NACC UNIFORM DATA SET

DOWN SYNDROME MODULE
Coding Guidebook Follow-up Visit Packet

UDS Version 3.0, March 2015
DS Module Version 1.0, August 2022

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Down Syndrome Module to the Uniform Data Set

Coding Guidebook for the Follow-up Visit Packet

INTRODUCTION

The Down Syndrome Module to the UDS is designed for:

- Persons with Down Syndrome

How to read the Guidebook

The Guidebook features a reproduction of each form in the DS Module, interspersed with explanatory notes and references. Throughout this document, all explanatory and reference text are on a white background.

Important notes

- ***Timing —*** The DS Module evaluation is intended to be completed as part of a UDS visit. If the UDS evaluation and the DS evaluation are separated into two days, please complete the DS evaluation within two weeks of the UDS evaluation.
- ***Visit Number —*** Even when the visit is split into two days, the same Visit Number MUST be used in the form header on all forms in both packets (UDS and DS) from both days.
- ***IVP vs. FVP —*** When a UDS enrollee is being given the DS Module evaluation for the first time, you should use the DS Module Initial Visit Packet, even if you are using the UDS Follow-up Visit Packet.

Form A1D: Participant Health History

INSTRUCTIONS: This form is to be completed by intake interviewer based on ADRC scheduling records, subject interview, medical records, and proxy co-participant report (as needed). Check only <u>one</u> box per question.

1. What are the participant's weekday activities?	
1a. Day program	☐ 0 No ☐ 1 Yes ☐ 9 Unknown
1b. Workshops	☐ 0 No ☐ 1 Yes ☐ 9 Unknown
1c. Stays at home	☐ 0 No ☐ 1 Yes ☐ 9 Unknown
1d. Community paid job	☐ 0 No ☐ 1 Yes ☐ 9 Unknown
1e. Other (SPECIFY): _____	☐ 0 No ☐ 1 Yes ☐ 9 Unknown
2. Age of participant's mother at participant's birth	____ ____ ____ *(777 = provided at previous visit; 999 = unknown)*

QUESTION 2: If the exact age of the participant's mother at the time of the participant's birth is unknown, ask the participant and/or co-participant to estimate. If s/he cannot estimate, enter **999 = Unknown**. If provided at a previous visit, enter **777 = provided at previous visit**.

QUESTIONS 3–7: For the following questions, record the presence or absence of a history of these congenital heart conditions at this visit, as determined by the clinician's best judgment following the medical history interview with the participant and/or co-participant.

A condition should be considered…		
Absent	IF	… it is not indicated by information obtained from the participant or co-participant interview
Present – resolved	IF	… it existed in the past but was resolved or there is no treatment currently under way
Present – repaired	IF	… it existed in the past and was repaired but not resolved or still requires active management
Present – unrepaired	IF	… it exists, was never repaired, or still requires active management
Unknown	IF	… there is insufficient information available from the participant or co-participant interview

3. Congenital heart disease — atrial septal defect	☐ 0 Absent ☐ 1 Present — resolved ☐ 2 Present — repaired ☐ 3 Present — unrepaired ☐ 9 Unknown
4. Congenital heart disease — ventricular septal defect	☐ 0 Absent ☐ 1 Present — resolved ☐ 2 Present — repaired ☐ 3 Present — unrepaired ☐ 9 Unknown
5. Congenital heart disease — atrioventricular (AV) canal defect	☐ 0 Absent ☐ 1 Present — resolved ☐ 2 Present — repaired ☐ 3 Present — unrepaired ☐ 9 Unknown
6. Congenital heart disease — tetralogy of Fallot	☐ 0 Absent ☐ 1 Present — resolved ☐ 2 Present — repaired ☐ 3 Present — unrepaired ☐ 9 Unknown
7. Congenital heart disease — other (SPECIFY): _____	☐ 0 Absent ☐ 1 Present — resolved ☐ 2 Present — repaired ☐ 3 Present — unrepaired ☐ 9 Unknown

QUESTIONS 8 – 34: For the following questions, record the presence or absence of a history of these conditions at this visit, as determined by the clinician's best judgment following the medical history interview with the participant and/or co-participant.

