Krams[i], Cynthia Lemere[j], Ira Lott[k], William Mobley[l], Seth Ness[i], Ralph Nixon[m], Huntington Potter[b,n], Roger Reeves[o], Marwan Sabbagh[p], Wayne Silverman[q,r], Benjamin Tycko[d], Michelle Whitten[s], and Thomas Wisniewski[t]

Amyloid precursor protein (*APP*) gene on chr21

Neuropathology consistent with AD by the 40s

90% of people with Down syndrome will develop Alzheimer's disease by age 65+

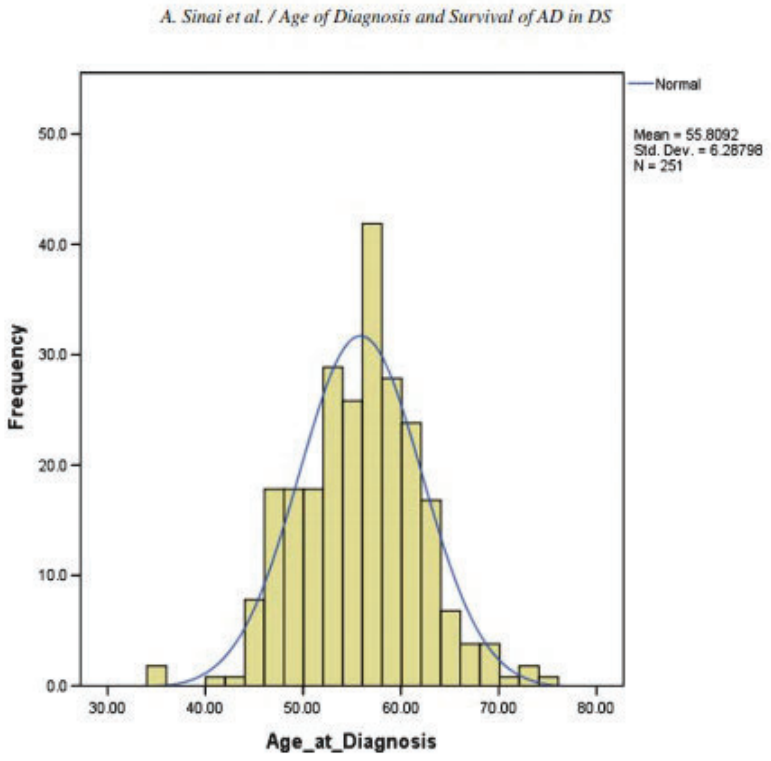


Fig. 1. Distribution of age at dementia diagnosis.

Hartley, et al. 2015.

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Adult Healthcare Guidelines published Oct 2020

Clinical Review & Education

JAMA | Special Communication

Medical Care of Adults With Down Syndrome
A Clinical Guideline

Amy Y. Tsou, MD, MSc; Peter Bulova, MD; George Capone, MD; Brian Chicoine, MD; Bryn Gelaro, MA, LSW; Terry Odell Harville, MD, PhD, D(ABMLI), D(ABHI); Barry A. Martin, MD; Dennis E. McGuire, PhD, LCSW; Kent D. McKelvey, MD; Moya Peterson, PhD, APRN, FNP-BC; Carl Tyler, MD, MSc; Michael Wells, BS; Michelle Sie Whitten, MA; for the Global Down Syndrome Foundation Medical Care Guidelines for Adults with Down Syndrome Workgroup

IMPORTANCE Down syndrome is the most common chromosomal condition, and average life expectancy has increased substantially, from 25 years in 1983 to 60 years in 2020. Despite the unique clinical comorbidities among adults with Down syndrome, there are no clinical guidelines for the care of these patients.

OBJECTIVE To develop an evidence-based clinical practice guideline for adults with Down syndrome.

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eFigure 2: GLOBAL Medical Care Guidelines for Adults with Down Syndrome Checklist

GLOBAL DOWN SYNDROME FOUNDATION

This checklist is intended to support the health of adults with Down syndrome directly or through their caregivers. We encourage this checklist to be shared with your medical professionals. Statements in blue represent our recommended, periodic health screenings/assessments that should begin at a specific age. Below each blue screening/assessment recommendation, there are blank boxes. Caregivers or individuals with Down syndrome can check off, date, or initial each blank box when the screening/assessment is completed. For screening/assessment recommendations with a time range (e.g. 1-2 years), the box size represents the longer possible time frame, such as 2 years versus 1. Statements in gray represent advisory recommendations that individuals with Down syndrome and caregivers should follow throughout adulthood.

Screening/Assessment | Advisory | Checkbox | No Recommendations

	21-29 Years	30-39 Years	40-49 Years	50-59 Years	60+ Years
Behavior	A review of behavioral, functional, adaptive, and psychosocial factors should be performed as part of an annual history that clinicians obtain from all adults with Down syndrome, their families, and caregivers.				
	When concern for a mental health disorder in adults with Down syndrome is present medical professionals should: a) Evaluate for medical conditions that may present with psychiatric and behavioral symptoms and b) Refer to a clinician knowledgeable about the medical, mental health disorders, and common behavioral characteristics of adults with Down syndrome.				
	When concern for a mental health disorder in adults with Down syndrome is present, medical professionals should follow guidelines for diagnosis in the Diagnostic and Statistical Manual of Mental Disorders (DSM 5). The Diagnostic Manual-Intellectual Disability 2 (DM-ID-2) also may be used to adapt diagnostic criteria from the DSM-5.				
Dementia	Caution is needed when diagnosing age-related, Alzheimer's Type Dementia in adults with Down syndrome less than age 40.		Medical professionals should assess adults with Down syndrome and interview their primary caregivers about changes from baseline function annually beginning at age 40. Decline in the six domains as per the National Task Group – Early Detection Screen for Dementia (NTG-EDSD) should be used to identify early-stage age-related Alzheimer's-type dementia and/or a potentially reversible medical condition.		
Cardiac			statin therapy should be assessed every 5 years starting at age 40 and using a 10-year risk calculator as recommended for adults without Down syndrome by the U.S. Preventive Services Task Force.		
	For adults with Down syndrome, risk factors for stroke should be managed as specified by the American Heart Association/American Stroke Association's Guidelines for the Primary Prevention of Stroke.				
	In adults with Down syndrome with a history of congenital heart disease, given the elevated risk of cardioembolic stroke, a periodic cardiac evaluation and a corresponding monitoring plan should be reviewed by a cardiologist.				
Obesity	Healthy diet, regular exercise, and calorie management should be followed by all adults with Down syndrome as part of a comprehensive approach to weight management, appetite control, and enhancement of quality of life.				
	Monitoring for weight change and obesity should be performed annually by calculating Body Mass Index in adults with Down syndrome. The U.S. Preventive Services Task Force Behavioral Weight Loss Interventions to Prevent Obesity-Related Morbidity and Mortality in Adults should be followed.				
Atlantoaxial Instability	In adults with Down syndrome, routine cervical spine x-rays should not be used to screen for risk of spinal cord injury in asymptomatic individuals.				
	Annual screening for adults with Down syndrome should be based on a review of signs and symptoms of cervical myelopathy using targeted history and physical exam.				
Osteoporosis	For primary prevention of osteoporotic fractures in adults with Down syndrome, there is insufficient evidence to recommend for or against applying established osteoporosis screening guidelines,including fracture risk estimation; thus, good clinical practice would support a shared decision-making approach to this issue would support a shared decision-making approach to this issue.				
	All adults with Down syndrome who sustain a fragility fracture should be evaluated for secondary causes of osteoporosis, including screening for hyperthyroidism, celiac disease, vitamin D deficiency, hyperparathyroidism and medications associated with adverse effects on bone health.				
Thyroid	Screening adults with Down syndrome for hypothyroidism should be performed every 1-2 years using a serum thyroid-stimulating hormone (TSH) test beginning at age 21.				
Celiac Disease	Adults with Down syndrome should receive an annual assessment for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease using targeted history, physical examination and clinical judgement of good practice.				

This checklist is not intended to be diagnostic. Presentation of medical and mental health conditions for people with Down syndrome may be atypical. Similar signs and symptoms may be a consequence of multiple reasons, including different disease processes. Thus, the patient evaluation should include considerations of additional causes for any detected sign or symptom. The development of new and/or changes in signs or symptoms should prompt a comprehensive evaluation with your clinician.

