Form D1b: Biomarkers and Etiological Diagnosis

ADRC name: _____ Participant ID: _____ Form date: ___ ___ / ___ ___ / ___ ___ ___ ___

Visit #: _____ Examiner's initials: _____

INSTRUCTIONS: *This form is to be completed by the clinician. For additional clarification and examples, see* UDS Coding Guidebook *for* **Initial Visit Packet**, **Form D1a**. *Check only one box per question.*

THIS FORM IS DIVIDED INTO (2) MAIN SECTIONS:

SECTION 1: Biomarkers, Imaging and Genetics—*neurodegenerative imaging and CSF biomarkers, imaging, evidence for CVD, and known genetic mutations for AD and FTLD.*

SECTION 2: Etiological Diagnoses—*presumed etiological diagnoses for the cognitive disorder.*

SECTION 1 - Biomarkers, Imaging and Genetics

Complete this section if any of the following biomarker measures were used to support **or exclude** a presumed etiologic diagnosis. If none were used, then **(SKIP TO SECTION 2)** »

1. MRI Findings—was MRI data or information used to support an etiologic diagnosis?

☐1 Yes **(COMPLETE QUESTIONS 1a-1e6)**

If yes, specify year scan was obtained: _____

2. Molecular Neuropathology (Amyloid, tau, synuclein, etc)—is there biomarker evidence of molecular neuropathology from **CSF, plasma, or PET tracer imaging** to support an etiologic diagnosis?

☐1 Yes **(COMPLETE QUESTIONS 2a-2h)**

3. FDG PET Findings—was FDG-PET data or information used to support an etiologic diagnosis?

☐1 Yes **(COMPLETE QUESTIONS 3a-3d)**

If yes, specify year scan was obtained: _____

4. DAT Scan Findings—was DAT Scan data or information used to support an etiologic diagnosis?

☐1 Yes **(COMPLETE QUESTION 4a)**

If yes, specify year scan was obtained: _____

5. Supportive Genetics—is there a known Autosomal Dominant pathogenic variant to support the diagnosis?

☐1 Yes **(COMPLETE QUESTIONS 5a-5c)**

6. No biomarkers were collected or biomarker status is unknown and not used to support **or exclude** a presumed etiologic diagnosis.

☐1 Yes **(SKIP to QUESTION 7)**

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SECTION 1 - Biomarkers, Imaging and Genetics *Continued...*

	MRI | Findings	No	Yes	Unknown/ Not Assessed
1a.	Hippocampal atrophy—was there an MRI finding of hippocampal atrophy, *according to your Center's standards?*	☐ 0	☐ 1	☐ 9
1b.	Were there structural MRI atrophy patterns suggestive of AD *(temporal parietal or biparietal atrophy)?*	☐ 0	☐ 1	☐ 9
1c.	Was there structural MR evidence for frontal or anterior temporal atrophy for FTLD?	☐ 0	☐ 1	☐ 9
1d.	Were there MRI findings of assymetric atrophy	☐ 0	☐ 1	☐ 9
1e.	Was there evidence for cerebrovascular disease (CVD) on imaging? **(IF NO, SKIP TO QUESTION 2a)**	☐ 0	☐ 1	☐ 9
	Imaging findings:			
	1e1. ☐ 1 Large vessel infarct(s)			
	1e2. ☐ 1 Lacunar infarct(s)			
	1e3. ☐ 1 Macrohemorrhage(s)			
	1e4. ☐ 1 Microhemorrhage(s)			
	1e5. ☐ 1 Moderate white-matter hyperintensity (CHS score 5-6)			
	1e6. ☐ 1 Extensive white-matter hyperintensity (CHS score 7-8+)			

Amyloid

	MOLECULAR NEUROPATHOLOGY | Blood, CSF, Tracer Pet Imaging	No	Yes	Unknown/ Not Assessed
2a.	**CSF**—were there CSF amyloid findings of abnormally low amyloid in CSF suggesting elevated cerebral amyloid, *according to your Center's standards?* **2a1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9
2b.	**Blood-based**—were there blood-based measures of amyloid suggesting evidence of elevated cerebral amyloid, *according to your Center's standards?* **2b1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9
2c.	**PET**—was there abnormally elevated amyloid on PET, *according to your Center's standards?* **2c1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9

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SECTION 1 - Biomarkers, Imaging and Genetics *Continued...*

Tau

2d.	**CSF**—was there abnormally elevated CSF ptau, *according to your Center's standards?* **2d1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9
2e.	**CSF**—as there abnormally elevated CSF total tau, *according to your Center's standards?* **2e1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9
2f.	**Blood-based**—were there blood-based measures supporting the presence of elevated cerebral tau, *according to your Center's standards?* **2f1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9
2g.	**PET**—was there tau PET evidence for AD? **2g1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9

Other

2h.	Was there other biomarker evidence of molecular neuropathology? **(SPECIFY):** _____ **2h1.** *If assessed, year of assessment:* _____	☐ 0	☐ 1	☐ 9