A condition should be considered…

Absent	IF	… it is not indicated by information obtained from the participant or co-participant interview
Recent/active	IF	… it happened within the last year or still requires active management and is consistent with information obtained from the participant and co-participant interview
Remote/inactive	IF	… it existed or occurred in the past (more than one year ago) but was resolved or there is no treatment currently under way
Unknown	IF	… there is insufficient information available from the participant or co-participant interview

8. Cardiovascular disease — hypotension	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
9. Cardiovascular disease — syncope	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
10. Pulmonary disease — pneumonia/aspiration	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
11. Hepatic conditions	☐ 1 Hepatitis B carrier ☐ 2 Hepatitis B infected ☐ 3 Hepatitis B immune ☐ 4 Had hepatitis B vaccine ☐ 9 Unknown

12. Dermatologic conditions — rosacea	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
13. Dermatologic conditions — alopecia	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
14. Dermatologic conditions — psoriasis	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
15. Musculoskeletal conditions — osteoporosis/osteopenia	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
16. Musculoskeletal conditions — gout	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
17. Musculoskeletal conditions — atlanto-axial subluxation	☐ 0 Absent ☐ 1 Present ☐ 9 Unknown
18. Musculoskeletal conditions — fractures in the past five years	☐ 0 No ☐ 1 Yes (SPECIFY): _____ ☐ 9 Unknown
19. Endocrine/metabolic conditions — hypothyroidism	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown

20. Endocrine/metabolic conditions — Hashimoto's thyroiditis	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
21. Endocrine/metabolic conditions — hyperthyroidism	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
22. Endocrine/metabolic conditions — currently on thyroid replacement medication	☐ 0 No ☐ 1 Yes (SPECIFY): _____ ☐ 9 Unknown
23. Endocrine/metabolic conditions — vitamin D deficiency	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
24. Menstrual history — has the participant ever menstruated?	☐ 0 No ☐ 1 Yes, active ☐ 2 Yes, menopausal ☐ 9 Unknown/not applicable
25. Menstrual history — age of onset of menses	____ ____ ____ *(777 = provided at previous visit; 888 = not applicable; 999 = unknown)*

QUESTION 25: If the exact age of the participant's onset of menses is unknown, ask the participant and/or co-participant to estimate. If s/he cannot estimate, enter **999=Unknown**. If provided at a previous visit, enter **777 = provided at previous visit**. If the participant has never menstruated, enter **888 = not applicable**.

26. Menstrual history — age of onset of menopause	____ ____ ____ *(777 = provided at previous visit; 888 = not applicable; 999 = unknown)*

QUESTION 26: If the exact age of the participant's onset of menopause is unknown, ask the participant and/or co-participant to estimate. If s/he cannot estimate, enter **999=Unknown**. If provided at a previous visit, enter **777 = provided at previous visit**. If the participant has never menstruated, enter **888 = not applicable**.

27. Hormone replacement therapy — has the participant received HRT?	☐ 0 No ☐ 1 Yes ☐ 9 Unknown
28. Hormone replacement therapy — at what age did HRT begin?	____ ____ ____ *(777 = provided at previous visit; 888 = not applicable; 999 = unknown)*

QUESTION 28: If the exact age that the participant began hormone replacement therapy is unknown, ask the participant and/or co-participant to estimate. If s/he cannot estimate, enter **999=Unknown**. If the participant did not receive hormone replacement therapy, enter **888=Not applicable**. If provided at a previous visit, enter **777 = provided at previous visit**.

29. Hormone replacement therapy — how many years has the participant been on HRT?	☐ 1 1 – 3 years ☐ 2 4 – 6 years ☐ 3 >6 years ☐ 9 Unknown/not applicable
30. Gastrointestinal conditions — celiac disease	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
31. Hematopoietic/lymphatic disease — anemia with iron deficiency	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
32. Hematopoietic/lymphatic disease — anemia with folate deficiency	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
33. Autoimmune conditions — lupus	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown

34. Autoimmune conditions — chronic neutropenia	☐ 0 Absent ☐ 1 Recent/active ☐ 2 Remote/inactive ☐ 9 Unknown
35. Cancer — solid tumor	☐ 0 No ☐ 1 Yes (SPECIFY PRIMARY SITE): _____ ☐ 9 Unknown
36. Cancer — leukemia	☐ 0 No ☐ 1 Yes, childhood transient myeloproliferative disorder ☐ 2 Yes, childhood leukemia ☐ 3 Yes, adult onset leukemia ☐ 9 Unknown

QUESTIONS 37 – 44: For the following questions, if the exact year of the most recent surgical procedure is unknown, ask the participant and/or the co-participant to estimate. If s/he cannot estimate, enter **9999 = Unknown**. If year was entered at a previous visit, enter **7777 = provided at previous visit**.

37. Major surgical procedures — congenital heart-defect repair	☐ 0 No (**SKIP TO QUESTION 38**) ☐ 1 Yes (SPECIFY): (**CONTINUE**) _____ ☐ 9 Unknown (**SKIP TO QUESTION 38**)
37a. Year of most recent congenital heart-defect repair	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
38. Major surgical procedures — adult cardiac surgery	☐ 0 No (**SKIP TO QUESTION 39**) ☐ 1 Yes (SPECIFY): (**CONTINUE**) _____ ☐ 9 Unknown (**SKIP TO QUESTION 39**)
38a. Year of most recent adult cardiac surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
39. Major surgical procedures — spinal surgery	☐ 0 No (**SKIP TO QUESTION 40**) ☐ 1 Yes (SPECIFY): (**CONTINUE**) _____ ☐ 9 Unknown (**SKIP TO QUESTION 40**)