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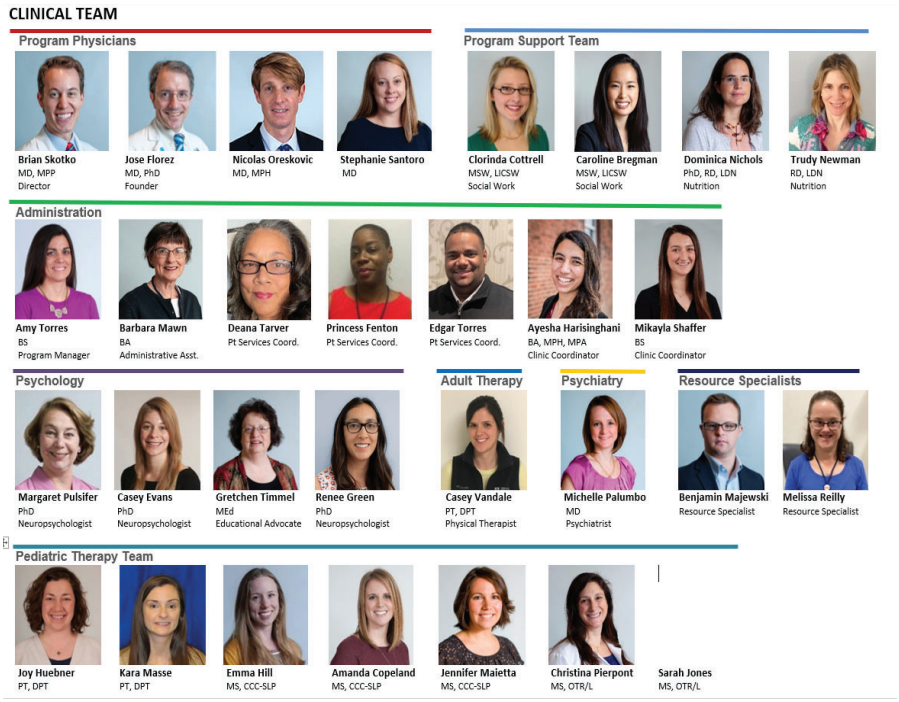
Protocol development

Developed in 2021, with multidisciplinary input

Piloted with a few families first, then launched in Dec 2021

Piloted with 2 physicians first, then added third physician in 2023

Protocol is multidisciplinary and begins at age 40



Dementia Protocol: Screening Tools

Screeners chosen by:
-caregiver-completed
-short time and # of items
-could be completed before visits
-freely-available
-produces score / cut-offs

All Adults age 40+:

- Routine health maintenance
- Screen for dementia (pre-visit in REDCap)
- Mental health screening (pre-visit in REDCap)
- Baseline neuropsychology eval if never done

The National Task Group on Intellectual Disabilities and Dementia Practices' Early Detection and Screen for Dementia (NTG-EDSD): The NTG-EDSD is an administrative screening tool used to record observed changes in function.[9]

American Association on Mental Deficiency (AAMD) Adaptive Behavior Scale: The AAMD behavior scales have been published in two versions, and includes 65 items covering ten domains.[12]

Dementia Questionnaire for People with Learning Disabilities (DLD): The DLD is a 50-item instrument, subdivided into two subcategories and eight subscales. Each item has three response categories; points of each subcategory are summed up to scores. The diagnostic criterion for dementia is based on score changes over time.[11,13] The DLD was studied in 78 adults with DS, ages 35 and older, with varied intellectual disability.[14] In a cohort with DS, using the lower cut-score of 4, the DLD was found to have a sensitivity of 100% and specificity of 69%.[14] One limitation of the DLD is that it requires repeated use to track score changes over time.

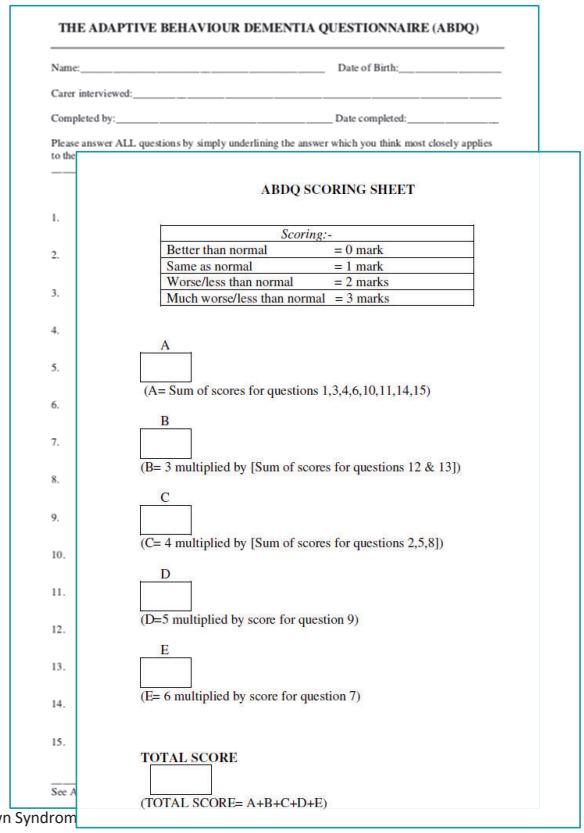
Screener Used First

Adaptive Behaviour Dementia Questionnaire (ABDQ)

- a short 15-item questionnaire
- yields a score with established values which indicate normal, mild dementia, moderate dementia or profound dementia
- Has been studied in adults with DS
- Studied in comparison to AD diagnosis on ICD-10 criteria in the past: the sensitivity was 89%, the specificity was 94%, the positive predictive value was 89%, and the negative predictive value 94%.[10] The authors describe that the overall percentage correct identification (accuracy) of AD and non-AD cases was 92%.[10]

Process: entered into REDCap, shared with caregivers before visit, score shared during pre-visit team huddle

Screeners chosen by:
-caregiver-completed
-short length and # of items
-could be completed before visits
-freely-available
-produces score / cut-offs



THE ADAPTIVE BEHAVIOUR DEMENTIA QUESTIONNAIRE (ABDQ)

Name:_____ Date of Birth:_____

Carer interviewed:_____

Completed by:_____ Date completed:_____

Please answer ALL questions by simply underlining the answer which you think most closely applies to the

1.
2.
3.
4.
5.
6.
7.
8.
9.
10.
11.
12.
13.
14.
15.

See A

ABDQ SCORING SHEET

Scoring:-	
Better than normal	= 0 mark
Same as normal	= 1 mark
Worse/less than normal	= 2 marks
Much worse/less than normal	= 3 marks

A
(A= Sum of scores for questions 1,3,4,6,10,11,14,15)

B
(B= 3 multiplied by [Sum of scores for questions 12 & 13])

C
(C= 4 multiplied by [Sum of scores for questions 2,5,8])

D
(D=5 multiplied by score for question 9)

E
(E= 6 multiplied by score for question 7)

TOTAL SCORE
(TOTAL SCORE= A+B+C+D+E)

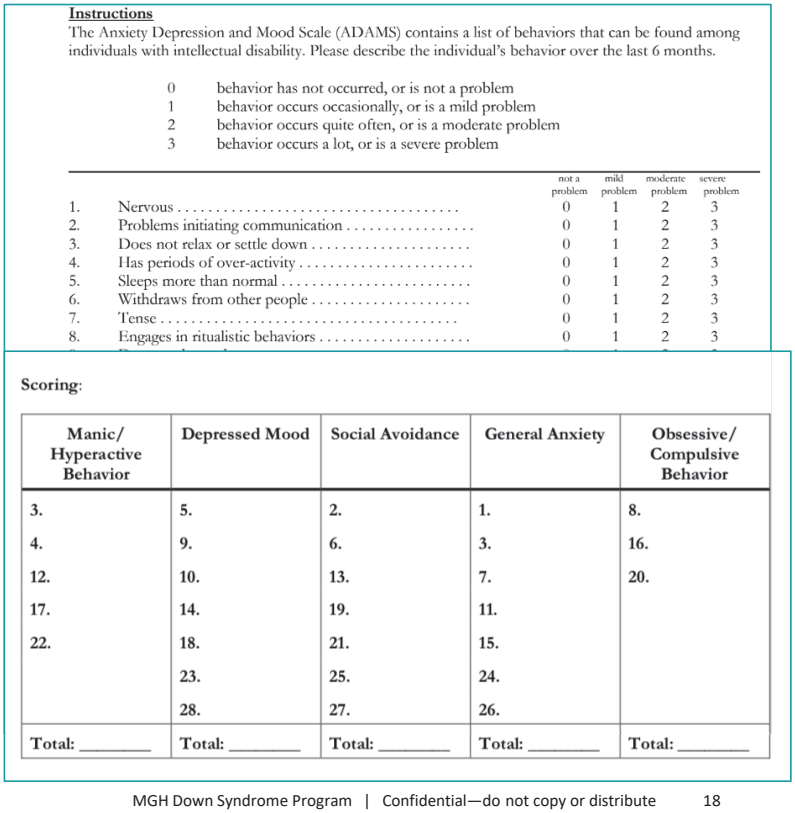
Screeners Used First

Anxiety, Depression and Mood Scale (ADAMS)

- validated questionnaire consisting of 28 questions with five subscales: Manic / Hyperactive Behavior, Depressed Mood, Social Avoidance, General Anxiety, and Compulsive Behavior
- Yields a score
- Has been studied before in DS[8]

Process: entered into REDCap, shared with caregivers before visit, score shared during pre-visit team huddle

Screeners chosen by:
-caregiver-completed
-short length and # of items
-could be completed before visits
-freely-available
-produces score / cut-offs



Instructions

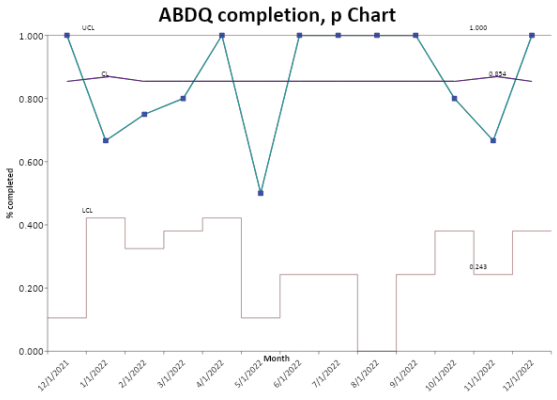
The Anxiety Depression and Mood Scale (ADAMS) contains a list of behaviors that can be found among individuals with intellectual disability. Please describe the individual's behavior over the last 6 months.