	FDG-PET | Findings	No	Yes	Unknown/ Not Assessed
3a.	FDG-PET Pattern of AD—were there FDG-PET findings of temporal parietal or posterior cingulate/precuneus hypometabolism consistent with AD or known AD-related syndromes, *according to your Center's standards?*	☐ 0	☐ 1	☐ 9
3b.	Was there FDG-PET evidence for frontal or anterior temporal hypometabolism consistent with FTLD related syndromes, *according to your Center's standards?*	☐ 0	☐ 1	☐ 9
3c.	Was there an FDG-PET finding of occipital hypometabolism or cingulate island sign consistent with LBD, *according to your Center's standards?*	☐ 0	☐ 1	☐ 9
3d.	Other **(SPECIFY):** _____	☐ 0	☐ 1	

	DAT-SCAN | Findings	No	Yes	Unknown/ Not Assessed
4a.	Were there abnormal Dopamine transporter scan (DAT scan) findings consistent with dopaminergic neuronal loss, *according to your Center's standards?*	☐ 0	☐ 1	☐ 9

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SECTION 1 - Biomarkers, Imaging and Genetics

Continued...

	SUPPORTIVE GENETIC INFORMATION	No	Yes	Unknown/ Not Assessed
5a.	Does the participant have a dominantly inherited AD pathogenic variant *(PSEN1, PSEN2, APP)?* *If yes, specify variant:* _____	☐ 0	☐ 1	☐ 9
5b.	Does the participant have a hereditary FTLD mutation *(e.g., GRN, VCP, TARBP, FUS, C9orf72, ChMP2B, MAPT)?* *If yes, specify variant:* _____	☐ 0	☐ 1	☐ 9
5c.	Does the participant have a hereditary mutation other than an AD or FTLD mutation? *If yes, specify variant:* _____	☐ 0	☐ 1	☐ 9

SECTION 2 - Etiologic Diagnoses

*Using all the available data (i.e. clincial, cognitive, biomarker, etc) please provide an etiologic diagnosis. For those with no biomarker data, enter a **presumed** etiological diagnosis.*

Must be filled out for all participants. Indicate whether a given condition is a primary, contributing, or non-contributing cause of the observed impairment, based on the clinician's best judgment. Select one or more syndrome from questions *(below)* as **Present**; all others will default to **Absent** in the NACC database ›› *only one diagnosis should be selected as **1 = Primary.***

For participants with normal cognition: Indicate the presence of any diagnoses by selecting **1 = Present**, and leave the questions on whether the diagnosis was primary, contributing, or non-contributing blank. Participants with positive biomarkers but no clinical symptoms of Alzheimer's disease, Lewy body disease, or frontotemporal lobar degeneration **should not** have these diagnoses selected as Present. Instead, the biomarker data from Section 2 can be used to identify the presence of preclinical disease.

	ETIOLOGIC DIAGNOSES	Present		Primary	Contributing	Non-contributing
7.	Alzheimer's disease	☐ 1	**7a.**	☐ 1	☐ 2	☐ 3
8.	Lewy body disease	☐ 1	**8a.**	☐ 1	☐ 2	☐ 3
	8b. Dementia with Lewy Bodies	☐ 1	**8b1.**	☐ 1	☐ 2	☐ 3
	8c. Parkinson's disease	☐ 1	**8c1.**	☐ 1	☐ 2	☐ 3
	8d. Parkinson's disease Dementia	☐ 1	**8d1.**	☐ 1	☐ 2	☐ 3
9.	Frontotemporal lobar degeneration	☐ 1	**9a.**	☐ 1	☐ 2	☐ 3
	9b. Progressive supranuclear palsy (PSP)	☐ 1	**9b1.**	☐ 1	☐ 2	☐ 3
	9c. Corticobasal degeneration (CBD)	☐ 1	**9c1.**	☐ 1	☐ 2	☐ 3
	9d. FTLD with motor neuron disease	☐ 1	**9d1.**	☐ 1	☐ 2	☐ 3
	9e. FTLD NOS	☐ 1	**9e1.**	☐ 1	☐ 2	☐ 3

continued...

SECTION 2 - Etiologic Diagnoses *Continued...*

9f. If FTLD (questions 9a-9d) is **Present**, *specify FTLD subtype:*

- ☐ 1 Tauopathy
- ☐ 2 TDP-43 proteinopathy
- ☐ 3 Other (**SPECIFY**): _____
- ☐ 9 Unknown

	ETIOLOGIC DIAGNOSES	Present		Primary	Contributing	Non-contributing
10.	Vascular brain injury (based on clinical and imaging evidence according to your Center's standards)	☐ 1	**10a.**	☐ 1	☐ 2	☐ 3
11.	Multiple system atrophy	☐ 1	**11a.**	☐ 1	☐ 2	☐ 3
12.	Chronic Traumatc Encephalopathy	☐ 1	**12a.**	☐ 1	☐ 2	☐ 3
13.	Down syndrome	☐ 1	**13a.**	☐ 1	☐ 2	☐ 3
14.	Huntington's disease	☐ 1	**14a.**	☐ 1	☐ 2	☐ 3
15.	Prion disease (CJD, other)	☐ 1	**15a.**	☐ 1	☐ 2	☐ 3
16.	Amyloid Angiopathy	☐ 1	**16a.**	☐ 1	☐ 2	☐ 3
17.	LATE: Limbic-predominant Age-Related TDP-43 Encephalopathy	☐ 1	**17a.**	☐ 1	☐ 2	☐ 3
18.	Other (**SPECIFY**): _____	☐ 1	**18a.**	☐ 1	☐ 2	☐ 3