39a. Year of most recent spinal surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
40. Major surgical procedures — lower-extremity orthopedic surgery	☐ 0 No **(SKIP TO QUESTION 41)** ☐ 1 Yes (SPECIFY): **(CONTINUE)** _____ ☐ 9 Unknown **(SKIP TO QUESTION 41)**
40a. Year of most recent lower-extremity orthopedic surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
41. Major surgical procedures — upper-extremity orthopedic surgery	☐ 0 No **(SKIP TO QUESTION 42)** ☐ 1 Yes (SPECIFY): **(CONTINUE)** _____ ☐ 9 Unknown **(SKIP TO QUESTION 42)**
41a. Year of most recent upper-extremity orthopedic surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
42. Major surgical procedures — thyroid surgery	☐ 0 No **(SKIP TO QUESTION 43)** ☐ 1 Yes (SPECIFY): **(CONTINUE)** _____ ☐ 9 Unknown **(SKIP TO QUESTION 43)**
42a. Year of most recent thyroid surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
43. Major surgical procedures — oncology surgery	☐ 0 No **(SKIP TO QUESTION 44)** ☐ 1 Yes (SPECIFY): **(CONTINUE)** _____ ☐ 9 Unknown **(SKIP TO QUESTION 44)**
43a. Year of most recent oncology surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*
44. Major surgical procedures — other surgery	☐ 0 No **(END FORM HERE)** ☐ 1 Yes (SPECIFY): **(CONTINUE)** _____ ☐ 9 Unknown **(END FORM HERE)**
44a. Year of most recent other surgery	____ ____ ____ ____ *(7777 = provided at previous visit; 9999 = unknown)*

Form B1D: NTG-EDSD

The NTG-Early Detection Screen for Dementia, adapted from the DSQIID*, can be used for the early detection screening of those adults with an intellectual disability who are suspected of or may be showing early signs of mild cognitive impairment or dementia. The NTG-EDSD is not an assessment or diagnostic instrument, but an administrative screen that can be used by staff and family caregivers to note functional decline and health problems and record information useful for further assessment, as well as to serve as part of the mandatory cognitive assessment review that is part of the Affordable Care Act's annual wellness visit for Medicare recipients. This instrument complies with Action 2.B of the US National Plan to Address Alzheimer's Disease.

It is recommended that this instrument be used on an annual or as indicated basis with adults with Down syndrome beginning with age 40, and with other at-risk persons with intellectual or developmental disabilities when suspected of experiencing cognitive change. The form can be completed by anyone who is familiar with the adult (that is, has known him or her for over six months), such as a family member, agency support worker, or a behavioral or health specialist using information derived by observation or from the adult's personal record.

The estimated time necessary to complete this form is between 15 and 60 minutes. Some information can be drawn from the individual's medical/health record. Consult the NTG-EDSD Manual for additional instructions (www.aadmd.org/ntg/ screening).

ntg
National Task Group on Intellectual Disabilities and Dementia Practices

NTG-EDSD

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

(7) **Sex:**

	Female
	Male

(8) **Best description of intellectual disability**

	No discernible intellectual disability
	Borderline (IQ 70-75)
	Mild ID (IQ 55-69)
	Moderate ID (IQ 40-54)
	Severe ID (IQ 25-39)
	Profound ID (IQ 24 and below)
	Unknown

(9) **Diagnosed condition (*check all that apply*)**

	Autism
	Cerebral palsy
	Down syndrome
	Fragile X syndrome
	Intellectual disability
	Prader-Willi syndrome
	Other:

Current living arrangement of person:

- ☐ **Lives alone**
- ☐ **Lives with spouse or friends**
- ☐ **Lives with parents or other family members**
- ☐ **Lives with paid caregiver**
- ☐ **Lives in community group home, apartment, supervised housing, etc.**
- ☐ **Lives in senior housing**
- ☐ **Lives congregate residential setting**
- ☐ **Lives in long term care facility**
- ☐ **Lives in other:** _____

(10) **General characteristics of current physical health:**

	Excellent
	Very good
	Good
	Fair
	Poor

(11) **Compared to one year ago, current physical health is:**

	Much better
	Somewhat better
	About the same
	Somewhat worse
	Much worse

(12) **Compared to one year ago, current mental health is:**

	Much better
	Somewhat better
	About the same
	Somewhat worse
	Much worse

(13) **Conditions present (*check all that apply*)**

	Vision impairment
	Blind (very limited or no vision)
	Vision corrected by glasses
	Hearing impairment
	Deaf (very limited or no hearing)
	Hearing corrected by hearing aids
	Mobility impairment
	Not mobile — uses wheelchair
	Not mobile — is moved about in wheelchair

(14) **Significant recent [in past year] life event (*check all that apply*)**

	Death of someone close
	Changes in living arrangement, work, or day program
	Changes in staff close to the person
	New roommate/housemates
	Illness or impairment due to accident
	Adverse reaction to medication or over-medication
	Interpersonal conflicts
	Victimization / abuse
	Other:

(15) **Seizures**

	Recent onset seizures
	Long term occurrence of seizures
	Seizures in childhood, not occurring in adulthood
	No history of seizures

If MCI or dementia is documented, complete 16, 17, & 18

(16) **Diagnostic History**

Mild cognitive impairment [MCI] or dementia previously diagnosed (Dx)?