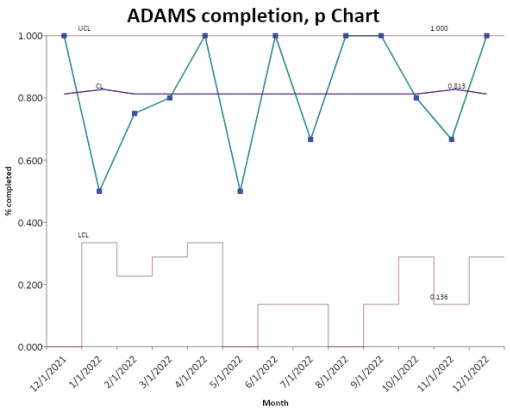
0 behavior has not occurred, or is not a problem
1 behavior occurs occasionally, or is a mild problem
2 behavior occurs quite often, or is a moderate problem
3 behavior occurs a lot, or is a severe problem

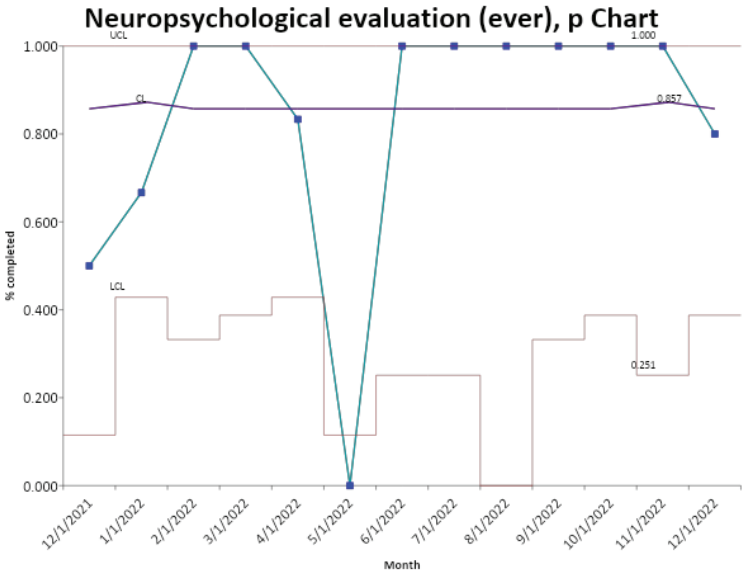
		not a problem	mild problem	moderate problem	severe problem
1.	Nervous	0	1	2	3
2.	Problems initiating communication	0	1	2	3
3.	Does not relax or settle down	0	1	2	3
4.	Has periods of over-activity	0	1	2	3
5.	Sleeps more than normal	0	1	2	3
6.	Withdraws from other people	0	1	2	3
7.	Tense	0	1	2	3
8.	Engages in ritualistic behaviors	0	1	2	3

Scoring:

Manic/ Hyperactive Behavior	Depressed Mood	Social Avoidance	General Anxiety	Obsessive/ Compulsive Behavior
3.	5.	2.	1.	8.
4.	9.	6.	3.	16.
12.	10.	13.	7.	20.
17.	14.	19.	11.	
22.	18.	21.	15.	
	23.	25.	24.	
	28.	27.	26.	
Total: _____	Total: _____	Total: _____	Total: _____	Total: _____







On average, 85% had ABDQ and 81% age 40+ had ADAMS in first year

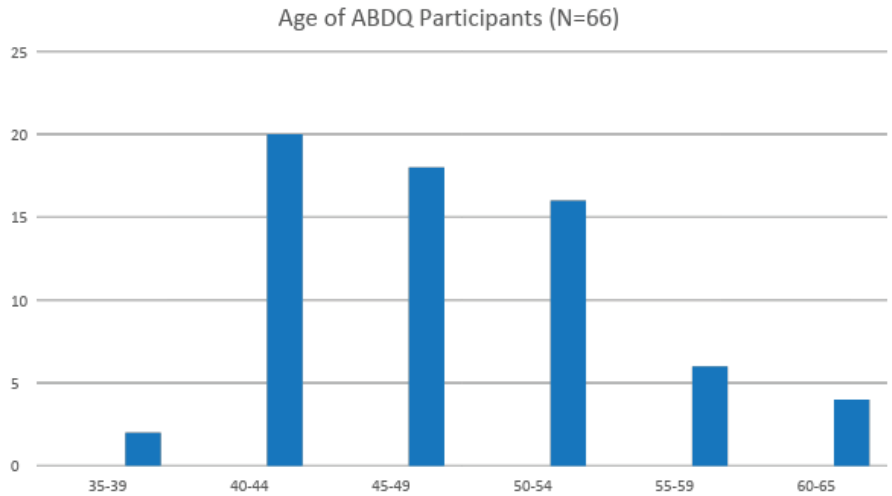
86% had ever had neuropsychology

Demographics Reviewed

56% male / 44% female
94% White / 97% Not Hispanic
Age: 39 to 63 years (mean=49±5.8)

Premorbid function:

- 20% Mild
- 5% Mild-moderate
- 45% Moderate
- 5% unspecified
- 25% not yet completed



Lessons learned: first screeners

- Success with implementing a novel protocol
- We noticed:
  - Dementia screener results did not match clinical suspicion
  - We kept track of ABDQ result and compared to our clinical notes
- Screeners:
  - Families had confusion with word “normal”
  - Families thought about “usual” in different ways

ABDQ: Dec 2021 to April 2023

	Clinical Assessment		
	Dementia and/or Cognitive Impairment	No Dementia and/or Cognitive Impairment	Total
Positive: ABDQ ≥ 78	0	1	1
Negative: ABDQ < 78	10	39	49
Total	10	40	50

Sensitivity: (0)/(0+10) = 0%
Specificity: (39)/(39+1) = 98%
PPV: (0)/(0+1) = 0%
NPV: (39)/(39+10) = 80%

Screener change from ABDQ to Use Of NTG-EDSD

	Always been the case	Always but worse in the past year	New symptom in past year	Does not apply
[19] **Activities of Daily Living**				
Needs help with washing and/or bathing				
Needs help with dressing				
Dresses inappropriately (e.g., back to front, incomplete, inadequately for weather)				
Undresses inappropriately (e.g., in public)				
Needs help eating (cutting food, mouthful amounts, choking)				
Needs help using the bathroom (finding, toileting)				
Incontinent (including occasional accidents)				
[20] **Language & Communication**				
Does not initiate conversation				
Does not find words				
Does not follow simple instructions				
Appears to get lost in middle of conversation				
Does not read				
Does not write (including printing own name)				
[21] **Sleep-Wake Change Patterns**				
Excessive sleep (sleeping more)				
Inadequate sleep (sleeping less)				
Wakes frequently at night				
Confused at night				
Sleeps during the day more than usual				
Wanders at night				
Wakes earlier than usual				
Sleeps later than usual				
[22] **Ambulation**				
Not confident walking over small cracks, lines on the ground, patterned flooring, or uneven surfaces				
Unsteady walk, loses balance				
Falls				
Requires aids to walk				

	Always been the case	Always but worse in the past year	New symptom in past year	Does not apply
[23] **Memory**				
Does not recognize familiar persons (staff/relatives/friends)				
Does not remember names of familiar people				
Does not remember recent events (in past week or less)				
Does not find way in familiar surroundings				
Loses track of time (time of day, day of the week, seasons)				
Loses or misplaces objects				
Puts familiar things in wrong places				
Problems with printing or signing own name				
Problems with learning new tasks or names of new people				
[24] **Behavior and Affect**				
Wanders				
Withdraws from social activities				
Withdraws from people				
Loss of interest in hobbies and activities				
Seems to go into own world				
Obsessive or repetitive behavior				
Hides or hoards objects				
Does not know what to do with familiar objects				
Increased impulsivity (touching others, arguing, taking things)				
Appears uncertain, lacks confidence				
Appears anxious, agitated, or nervous				
Appears depressed				
Shows verbal aggression				
Shows physical aggression				
Temper tantrums, uncontrollable crying, shouting				
Shows lethargy or listlessness				
Talks to self				
[25] **Adult's Self-reported Problems**				
Changes in ability to do things				
Hearing things				
Seeing things				
Changes in 'thinking'				
Changes in interests				
Changes in memory				
[26] **Notable Significant Changes Observed by Others**				
In gait (e.g., stumbling, falling, unsteadiness)				
In personality (e.g., subdued when was outgoing)				
In friendliness (e.g., now socially unresponsive)				
In attentiveness (e.g., misses cues, distracted)				
In weight (e.g., weight loss or weight gain)				
In abnormal voluntary movements (head, neck, limbs, trunk)				

Process: administered on paper-and-pencil format, shared with team after caregivers complete

NTG-EDSD: 12 months of pilot data (Sept 2023 to September 2024)

	Clinical Assessment	
	Yes – Dementia/ MCI	No – Dementia/ MCI
NTG-EDSD (≥1)	14	23
NTG-EDSD (< 1)	0	17

Sensitivity: (14)/(14+0) = 100%
Specificity: (17)/(23+17) = 43%
PPV: (14)/(23+17) = 38%
NPV: (17)/(17+0) = 100%

Clinical Care:
MGH DSP Dementia Screening Protocol

Nicolas M. Oreskovic, MD, MPH

Massachusetts General Hospital
Assistant Professor of Pediatrics, Harvard Medical School

MGH Down Syndrome Program: Dementia Screening Protocol

- Begins at age 40 years
- Interdisciplinary and collaborative process
- Baseline neuropsychological assessment
- Annual screening
- Caregiver report(s) + clinical assessment
- Laboratory testing as indicated
- Specialist consultation as indicated

Age and Timing

- Dementia protocol begins in all patients at age 40 years

During Visit:

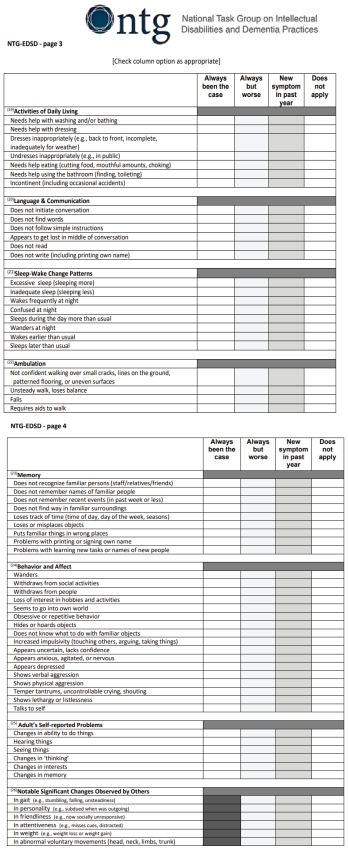
- Begin discussing process with patients and caregiver(s)
- MD assessment, SW assessment
- Initial dementia screening questionnaire at age 40 years
    - Repeat screening at each annual visit thereafter
- Documentation in EHR - use of MGH DSP dementia protocol template

Referral:

- Baseline neuropsychological assessment (PhD) at age 40 years (if not previously obtained)
    - Repeat neuropsychological assessment as indicated if concern about change in status

Screening Questionnaire

- Caregiver-reported questionnaire
  - Initially tried ABDQ - performed poorly
  - Currently using modified NTG-EDSD
    - Using pages 3 & 4 to identify any change in function
    - Completed by caregiver
    - Screener, not diagnostic → purpose is to identify possible concerns & initiate discussion **(2 grey columns)**
    - Rapid (5 minutes for caregiver to complete, 30 seconds for MD to review)
    - Structured Domains: Identifies domains of potential concern (ADLs, communication, sleep, behavior, memory, etc)



ntg National Task Group on Intellectual Disabilities and Dementia Practices

NTG-EDSD - page 3

[Check column option as appropriate]

	Always been the case	Always but worse	New symptom in past year	Does not apply
[18] **Activities of Daily Living**				
Needs help with washing and/or bathing				
Needs help with dressing				
Dresses inappropriately (e.g., back to front, incomplete, inadequately for weather)				
Undresses inappropriately (e.g., in public)				
Needs help eating (cutting food, mouthful amounts, choking)				
Needs help using the bathroom (finding, toileting)				
Incontinent (including occasional accidents)				
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Wanders at night				
Wakes earlier than usual				
Sleeps later than usual				
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Not confident walking over small cracks, lines on the ground, patterned flooring, or uneven surfaces				
Unsteady walk, loses balance				
Falls				
Requires aids to walk				

NTG-EDSD - page 4

	Always been the case	Always but worse	New symptom in past year	Does not apply
[23] **Memory**				
Does not recognize familiar persons (staff/relatives/friends)				
Does not remember names of familiar people				
Does not remember recent events (in past week or less)				
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Loses track of time (time of day, day of the week, seasons)				
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Problems with printing or signing own name				
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Changes in memory				
[26] **Notable Significant Changes Observed by Others**				
In gait (e.g., stumbling, falling, unsteadiness)				
In personality (e.g., subdued when was outgoing)				
In friendliness (e.g., now socially unresponsive)				
In attentiveness (e.g., misses cues, distracted)				
In weight (e.g., weight loss or weight gain)				
In abnormal voluntary movements (head, neck, limbs, trunk)				

Clinical Assessment

- Assess & Treat alternate medical explanations:
  - Universally recommend: Vision testing, audiology, Sleep Study, Thyroid function tests, TTg IgA, CBC, Vitamin B12, Folate, Urinalysis & culture, Abdominal Xray (constipation), brain MRI
  - Consider: RPR, HIV, Vitamin B1 (Thiamine), EEG

- Referral to specialist as indicated
  - Ophtho/Otology
  - Sleep Medicine
  - Neurology (seizures, dementia)
  - Dental
  - Other

Clinical Documentation

Use preformatted and editable ‘smartphrase’ in Epic (Electronic Health Record)

Dementia Screening:

Adults with Down syndrome are now living longer and fuller lives, with the current average life expectancy of individuals with Down syndrome into the 60's. There are several medical issues that can occur in older age adults with Down syndrome, one of the biggest concerns for families is the possible development of dementia. Dementia is a general term for a decline in mental ability severe enough to interfere with daily life. Dementia is not a single disease; it is the umbrella term for an individual's changes in memory, thinking, or reasoning. There are many possible causes of dementia, including Alzheimer's – disorders grouped under the general term "dementia" are caused by abnormal brain changes. We discussed the increased prevalence of Alzheimer's disease in Down syndrome due to the genetic link between Down syndrome and the neuropathology of Down syndrome, although not everyone with Down syndrome will develop this condition. Individuals with Down syndrome experience accelerated aging and may "slow down" in their 40's and 50's, even without a formal diagnosis of dementia or Alzheimer's. The prevalence of Alzheimer's in adults with Down syndrome varies based on numerous epidemiologic studies, but can begin between the ages of 35-40 years, and in general about 35% of individuals with Down syndrome will have dementia in their 50's and about 55% in their 60's. As nearly all patients with Down syndrome have signs of neuropathology present by their early 40's, and as these changes may not always be noticeable to caregivers or readily detectable clinically, the MGH Down Syndrome Programs screens all patients ages 40 and older annually for dementia. @FNAME@ is at the age where adults with Down syndrome are at increased risk for dementia, and was therefore screened as part of today's visit, using caregiver report and a clinical evaluation. We also conducted the National Task Group-Early Detection Screen for Dementia (NTG-EDSD) (https://www.the-ntg.org/ntg-edsd) as part of an ongoing process being developed by the MGH Down Syndrome Program aimed at providing annual dementia screening to all patients 40 years and older with Down syndrome.

Based on @FNAME@'s clinical evaluation and NTG-EDSD score, @HIS@ screening results indicate that it is likely/unlikely that @FNAME@ has dementia at this time.

As part of our comprehensive work-up, we recommend that all patients have a baseline neuropsychologic assessment at or after 40 years of age. @FNAME@ {Blank single:19197::"has","has not"} had a baseline neuropsychologic assessment. A baseline assessment, performed while an adult is healthy and well, will allow us to have a basis for comparison should signs of dementia develop in the future.

In addition, we recommended/provided/discussed the following resources with the caregiver(s):

- Down Syndrome Brain Train: an online program reviewing evidence-based recommendations aimed at optimizing brain health, including information on physical activity, diet, and socialization, and treatment of co-occurring medical conditions. More at www.downsyndromebraintrain.com
- NDSSS Aging and Down Syndrome: A Health & Well-Being Guidebook www.ndss.org/wp-content/uploads/2017/11/Aging-and-Down-Syndrome.pdf

Resources for Families

We recommended/provide:

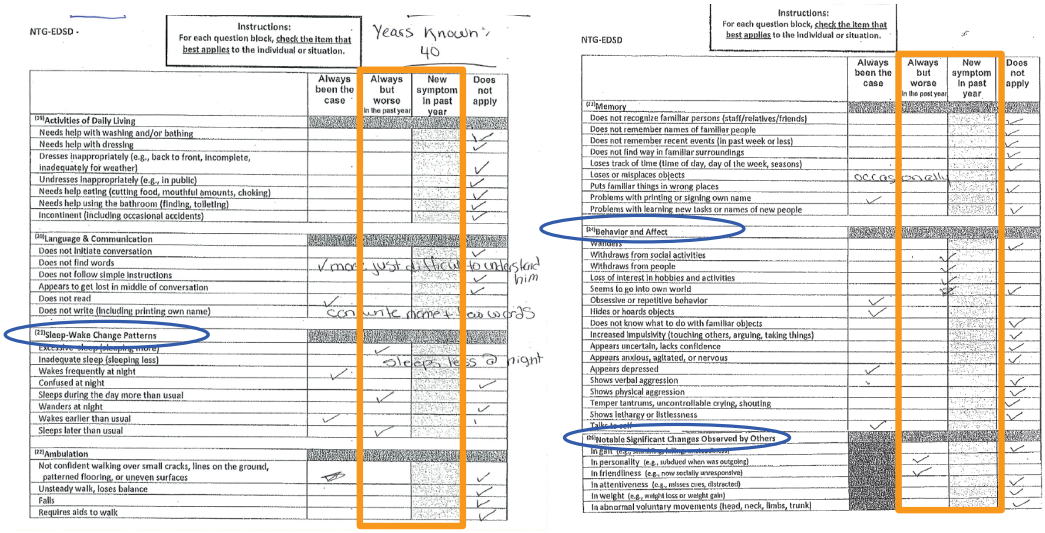
- Down Syndrome Brain Train: online program reviewing evidence-based recommendations aimed at optimizing brain health [physical activity, diet, socialization, and treatment of co-occurring medical conditions] www.downsyndromebraintrain.com

- NDSS Aging and Down Syndrome: A Health & Well-Being Guidebook www.ndss.org/wp-content/uploads/2017/11/Aging-and-Down-Syndrome.pdf

Case: Mr. H

- 40 year old man, no prior memory concerns, presents for annual DS visit
- Caregiver report of changes in sleep patterns and social interaction (more withdrawn) over past year
- Followed by Psychiatry for depression and OCD-like behaviors, on SSRI
- PMH: OSA cannot tolerate CPAP, myopia will not wear glasses

NTG -EDSD screen: caregiver completed, physician reviewed



NTG-EDSD

Instructions: For each question block, check the item that best applies to the individual or situation.