[ ] No

[ ] Yes, MCI

Date of Dx:

[ ] Yes, dementia

Date of Dx:

Type of dementia:

Diagnosed by:
☐ Geriatrician
☐ Neurologist
☐ Physician
☐ Psychiatrist
☐ Psychologist
☐ Other:

(17) **Reported date of onset of MCI/dementia**
[When suspicion of dementia first arose]
Note approximate year and month:

(18) **Comments / explanations about dementia suspicions:**

"Always has been the case" means the need, problem, or behavior has been present for a very long time.
"Always but worse" means the existing need, problem, or behavior has further declined, requiring more personal assistance.
"New symptom in past year" means this need, problem, or behavior was not present until recently.
"Does not apply" means these needs, problems, or behaviors are not present.

[Check column option as appropriate]

	Always been the case	Always but worse	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	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)				

[Check column option as appropriate]

	[27] Chronic Health Conditions*	Recent condition (past year)	Condition diagnosed in last 5 years	Lifelong condition	Condition not present
	Bone, Joint, and Muscle				
1	Arthritis				
2	Osteoporosis				
	Heart and Circulation				
3	Heart condition				
4	High cholesterol				
5	High blood pressure				
6	Low blood pressure				
7	Stroke				
	Hormonal				
8	Diabetes (type 1 or 2)				
9	Thyroid disorder				
	Lungs/breathing				
10	Asthma				
11	Chronic bronchitis, emphysema				
12	Sleep disorder				
	Mental Health				
13	Alcohol or substance abuse				
14	Anxiety disorder				
15	Attention deficit disorder				
16	Bipolar disorder				
17	Dementia/Alzheimer's disease				
18	Depression				
19	Eating disorder (anorexia, bulimia)				
20	Obsessive-compulsive disorder				
21	Schizophrenia				
22	Other:				
	Pain / Discomfort				
23	Back pain				
24	Constipation				
25	Foot pain				
26	Gastrointestinal pain or discomfort				
27	Headaches				
28	Hip/knee pain				
29	Neck/shoulder pain				
	Sensory				
30	Dizziness / vertigo				
31	Impaired hearing				
32	Impaired vision				
	Other				
33	Cancer — type:				
34	Chronic fatigue				
35	Epilepsy / seizure disorder				
36	Heartburn / acid reflux				
37	Urinary incontinence				
38	Sleep apnea				
39	Tics/movement disorder/spasticity				
40	Dental pain				

*Items drawn from the Longitudinal Health and Intellectual Disability Survey (University of Illinois at Chicago)

(28) **Current Medications**

Yes	No	Indicate type
☐	☐	Treatment of chronic conditions
☐	☐	Treatment of mental health disorders or behavior problems
☐	☐	Treatment of pain

For reviews, attach list of current medications, dosage, and when prescribed

☐ List is attached for reviews

(29) **Comments related to other notable changes or concerns:**

(30) **Next Steps / Recommendations**

☐ Refer to treating physician for assessment

☐ Review internally by clinical personnel

☐ Include in annual review / annual wellness visit

☐ Repeat in _____ months

Acknowledgment: Derived from the DSQIID (*Dementia Screening Questionnaire for Individuals with Intellectual Disabilities; Deb, S., 2007) as adapted into the Southeast PA Dementia Screening Tool (DST) – with the assistance of Carl V. Tyler, Jr., MD – and the LHIDS (Longitudinal Health and Intellectual Disability Survey; Rimmer & Hsieh, 2010) and as further adapted by the National Task Group on Intellectual Disabilities and Dementia Practices as the NTG Early Detection Screen for Dementia for use in the USA.

www.aadmd.org/ntg/screening

Form B2D: DLD Summary Page

The Dementia Questionnaire for People with Learning Disabilities (DLD) must be purchased from Pearson UK: https://www.pearsonclinical.co.uk/store/ukassessments/en/Store/Professional-Assessments/Cognition-%26-Neuro/Dementia-Questionnaire-for-People-with-Learning-Disabilities/p/P100009213.html

The DLD is a Qualification Level B assessment and may be purchased by individuals meeting one of Pearson's qualification requirements. Further information about assessment qualification codes can be found at https://www.pearsonclinical.co.uk/ordering/how-to-order/qualifications/qualifications-policy.html

Purchases on behalf of a qualified user should follow the guidance at https://www.pearsonclinical.co.uk/ordering/how-to-order/qualifications/purchasing-on-behalf-of-a-qualified-user.html

Ordering questions should be directed to Pearson UK (https://www.pearsonclinical.co.uk/contact-us.html).

Summary: Dementia Questionnaire for People With Learning Disabilities

Category → / Totals ↓	1 Short-term memory	2 Long-term memory	3 Spatial & temporal orientation	4 Speech	5 Practical skills	6 Mood	7 Activity & interest	8 Behavioral disturbance
Page 2					X	X	X	X
Page 3	X			X		X		
Page 4		X				X		
Page 5		X	X			X		X
Page 6					X	X		
Page 7		X	X	X	X			
Page 8	X			X			X	
Page 9	X			X	X		X	X
Page 10	X	X	X	X			X	X
Category total								

SCS = Sum of cognitive scores (1 – 3): ☐

SOS = Sum of social scores (4 – 8): ☐

Form C1D: Neuropsychological Battery Scores

INSTRUCTIONS: This form should be completed by ADRC or clinic staff.

KEY: If the subject cannot complete any of the following exams, please give the reason by entering one of the following codes: 95 = Physical problem; 96 = Cognitive/behavior problem; 97 = Other problem; 98 = Verbal refusal

1. Down Syndrome Mental Status Examination (DSMSE)

1. Was any part of the DSMSE administered?

0 ☐ No (If No, enter reason code, 95 – 98: ___ ___ and **SKIP TO QUESTION 2**)

1 ☐ Yes (If Yes, **CONTINUE**)

			TOTALS
Personal information IA+IIIA+IIIB 1a1. ___ *(0 – 7; 8=Not assessed)* Season/day IIIC+IIID 1b1. ___ *(0 – 4; 8=Not assessed)*			1b2. ___ ___ *(0 – 11; 88=Not assessed)* (1a1 + 1b1)
Shoebox Memory			
Object IIA	Immediate 1c1. ___ ___ *(0 – 9; 88=Not assessed)*	VA Delay 1c2. ___ ___ *(0 – 9;88=Not assessed)*	1c3. ___ ___ *(0 – 18; 88=Not assessed)* (1c1 + 1c2)
Memory			
Place VIIA	Immediate 1d1. ___ *(0 – 3; 8=Not assessed)*	XA Delay 1d2. ___ *(0 – 3; 8=Not assessed)*	1d3. ___ *(0 – 6; 8=Not assessed)* (1d1 + 1d2)
Apraxia			
Intransitive XIA 1e1. ___ *(0 – 2; 8=Not assessed)* Transitive XIB 1e2. ___ . ___ *(0.0 – 2.0; 8.8=Not assessed)*			1e3. ___ . ___ *(0.0 – 4.0; 8.8=Not assessed)* (1e1 + 1e2)
Language			
Naming VIA+VIIIA 1f1. ___ ___ *(0 – 11; 88=Not assessed)* Repetitions IVA 1f2. ___ ___ *(0 – 30; 88=Not assessed)* Comprehension VIB 1f3. ___ ___ . ___ *(0.0 – 12; 88.8=Not assessed)*			1f4. ___ ___ . ___ *(0.0 – 53; 88.8=Not assessed)* (1f1 + 1f2 + 1f3)
Visuospatial			
IXA+IXB 1g1. ___ . ___ *(0.0 – 8.0; 8.8=Not assessed)*			1g2. ___ . ___ *(0.0 – 8.0; 8.8=Not assessed)*

1. Down Syndrome Mental Status Examination (DSMSE)

Knowledge of the Examiner

IIIE+IIIF	1h1. ___ *(0 – 3; 8=Not assessed)*	1h2. ___ *(0 – 3;8= Not assessed)*
	DSMSE TOTAL SCORE:	**TOTAL SCORE:** 1i1. ___ ___ ___ . ___ *(0.0 – 103.0; 995.0 – 998.0)*

2. Cued Recall Task (whole integer range)

2a. Was any part of the Cued Recall Task administered?

0 ☐ No (If No, enter reason code, 95 – 98: ___ ___ and **SKIP TO QUESTION 3**)

1 ☐ Yes (If Yes, **CONTINUE**)

2b. Indicate which cue card set was used:

1 ☐ Version 1 (Set A)

2 ☐ Version 2 (Set B)

(NOTE: Set B no longer used by ABC-DS. See Down Syndrome Module neuropsychological battery instructions.)