Yeass known: 40

	Always been the case	Always but worse in the past year	New symptom in past year	Does not apply
[18] Activities of Daily Living				
Needs help with washing and/or bathing				✓
Needs help with dressing				✓
Dresses inappropriately (e.g., back to front, incomplete, inadequately for weather)				✓
Undresses inappropriately (e.g., in public)				✓
Needs help eating (cutting food, mouthful amounts, choking)				✓
Needs help using the bathroom (finding, toileting)				✓
Incontinent (including occasional accidents)				✓
[19] Language & Communication				
Does not initiate conversation				✓
Does not find words				✓ more just difficult to understand him
Does not follow simple instructions				✓
Appears to get lost in middle of conversation				✓
Does not read				✓
Does not write (including printing own name)	✓			can write name + few words
[20] Sleep-Wake Change Patterns				
Excessive sleep (sleeping more)				✓
Inadequate sleep (sleeping less)				sleeps less @ night
Wakes frequently at night	✓			
Confused at night				✓
Sleeps during the day more than usual				✓
Wanders at night				✓
Wakes earlier than usual	✓			
Sleeps later than usual				✓
[21] Ambulation				
Not confident walking over small cracks, lines on the ground, patterned flooring, or uneven surfaces	✓			✓
Unsteady walk, loses balance				✓
Falls				✓
Requires aids to walk				✓

NTG-EDSD

Instructions: For each question block, check the item that best applies to the individual or situation.

	Always been the case	Always but worse in the past year	New symptom in past year	Does not apply
[22] Memory				
Does not recognize familiar persons (staff/relatives/friends)				✓
Does not remember names of familiar people				✓
Does not remember recent events (in past week or less)				✓
Does not find way in familiar surroundings				✓
Loses track of time (time of day, day of the week, seasons)				✓
Loses or misplaces objects	occas	occas		
Puts familiar things in wrong places				✓
Problems with printing or signing own name	✓			
Problems with learning new tasks or names of new people				✓
[23] Behavior and Affect				
Wanders				✓
Withdraws from social activities			✓	
Withdraws from people			✓	
Loss of interest in hobbies and activities			✓	
Seems to go into own world				✓
Obsessive or repetitive behavior	✓			
Hides or hoards objects	✓			
Does not know what to do with familiar objects				✓
Increased impulsivity (touching others, arguing, taking things)				✓
Appears uncertain, lacks confidence				✓
Appears anxious, agitated, or nervous				✓
Appears depressed	✓			
Shows verbal aggression				✓
Shows physical aggression				✓
Temper tantrums, uncontrollable crying, shouting				✓
Shows lethargy or listlessness				✓
Calls to self	✓			
[24] Notable Significant Changes Observed by Others				
In personality (e.g., subdued when was outgoing)		✓		
In friendliness (e.g., now socially unresponsive)		✓		
In attentiveness (e.g., misses cues, distracted)				✓
In weight (e.g., weight loss or weight gain)				✓
In abnormal voluntary movements (head, neck, limbs, trunk)				✓

Case: Mr. H (continued)

- 40 year old man with no prior history of dementia but possible confounding co-occurring medical conditions
- Presents with caregiver reported changes in multiple domains on NTG screen

= Positive Screen

→ EHR documentation: *"Based on Mr. H's clinical evaluation and NTG-EDSD score, his screening results indicate that Mr. H may have some symptoms which could represent early signs of dementia."*

1. Referred back to ENT for HNS evaluation
2. Referred for audiology (last done 3 years ago)
3. Routine blood tests for AD protocol (TSH, cbc, iron studies, Vitamin B12)
4. Referred for baseline neuropsychological testing
5. Provided printed NDSS resources to family

Nutritional Considerations for Individuals with DS-AD or DS-Dementia

Amanda Laezza, RD, LDN & Dominica Nichols PhD, RD, LDN
Registered Dietitians at MGH Down Syndrome Program
Massachusetts General Hospital

Massachusetts General Hospital
Founding Member, Mass General Brigham

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Changes in **food routines** might be the first place that familiar caregivers notice cognitive change, especially if there are no other reasonable explanations for these changes.

www.istockphoto.com

Red Flags in Feeding and Nutrition

- new schedule/routine changes
- new concerns with chewing or swallowing, or fine motor skills
- frustration with choosing foods, unsure what foods are preferred foods
- increased caregiver concerns with mealtime safety

www.discovermagazine.com

Nutrition Status Changes

- unplanned weight loss (Fleming et al. 2024) and/or observed muscle loss ie. cachexia (Minaglia et al. 2019)

- bone density loss (Bettis et al. 2021)
- protein/calorie malnutrition (Loda et al. 2024)
- dehydration (Beck et al. 2021, Liska et al. 2019)

www.specialstrong.com

"***Malnutrition*** *is associated with adverse health outcomes, including faster cognitive and functional decline.*" (Loda et al. 2024)

Malnutrition is not defined by BMI or changes in BMI alone.

- Micronutrient malnutrition can lead to "adverse health outcomes" without changes in overall body appearance.
- Macronutrient deficiencies can also lead to "adverse health outcomes."

www.wayfair.com

Registered Dietitian (RD) tool kit:

The scope of practice of the RD includes:

1. Nutrition-focused physical exam
2. Collection and analysis of diet recall and food frequency report
3. Collection of caregiver feedback including report of mealtime behaviors
4. Review and interpretation of critical lab values

The scope of practice of the RD does *not* include:

1. Assessment of chewing and swallowing (efficiency or safety)

Goals of Nutrition in DS-AD

It is important to support **adequate nutrition** to maintain weight status and lean tissue: calories, protein, fat and fluid, micronutrients (vitamins, minerals).

Common interventions in collaboration with a **nutrition expert** (RD) and/or **SLP**:

1. discussing MIND diet and adequate nutrition with patients and families BEFORE the onset of clinically-relevant cognitive changes
2. adding key (accepted) ingredients to food plan
3. adding an oral nutritional supplement or food additive (Duocal, Polycal, BeneProtein)
4. changing food and drink texture (using IDDSI standards) to encourage intake in the least restrictive way with the dual goals of safety and QoL

Nutrition References

Fleming V, Helsel BC, Ptomey LT, Rosas HD, Handen B, Laymon C, Christian BT, Head E, Mapstone M, Lai F, Krinsky-McHale S, Zaman S, Ances BM, Lee JH, Hartley SL; Alzheimer's Biomarker Consortium –Down Syndrome (ABC-DS) Consortium. Weight Loss and Alzheimer's Disease in Down Syndrome. J Alzheimers Dis. 2023;91(3):1215-1227. doi: 10.3233/JAD-220865. PMID: 36565120; PMCID: PMC9940268.

Minaglia C, Giannotti C, Boccardi V, Mecocci P, Serafini G, Odetti P, Monacelli F. Cachexia and advanced dementia. J Cachexia Sarcopenia Muscle. 2019 Apr;10(2):263-277. doi: 10.1002/jcsm.12380. Epub 2019 Feb 22. PMID: 30794350; PMCID: PMC6463474.

Bettis T, Kim BJ, Hamrick MW. Impact of muscle atrophy on bone metabolism and bone strength: implications for muscle-bone crosstalk with aging and disuse. Osteoporos Int. 2018 Aug;29(8):1713-1720. doi: 10.1007/s00198-018-4570-1. Epub 2018 May 18. PMID: 29777277; PMCID: PMC7861141.

Loda I, D'Angelo E, Marzetti E, Kerminen H. Prevention, Assessment, and Management of Malnutrition in Older Adults with Early Stages of Cognitive Disorders. *Nutrients*. 2024; 16(11):1566. https://doi.org/10.3390/nu16111566

Beck AM, Seemer J, Knudsen AW, Munk T. Narrative Review of Low-Intake Dehydration in Older Adults. Nutrients. 2021 Sep 9;13(9):3142. doi: 10.3390/nu13093142. PMID: 34579019; PMCID: PMC8470893.

Liska D, Mah E, Brisbois T, Barrios PL, Baker LB, Spriet LL. Narrative Review of Hydration and Selected Health Outcomes in the General Population. *Nutrients*. 2019; 11(1):70. https://doi.org/10.3390/nu11010070

Books to Check Out

Kelli McGrane, MS, RD. **MIND DIET for Beginners:** 85 Recipes and a 7-day Kick-Start Plan to Boost Your Brain Health https://www.amazon.com/MIND-Diet-Beginners-Recipes-Kickstart/dp/1647398185

Luna Regina and Kelli McGrane, MS, RD. **Healthy Eating 101:** 45 Effortless Recipes to Step up Your Diet Game: https://www.barnesandnoble.com/w/healthy-eating-101-luna-regina/1133504497

Psychosocial Considerations for Individuals with DS-AD or DS-Dementia

Caroline Bregman, LICSW
Clinical Social Worker at MGH Down Syndrome Program
Massachusetts General Hospital

Massachusetts General Hospital
Founding Member, Mass General Brigham

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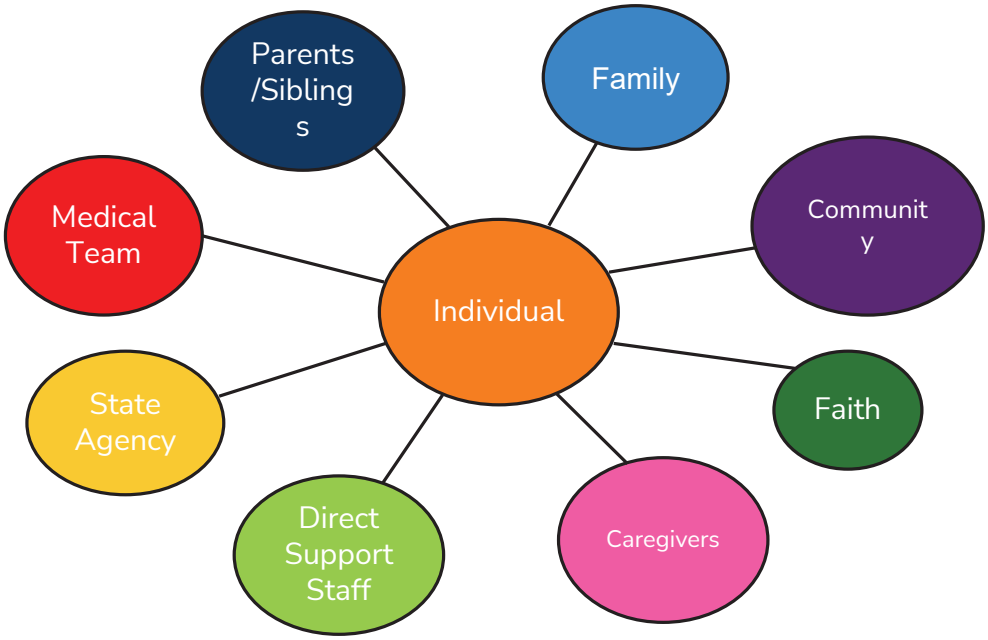
Psychosocial Assessment

What is the person's circle of support?