2c. Training trial

	TRIAL 1	TRIAL 2	TRIAL 3
Card 1	2c1. ___ *(0 – 4)*	2c4. ___ *(0 – 4)*	2c7. ___ *(0 – 4)*
Card 2	2c2. ___ *(0 – 4)*	2c5. ___ *(0 – 4)*	2c8. ___ *(0 – 4)*
Card 3	2c3. ___ *(0 – 4)*	2c6. ___ *(0 – 4)*	2c9. ___ *(0 – 4)*

2d. Test trials

	FREE RECALL	INTRUSIONS TO FR	CUED RECALL	INTRUSIONS TO CR
Trial 1	2d1. ___ *(0 – 12)*	2d2. ___ ___ *(no limit)*	2d3. ___ *(0 – 12)*	2d4. ___ ___ *(no limit)*
Trial 2	2d5. ___ *(0 – 12)*	2d6. ___ ___ *(no limit)*	2d7. ___ *(0 – 12)*	2d8. ___ ___ *(no limit)*
Trial 3	2d9. ___ *(0 – 12)*	2d10. ___ ___ *(no limit)*	2d11. ___ *(0 – 12)*	2d12. ___ ___ *(no limit)*
TOTAL SCORE	2d13. ___ ___ *(0 – 36)*	2d14. ___ ___ *(no limit)*	2d15. ___ ___ *(0 – 36)*	2d16. ___ ___ *(no limit)*

3. Appraisal of participant engagement

Select the best description of the participant's behavior during each test:

	COOPERATIVE AND ENGAGED	COOPERATIVE BUT DISTRACTED	UNCOOPERATIVE	NOT ADMINISTERED
3a. DSMSE	1 ☐	2 ☐	3 ☐	9 ☐
3b. Cued Recall Task	1 ☐	2 ☐	3 ☐	9 ☐

Form D1D: Clinician Exam and Diagnosis

INSTRUCTIONS: This form is to be completed by the clinician. Check only one box per question.

1. Head circumference	____ ____ ____ cm *(777 = provided at previous visit; 999 = Not assessed)*

QUESTION 1: Measure the largest possible circumference — from the most prominent part of the forehead (often 1 – 2 fingers above the eyebrow) around to the widest part of the back of the head. If head circumference cannot be measured (e.g., if participant is uncooperative), enter **888=Not assessed.** If provided at a previous visit, enter **777 = provided at previous visit**.

QUESTIONS 2 – 4, below: For individuals with Down syndrome, individual primitive reflexes may be present throughout the lifespan with or without the presence of dementia. However, individuals with Down syndrome who have dementia tend to show a larger aggregate of these primitive reflexes.

DEFINITIONS:

Palmar grasp: Examiner's finger placed between participant's thumb and fifth digit causes participant's fingers to close reflexively.

Snout: Moderate tap to the philtrum results in pursing or pouting of the lips.

Rooting: When examiner touches corner of participant's mouth, participant's head moves in that direction.

2. Pathological reflexes — palmar grasp	☐ 1 Absent ☐ 2 Present ☐ 9 Unknown/not assessed
3. Pathological reflexes — snout	☐ 1 Absent ☐ 2 Present ☐ 9 Unknown/not assessed
4. Pathological reflexes — rooting	☐ 1 Absent ☐ 2 Present ☐ 9 Unknown/not assessed
5. What is the participant's chromosome diagnosis?	☐ 1 Trisomy 21 ☐ 2 Translocation DS ☐ 3 Mosaic DS ☐ 9 Unknown/not assessed

6. What is the participant's cognitive status?	☐ 1 Cognitively stable ☐ 2 MCI-DS ☐ 3 Dementia ☐ 9 Unable to determine

QUESTION 6: Recommended guidelines for case consensus process

OVERVIEW

Determination of overall Alzheimer's disease (AD)-related clinical status is based on overall profile of performance on the directly administered neuropsychological measures and caregiver-reported measures of dementia symptoms, combined with clinical judgment and in consideration of baseline IQ, medical/psychiatric history, neurological exam, and recent life events. All available time points of data (i.e., previous study visits) are reviewed in this process.

Weighing available information:

Consensus Summary Table

AREA	SOURCE		
	Caregiver report	Cognitive scores	Other*
Memory			
Non-memory, cognitive			
Emotional/behavioral			
Functional behavior			

**(e.g., neuro exam, research team observations)*

Instructions: For each source and area, enter the appropriate value:

0 = No concerns about decline/change

1 = Mild or inconsistent concerns about decline/change

2 = Moderate/consistent concerns about decline/change

Shaded boxes indicate that there are no scores in this area from that source.

INTERPRETING THE DATA

Information to be used in the determination of diagnostic status will be primarily from the caregiver report and from direct testing/observation of the participant. But there also may be other sources of information, such as a report from a local neurologist who has given the participant an MCI or dementia diagnosis and review of medical records. The Down Syndrome Module Working Group ("Working Group") suggests organizing the information using the above table. As there aren't well-normed tests for adults with Down syndrome, it can be difficult to make a diagnosis of MCI-DS or dementia following a single visit. A key to interpreting any data is to know the individual's level of functioning and/or premorbid IQ (or prior functioning for those who come with concerns regarding possible dementia).