Education/Anticipatory Guidance

- Mental health considerations, treat or rule out if needed.
- Behavior changes and support
- Safety Planning
- Home Modifications?

Communication

"It's not what you say, it's how you say it"

- Try to avoid arguing or correcting the person's reality, gently respond and use 'therapeutic fiblets' to diffuse a situation.
- Yes/no questions vs open ended questions
- Try to maintain a calm environment
- Anticipate cues and needs
- Use visual supports

Caregiver Support

- What is the family/caregiver support system?
- Unique needs of aging parents providing caregiving support, who may also be experiencing aging/dementia at the same time.

“Widening the Support Network”

- Respite - formal and informal Supports
- State Agency (DDS)
- Aging Services Access Points
- Caregiver Support groups
- Alzheimer’s Association

Advanced Care Planning

- Guardianship/HCP/Supported Decision Making
- Advanced Directives/MOLST/DNR/DNI - Authorization to sign for orders - know your state’s rules so you can provide anticipatory guidance
- SNT/ABLE Accounts
- Does the individual have the ability to age in place in their current setting?
- Goals of care/values
- How to preserve the dignity, choice, wishes, and preferences of the individual
- End of life care - palliative care, hospice, comfort care options

Grief and Bereavement

- Honoring the person, the caregivers, and caregiving team

Resources

National Down Syndrome Society Publications- link: https://ndss.org/publications

· *Caregiving and Down Syndrome: A Companion Guidebook to aging and Down Syndrome:* A health and wellbeing guidebook. Provides guidance, information, and support to families and caregivers of older adults with Down syndrome. Prepare families and caregivers with the key areas of support needed to navigate adult life and aging for individuals with Down syndrome. Empower families and caregivers with information and resources to take proactive, team-based steps to address the needs of an adult with Down syndrome over the course of their lifespan. Provide advocacy and self-care tips for families and caregivers.

· *Aging and Down Syndrome: A Health & Well-Being Guidebook:* This guidebook is a resource for families and caregivers of adults with Down syndrome. It provides accurate information and education about what to anticipate as a part of growing older, so they can set the stage for successful aging. It is intended to be used by various learners: families, professionals, direct caregivers or anyone concerned with the general welfare of someone with Down syndrome.

· *Alzheimer's Disease & Down Syndrome: A Practical Guidebook for Caregivers:* This booklet was written to help empower families and caregivers with knowledge about the connection between Down syndrome and Alzheimer's disease, suggestions about how to carefully and thoughtfully evaluate changes that may be observed with aging, and guidance about how to adapt and thrive within an ever-changing caregiving role when a diagnosis is made.

Resources

End of Life and Down Syndrome, A Companion Guidebook to Aging and Down Syndrome: A Health and Well-Being Guidebook: National Down Syndrome Society, in partnership with The National Task Group on Intellectual Disabilities and Dementia Practices (NTG) and the Alzheimer's Association, released a new guidebook with the aim to provide guidance for the crossroads and decisions that arise in late life and at the end of life. This also offers a proactive advice about support and planning across the lifespan

Alzheimer's Association Family Care Guide: A Guide for Families Caring for Someone with Alzheimer's Disease or a Related Dementia: this guide shares best practices around communication, daily living, understanding behaviors, safety considerations, planning ahead, and caregiver support. Link: https://www.alz.org/media/manh/documents/alzheimer_s-family-care-guide-(fcg).pdf

National Institute on Aging Home Safety Checklist for Alzheimer's Disease: Use the following room-by-room checklist to alert you to potential hazards and to record any changes you need to make to help keep a person with dementia safe. You can buy products or gadgets necessary for home safety at stores carrying hardware, electronics, medical supplies, and children's items. Link: https://www.nia.nih.gov/health/home-safety-checklist-alzheimers-disease

Considering Brain Donation

Brian Skotko, MD, MPP
Emma Campbell Endowed Chair on Down Syndrome
Director, Down Syndrome Program
Massachusetts General Hospital
Professor of Pediatrics, Harvard Medical School

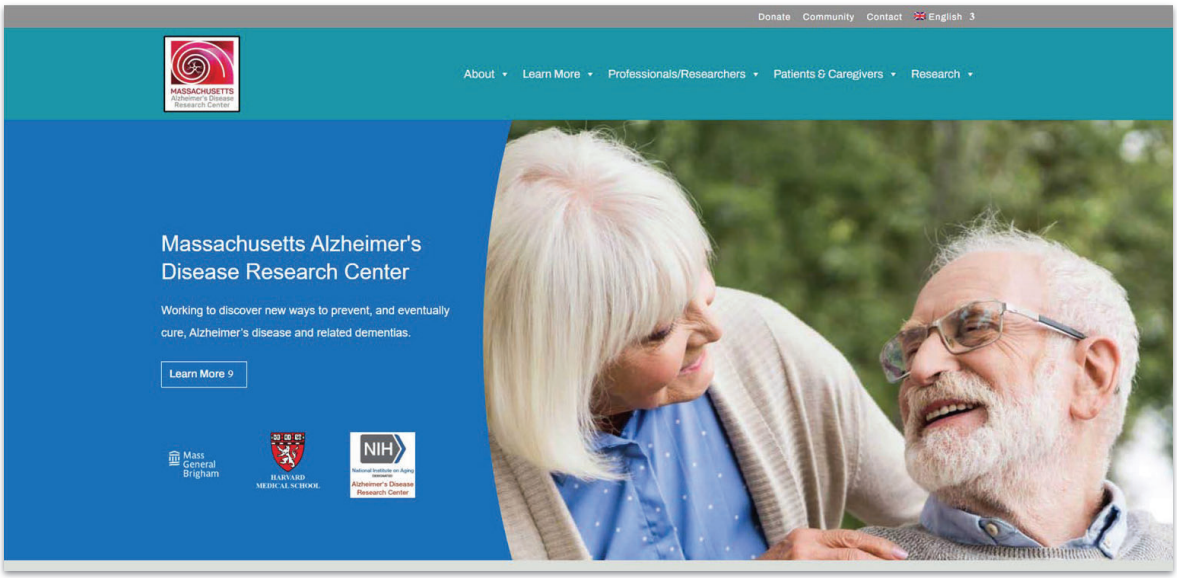
Massachusetts General Hospital
Founding Member, Mass General Brigham

HARVARD
MEDICAL SCHOOL

Reasons for brain donation

- Valuable gift that can provide tissue for numerous research studies
- Especially important as scientists begin to unravel the mysteries of Alzheimer's disease in people with Down syndrome
- Such a donation can enhance our ability to treat and perhaps someday cure Alzheimer's disease in people with Down syndrome

We have partnered with MADRC



www.madrc.org

Handout is available for families

The Importance of Brain Donation

Brain donation, at the time of death, is the most generous gift a patient and their family can give to research. Being able to examine brain tissue from a post-mortem donor helps scientists discover new ways to treat and cure Alzheimer's disease and related dementias.

Most diseases that affect the brain can only be diagnosed with certainty by the examination of brain tissue after death. Many families want to know for sure what condition affected their relative. This is especially important when more than one person in the family has suffered from the disease.

One donated brain can provide tissue for hundreds of research studies. Thus, providing the gift of hope for future generations at risk.

Mass General Brigham

MASSACHUSETTS Alzheimer's Disease Research Center

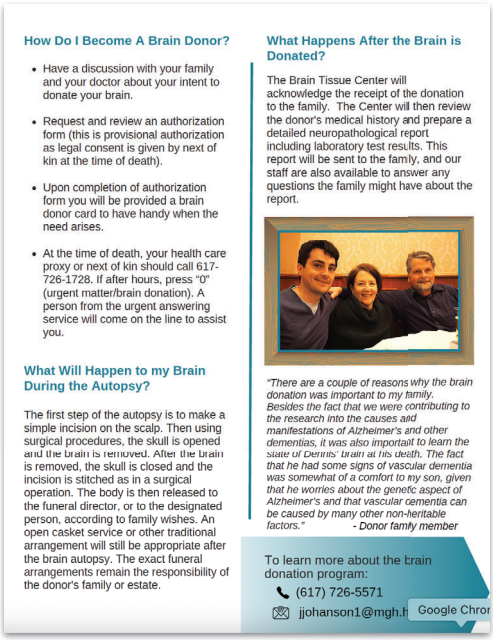
Who Can Donate Their Brain?

The Massachusetts Alzheimer's Disease Research Center (MADRC) has a brain donation program at Massachusetts General Hospital (MGH) for people enrolled in our research studies and other patients in the Mass General Brigham system.

Healthy donors - without memory or other cognitive problems - are also welcome to donate their brains. Information from these donors helps identify what changes in the brain are related to normal aging, versus what is associated with Alzheimer's disease or other dementias.

Is There a Cost for the Brain Autopsy Procedure?

Generally, not as long as the donor has been a patient or research participant at Mass General Brigham. However, if the individual is not a participant in our clinical trials or other specific research studies, the family may need to pay for transportation of the body, and for brain removal and transportation if the autopsy is performed elsewhere.



How Do I Become A Brain Donor?

- Have a discussion with your family and your doctor about your intent to donate your brain.
- Request and review an authorization form (this is provisional authorization as legal consent is given by next of kin at the time of death).
- Upon completion of authorization form you will be provided a brain donor card to have handy when the need arises.
- At the time of death, your health care proxy or next of kin should call 617-726-1728. If after hours, press "0" (urgent matter/brain donation). A person from the urgent answering service will come on the line to assist you.

What Will Happen to my Brain During the Autopsy?

The first step of the autopsy is to make a simple incision on the scalp. Then using surgical procedures, the skull is opened and the brain is removed. After the brain is removed, the skull is closed and the incision is stitched as in a surgical operation. The body is then released to the funeral director, or to the designated person, according to family wishes. An open casket service or other traditional arrangement will still be appropriate after the brain autopsy. The exact funeral arrangements remain the responsibility of the donor's family or estate.

What Happens After the Brain is Donated?

The Brain Tissue Center will acknowledge the receipt of the donation to the family. The Center will then review the donor's medical history and prepare a detailed neuropathological report including laboratory test results. This report will be sent to the family, and our staff are also available to answer any questions the family might have about the report.

"There are a couple of reasons why the brain donation was important to my family. Besides the fact that we were contributing to the research into the causes and manifestations of Alzheimer's and other dementias, it was also important to learn the state of Dennis' brain at his death. The fact that he had some signs of vascular dementia was somewhat of a comfort to my son, given that he worries about the genetic aspect of Alzheimer's and that vascular dementia can be caused by many other non-heritable factors." *- Donor family member*

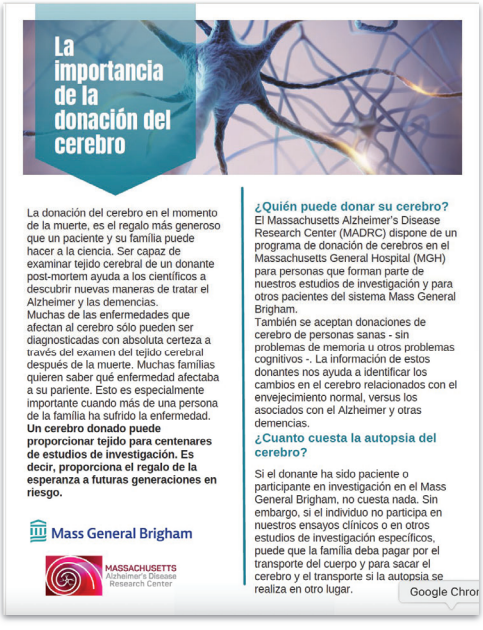
To learn more about the brain donation program:

(617) 726-5571

jjohanson1@mgh.h

Google Chro

Handout is available in Spanish, too



La importancia de la donación del cerebro

La donación del cerebro en el momento de la muerte, es el regalo más generoso que un paciente y su familia puede hacer a la ciencia. Ser capaz de examinar tejido cerebral de un donante post-mortem ayuda a los científicos a descubrir nuevas maneras de tratar el Alzheimer y las demencias.
Muchas de las enfermedades que afectan al cerebro sólo pueden ser diagnosticadas con absoluta certeza a través del examen del tejido cerebral después de la muerte. Muchas familias quieren saber qué enfermedad afectaba a su pariente. Esto es especialmente importante cuando más de una persona de la familia ha sufrido la enfermedad.
Un cerebro donado puede proporcionar tejido para centenares de estudios de investigación. Es decir, proporciona el regalo de la esperanza a futuras generaciones en riesgo.

Mass General Brigham

MASSACHUSETTS Alzheimer's Disease Research Center

¿Quién puede donar su cerebro?

El Massachusetts Alzheimer's Disease Research Center (MADRC) dispone de un programa de donación de cerebros en el Massachusetts General Hospital (MGH) para personas que forman parte de nuestros estudios de investigación y para otros pacientes del sistema Mass General Brigham.
También se aceptan donaciones de cerebro de personas sanas - sin problemas de memoria u otros problemas cognitivos -. La información de estos donantes nos ayuda a identificar los cambios en el cerebro relacionados con el envejecimiento normal, versus los asociados con el Alzheimer y otras demencias.

¿Cuanto cuesta la autopsia del cerebro?

Si el donante ha sido paciente o participante en investigación en el Mass General Brigham, no cuesta nada. Sin embargo, si el individuo no participa en nuestros ensayos clínicos o en otros estudios de investigación específicos, puede que la familia deba pagar por el transporte del cuerpo y para sacar el cerebro y el transporte si la autopsia se realiza en otro lugar.

Google Chro

¿Cómo consigo ser donante de cerebro?

- Hable con su familia y con su médico sobre su intención de donar su cerebro.
- Pida y revise un formulario de autorización (esto es una autorización provisional de consentimiento legal que da a su pariente más cercano en el momento de la muerte).
- Después de completar el formulario de autorización se le suministrará una targeta de donante de cerebro para tener a mano cuando sea necesario.
- En el momento de la muerte, su apoderado de atención médica o su pariente más cercano debe llamar al 617-726-1728. Si es fuera del horario laboral, marque "0" (cuestión urgente/donación de cerebro). Una persona del servicio de respuesta urgente le asistirá.

¿Qué le sucederá a mi cerebro durante la autopsia?

El primer paso de la autopsia es realizar una incisión simple en el cuero cabelludo. Después, con procedimientos quirúrgicos, se abre el cráneo y se saca el cerebro. Después de sacar el cerebro, se cierra el cráneo y se dan puntos en la incisión como en una operación quirúrgica. El cuerpo se entrega al director del funeral o a la persona designada, siguiendo los designios de la familia. Después de la autopsia del cerebro, continúa siendo apropiado un servicio con el ataúd abierto u otra disposición tradicional. Las disposiciones exactas del funeral continúan siendo responsabilidad de la familia del donante o del estado.

¿Qué sucede después que se haya donado el cerebro?

El centro de tejido cerebral dará un acuse de recibo de la donación a la familia. Después, el centro revisará el historial médico del donante y preparará un informe neuropatológico detallado que incluya resultados de pruebas de laboratorio. Este informe se enviará a la familia, y nuestro personal está también disponible para contestar cualquier pregunta que la familia pueda tener en referencia al informe.

"Hay un par de motivos por los que la donación del cerebro era importante para mi familia. Aparte del hecho de contribuir en la investigación de las causas y manifestaciones del Alzheimer y otras demencias, también era importante saber el estado del cerebro de Dennis cuando murió. El hecho de tener algunos signos de demencia vascular fue reconfortante para mi hijo, puesto que está preocupado sobre el aspecto genético del Alzheimer y la demencia vascular puede ser causada por otros factores no hereditarios."

- Familiar de un donante

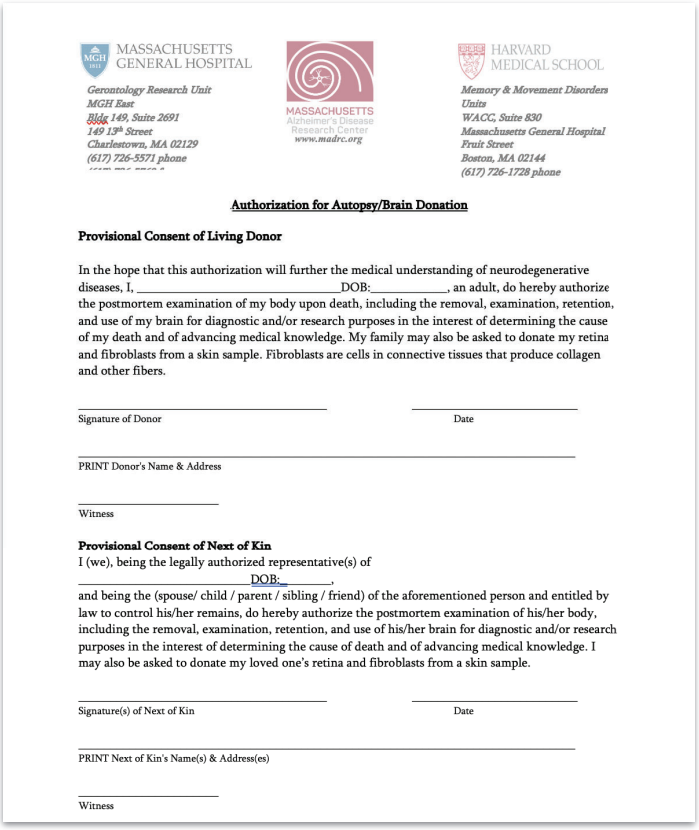
Para saber más sobre el programa de donación de cerebros:

(617) 726-5571

jjohanson1@mgh.harvard.edu

Some logistical considerations

- All major **religions support organ and tissue donation**. However, since brain donation is a unique gift, we encourage people to talk to their religious leaders if they have questions.
- When the brain is donated to the MGH Brain Tissue Center, there are **no restrictions on the donation of other organs**, which can be facilitated at time of autopsy.
- In Massachusetts, a living person can signal intent to donate, but the deceased next of kin or legal representative must provide **consent**.
- The brain donation and autopsy have no costs, but family is responsible for **transportation costs** of the body
- An **open casket service** is still possible after brain donation.