Interpreting individual measures: The Working Group strives towards using the same evaluators and the same caregivers for all visits. If that is not the case (especially if the caregiver is different), the team may need to wait for data from a subsequent visit before agreeing on an MCI-DS or dementia diagnosis (depending upon how reliable

the new information is judged). A second challenge in the interpretation of symptoms involves situations where there has been a worsening of performance, but this has occurred in the context of significant environmental changes (e.g., moving to a different home, death of a parent, a major medical issue). In such cases, it may be best to use the "unable to determine" diagnostic category until the participant is seen again.

Although the Working Group provides guidelines for interpreting performance, it should be noted that determination of consensus diagnosis is based on clinical judgment and the overall pattern of findings across measures (think of it as a gestalt interpretation) rather than through the use of cut-off scores or a single measure.

a) **Dementia Questionnaire for People with Learning Disabilities (DLD)**: The initial studies by measure authors of the DLD have suggested that a change of >7 points on the Sum of Cognitive Scores (SCS) and a change of >5 points on the Sum of Social Scores (SOS) are associated with a diagnosis of dementia.[1] Individuals functioning in the moderate to severe ranges of ID will likely have high DLD scores at baseline. Change in both the SCS and SOS have previously found to correspond to MCI and dementia status in Down syndrome,[2] with evidence that change in SCS may occur first.[3] However, as the DLD manual indicates sensitivity and specificity are imperfect for these diagnostic criteria, DLD findings need to be interpreted in the context of other evidence (e.g., change should be compared with change noted in other measures).

b) **The Neuropsychiatric Inventory Questionnaire (NPI-Q)**: The NPI assesses 13 neuropsychiatric areas, which are scored as "not present" and as mild/moderate/severe if present. There is a total score ranging from 0 to 36 points. The emergence of new concerns or increases in the severity of existing neuropsychiatric problems may occur with MCI or dementia, but can also be due to other factors.

c) **NTG Early Detection Screen for Dementia (NTG)**: The original data on the NTG suggested that >=20 areas of worsening symptoms were suggestive of dementia in adults with DS. No data was available on MCI-DS. Hence, individuals with MCI-DS are generally expected to have >9 and <20 areas of worsening symptoms. Having one or more concern in the language or memory domain had good sensitivity and specificity for MCI-DS.

d) **Neuropsychological measures**: The DSMSE Total provides a summary mental status score (as well as specific scores on areas such as memory, visual-spatial, etc). This measure tends to be stable across time while individuals are Cognitively Stable.[4] Additionally, the Working Group's research to date suggests that episodic memory (Cued Recall) is the most sensitive measure for early MCI-DS and dementia.[4–8]

Recommendations for how to approach a diagnostic decision

1. Review the available data in each area using the **Consensus Conference Form**. If data from only a single visit is available, use premorbid level of functioning or IQ to determine if any of the neuropsychological and caregiver measures are consistent with that level. If there are multiple visits, look for significant change.
    - **Memory:** DLD (memory subscales), NTG (memory items), Cued Recall and DSMSE (memory items), NPI, notes from staff (e.g., participant no longer seemed to remember any of the staff at the hospital), notes from neuro exam.
    - **Non-memory, cognitive**: DLD, NTG, NPI (psychomotor)
    - **Emotional/behavioral**: NTG (behavioral Items), NPI, notes from staff (e.g., participant frequently refused to comply, tearful) and from neuro exam.

- **Functional**: NTG (activities of daily living items), NPI, notes from staff and neuro exam

2. Complete the **Consensus Summary Table**, indicating which source is identifying concerns ("0" for no concerns, "1" for mild or inconsistent concerns, and "2" for moderate/consistent concerns). Some rules of thumb are:
   - Consider whether changes may be due to major environmental changes (moving, death of parent) or a major illness (see NTG for checklist of recent life stressful events).
   - For MCI-DS, problems are expected in memory and/or non-memory, cognitive domains that could impact other areas (e.g., emotional/behavioral or adaptive/functional). If neuro-psychological test results indicate that a participant has improved or stayed the same as baseline in a number of areas, it would be difficult to justify an MCI-DS diagnosis.
   - For dementia, "2"-level concerns in multiple areas and across sources are expected. For example, changes in memory and non-memory, cognitive domains on directly administered neuropsychological assessment are expected, as well as declines in adaptive and functional behavior reported by caregivers.
   - There will be cases where the caregiver reports no concerns but the neuropsych testing shows a decline. It is possible that the participant was tired or just had a difficult time with the testing. In these cases, it will be important to consider anything that may have impacted testing. All aspects of data should be considered in making the rating.