MASSACHUSETTS GENERAL HOSPITAL

Gerontology Research Unit
MGH East
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(617) 726-5571 phone

MASSACHUSETTS Alzheimer's Disease Research Center
www.madrc.org

HARVARD MEDICAL SCHOOL

Memory & Movement Disorders Units
WACC, Suite 830
Massachusetts General Hospital
Fruit Street
Boston, MA 02144
(617) 726-1728 phone

Authorization for Autopsy/Brain Donation

Provisional Consent of Living Donor

In the hope that this authorization will further the medical understanding of neurodegenerative diseases, I, _____DOB:_____, an adult, do hereby authorize the postmortem examination of my body upon death, including the removal, examination, retention, and use of my brain for diagnostic and/or research purposes in the interest of determining the cause of my death and of advancing medical knowledge. My family may also be asked to donate my retina and fibroblasts from a skin sample. Fibroblasts are cells in connective tissues that produce collagen and other fibers.

_____ _____
Signature of Donor Date

_____
PRINT Donor's Name & Address

_____
Witness

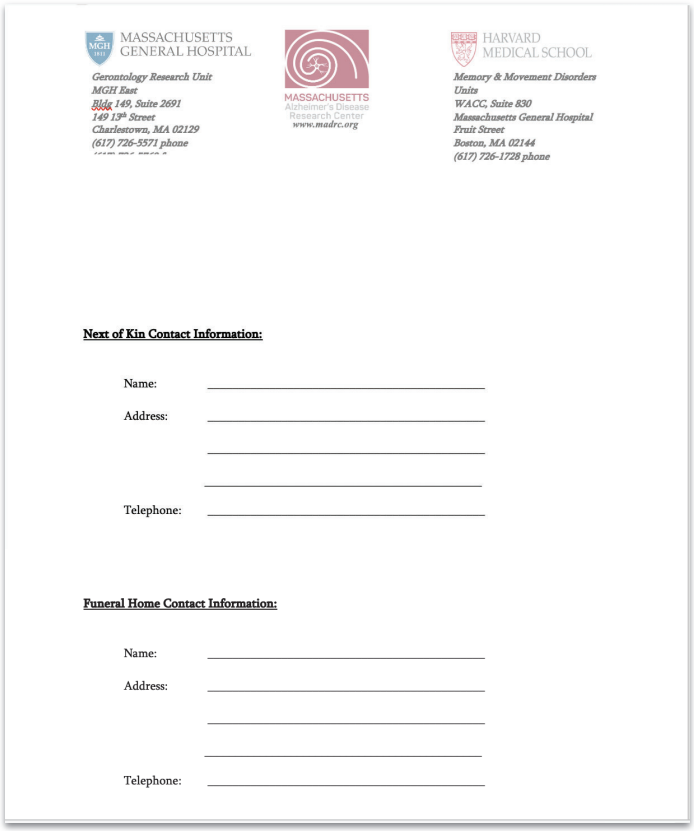
Provisional Consent of Next of Kin

I (we), being the legally authorized representative(s) of _____DOB:_____, and being the (spouse/ child / parent / sibling / friend) of the aforementioned person and entitled by law to control his/her remains, do hereby authorize the postmortem examination of his/her body, including the removal, examination, retention, and use of his/her brain for diagnostic and/or research purposes in the interest of determining the cause of death and of advancing medical knowledge. I may also be asked to donate my loved one's retina and fibroblasts from a skin sample.

_____ _____
Signature(s) of Next of Kin Date

_____
PRINT Next of Kin's Name(s) & Address(es)

_____
Witness



MASSACHUSETTS GENERAL HOSPITAL

Gerontology Research Unit
MGH East
Bldg 149, Suite 2691
149 13th Street
Charlestown, MA 02129
(617) 726-5571 phone

MASSACHUSETTS Alzheimer's Disease Research Center
www.madrc.org

HARVARD MEDICAL SCHOOL

Memory & Movement Disorders Units
WACC, Suite 830
Massachusetts General Hospital
Fruit Street
Boston, MA 02144
(617) 726-1728 phone

Next of Kin Contact Information:

Name: _____

Address: _____

_____

_____

Telephone: _____

Funeral Home Contact Information:

Name: _____

Address: _____

_____

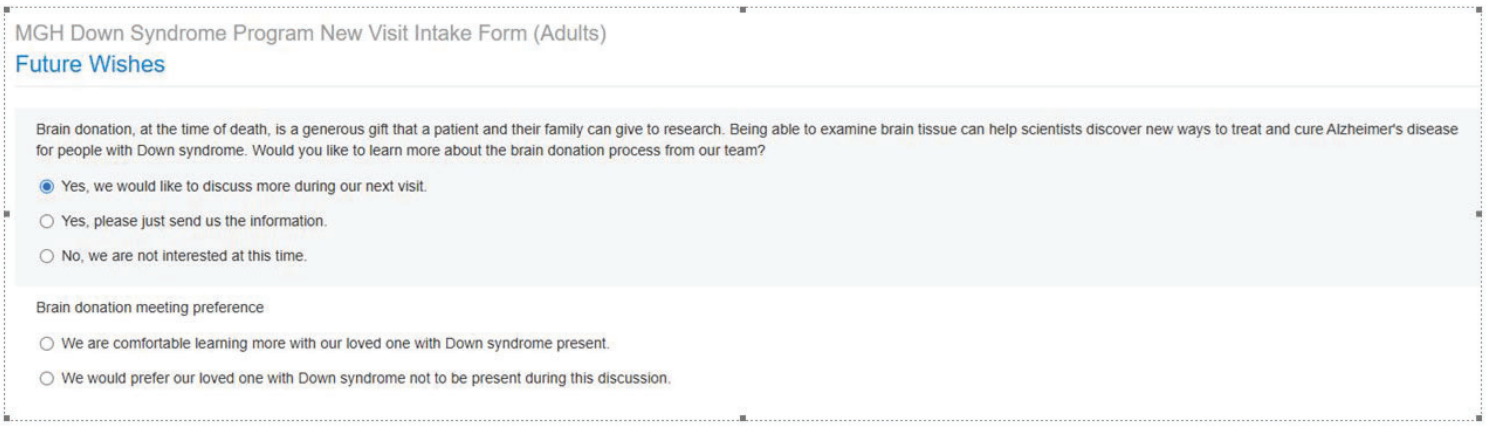
_____

Telephone: _____

Autopsy and brain donation

- The MGH Brain Tissue Center will acknowledge the receipt of the donation to the family.
- The Center will review the donor's medical history and prepare a detailed neuropathological report.
- This report will be sent to the family and the clinical team.
- Researchers, worldwide, who have IRB-approved studies can contact the MGH Brain Tissue Center to request samples.

Step 1: ask family's interest on our intake form



MGH Down Syndrome Program New Visit Intake Form (Adults)

Future Wishes

Brain donation, at the time of death, is a generous gift that a patient and their family can give to research. Being able to examine brain tissue can help scientists discover new ways to treat and cure Alzheimer's disease for people with Down syndrome. Would you like to learn more about the brain donation process from our team?

- (●) Yes, we would like to discuss more during our next visit.
- ( ) Yes, please just send us the information.
- ( ) No, we are not interested at this time.

Brain donation meeting preference

- ( ) We are comfortable learning more with our loved one with Down syndrome present.
- ( ) We would prefer our loved one with Down syndrome not to be present during this discussion.

Step 2: “send me information”

- The patient’s physician or social worker will either (a) send an e-mail to the family or (b) send a Patient Gateway message in Epic to the family.
- We developed a smart phrase in Epic to streamline our processes.
- If a family then responds that they would like to proceed with indicating an **intent** to donate, they should complete the "Authorization for Autopsy/Brain Donation."

Step 2: “discuss during next visit”

- The patient’s doctor and/or social worker should discuss who and how to best structure the clinical visit. Every family and patient is different, and we will defer to the clinical team to customize what works best.
- Families should be given (1) the two-page flyer in English or Spanish and (2) the Q&A sheet, available in English.
- For families who wish to proceed with indicating an **intent** to donate, they should complete the "Authorization for Autopsy/Brain Donation."
- If they give us the Authorization form, we then send this to the MGH Brain Donation Program
- Once the Brain Donation receives the Authorization form, they will follow-up with the family directly with confirmation letter and with the Brian Donation card.

Step 2: after death

- Upon death, the family **still needs to call** the MGH Brain Donation Program at 617-726-5571 who will handle the logistics from there.
- All costs are covered EXCEPT for the transportation of the body, which will be a cost that is billed to the family from the funeral home.

Take-home messages

- We spent more than one year working out the logistics with our Brain Donation center within our hospital.
- We then spent time as a team thinking about how best to incorporate the information thoughtfully for families.
- This coming year, we will be evaluating how the process goes with our families.
- In short, we hope that this will give patients and families, who are interested in brain donation, an opportunity to contribute to future scientific discoveries for people with Down syndrome.

Contact information for MGH Down Syndrome Program team

- **E-mail:** downsyndrome@partners.org
- **Phone**: 617-643-8912
- **Facebook:** https://www.facebook.com/MGHDownSyndrome/
- **Twitter**: https://x.com/MGHDownSyndrome
- **E-Newsletters**: http://eepurl.com/dAJGu1
- **Web page**: www.massgeneral.org/downsyndrome