References:

1. Evenhuis HM. Further evaluation of the Dementia Questionnaire for Persons with Mental Retardation (DMR). J Intellect Disabil Res. 1996;40(4):369-373. doi:10.1111/j.1365-2788.1996.tb00642.x
2. Startin CM, Lowe B, Hamburg S, et al. Validating the cognitive scale for down syndrome (CS-DS) to detect longitudinal cognitive decline in adults with down syndrome. Front Psychiatry. 2019;10(APR):158. doi:10.3389/fpsyt.2019.00158
3. Firth NC, Startin CM, Hithersay R, et al. Aging related cognitive changes associated with Alzheimer's disease in Down syndrome. Ann Clin Transl Neurol. 2018;5(6):741-751. doi:10.1002/acn3.571
4. Krinsky-McHale SJ, Zigman WB, Lee JH, et al. Promising outcome measures of early Alzheimer's dementia in adults with Down syndrome. Alzheimer's Dement Diagnosis, Assess Dis Monit. 2020;12(1):1-11. doi:10.1002/dad2.12044
5. Devenny DA, Zimmerli EJ, Kittler P, Krinsky-McHale SJ. Cued recall in early-stage dementia in adults with Down's syndrome. J Intellect Disabil Res. 2002;46(6):472-483. doi:10.1046/j.1365-2788.2002.00417.x
6. Hartley SL, Handen BL, Devenny DA, et al. Cognitive functioning in relation to brain amyloid-β in healthy adults with Down syndrome. Brain. 2014;137(9):2556-2563. doi:10.1093/brain/awu173
7. Hartley SL, Handen BL, Devenny D, et al. Cognitive decline and brain amyloid-β accumulation across 3 years in adults with Down syndrome. Neurobiol Aging. 2017;58:68-76. doi:10.1016/j.neurobiolaging.2017.05.019
8. Hartley SL, Handen BL, Devenny D, et al. Cognitive Indicators of Transition to Preclinical and Prodromal Stages of Alzheimer's Disease in Down Syndrome. Alzheimer's and Dementia. Alzheimer's Dement Diagnosis, Assessment, Dis Monit. Published online 2020. doi:10.1002/dad2.12096

Subject ID: _____ Consensus conference date: _____

Sex: _____ YOB: _____ Age V1: _____ Age V2: _____ Age V3: _____

IQ: _____ Test: _____ Date administered: _____

Level of intellectual disability: _____

Consensus conference form

CONSENSUS CONFERENCE DECISIONS

	CCD primary	Date	CCD secondary	Date
Visit 1				
Visit 2				
Visit 3				

Consensus conference decision (CCD)
0 = Cognitively stable
1 = Mild cognitive impairment-DS
2 = Possible dementia
3 = Definite dementia
4 = Status uncertain (complications)
5 = Indeterminable
6 = Decision on hold

Differential diagnosis (DDx)
1 = AD (possible)
2 = AD (probable)
3 = Mixed AD
4 = Vascular dementia
5 = Parkinson's dementia
6 = Other dementia
7 = Unknown

Participants with CCD 2 or 3:

DDx: _____

ALL participants:

Age @ 1st concern: _____

Age @ MD Dx: _____

NEUROPSYCHOLOGICAL BATTERY

Visit 1 date: _____ **Visit 2 date:** _____ **Visit 3 date:** _____

Down Syndrome Mental Status Examination

	Total score 1	Total score 2	Participant engagement
Visit 1			
Visit 2			
Visit 3			
Min – max:	(0 – 81)	(0 – 103)	

Cued Recall Task — Immediate

Totals:	Free recall	Intrusions to free recall	Cued recall	Intrusions to cued recall	Participant engagement
Visit 1					
Visit 2					
Visit 3					
Min – max:	(0 – 36)		(0 – 36)		

Subject ID: _____

INFORMANT QUESTIONNAIRES

Visit 1 date: _____ **Visit 2 date:** _____ **Visit 3 date:** _____

Dementia Questionnaire for People with Learning Disabilities (DLD)

	DLD total	DLD SCS	DLD SOS
Visit 1			
Visit 2			
Visit 3			
Min – Max:	(0 – 104)	(0 – 44)	(0 – 60)

NTG — Early Detection Screen for Dementia

		Visit 1	Visit 2	Visit 3
Category	**Min – Max**	**Number of concerns per category**		
Activities of daily living	0 – 7			
Language and communication	0 – 6			
Sleep-wake change pattern	0 – 8			
Ambulation	0 – 4			
Memory	0 – 9			
Behavior and affect	0 – 17			
Adult's self-reported problems	0 – 6			
Significant changes observed by others	0 – 6			
Total score	0 – 63			

CONSENSUS CONFERENCE COMMENTS

Visit 1 Conference **Conference date(s):** _____

Notes:

Visit 2 Conference **Conference date(s):** _____

Notes:

Visit 3 Conference **Conference date(s):** _____

Notes: