

# The Genetics Module – Bud Kukull's slides



**INITIAL VISIT PACKET** NACC UNIFORM DATA SET (UDS) — FTLD MODULE

## Form A3aF: Record of Consent for Biologic Specimen Use

Center: \_\_\_\_\_ Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

**NOTE:** This form is to be completed by clinic staff responsible for obtaining consents, coded from an existing consent at clinic. For additional clarification and examples, see FTLD Coding Guidebook for Initial Visit Packet, Form A3aF.

Visit #: \_\_\_\_\_

Examiner's initials: \_\_\_\_\_

One of these forms will be completed for each relative who provides a specimen.

**1. What relative's consent is being recorded on this form?**

- ☐ 1 Mother  
☐ 2 Father  
☐ 3 Sibling (sibling's birth year: \_\_\_\_\_)  
☐ 4 Child (child's birth year: \_\_\_\_\_)

**NOTE:** "Unknown" (9999) is not a permissible value for sibling's or child's birth year. If birth year is unknown, please provide an approximate year on **UDS Initial Visit Form A3** so that the sibling or child ends up in correct birth order relative to the other siblings or children.

*"Sibling's birth year" or "child's birth year" on this form **MUST** agree with the birth year listed for that person on UDS Initial Visit Form A3 and FTLD Module Form A3F.*

Please indicate that subject provided consent for the following:

1a.	"I permit my sample to be stored and used in future research of neurologic disease at (home institution)."	<input type="checkbox"/> 0 No	<input type="checkbox"/> 1 Yes
1b.	"I permit my sample to be stored and used in future research at (home institution) to learn about, prevent, or treat other health problems."	<input type="checkbox"/> 0 No	<input type="checkbox"/> 1 Yes
1c.	"There is a small chance that some commercial value may result from my sample at the National Cell Repository for Alzheimer's Disease (NCRAD). If that would happen, I would not be offered a share in any profits. I permit (home institution) to give my sample to researchers at other institutions."	<input type="checkbox"/> 0 No	<input type="checkbox"/> 1 Yes



## A3F - record of consent for family members

- Use only for relatives of the Proband
- Complete one form for each relative enrolled from whom specimens are obtained
- Use the same Birth year as recorded on UDS Family History
  - *Estimate birth year based on other information about Proband and family*
  - *“Unknown” (9999) is UN-HELPFUL: try to get a real year and also replace if “unknown” on UDS*

## NACC Uniform Data Set (UDS) – Initial Visit Packet

### Form A3: Subject Family History

Center: \_\_\_\_\_ ADC Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

**NOTE:** This form is to be completed by intake interviewer per subject/informant report. For additional clarification and examples, see UDS Coding Guidebook for Initial Visit Packet, Form A3.

ADC Visit #: \_\_\_\_\_

Examiner's initials: \_\_\_\_\_

For the following questions:

Dementia refers to progressive loss of memory and cognition, and is often described as senility, dementia, Alzheimer's Disease, hardening of the arteries, or other causes that compromised the subject's social or occupational functioning and from which they did not recover.

Age at onset refers to the age at which dementia symptoms began, not the age at which the diagnosis was made.

Please consider blood relatives only.

#### PARENTS:

	a. Year of birth  (9999=unknown)	b. Is the parent still living?			c. If deceased, indicate year of death  (9999=unknown)	d. Does/did this parent have dementia (defined above), as indicated by symptoms, history or diagnosis?			e. If yes, indicate age at onset  (999=unknown)
		Yes	No	Unknown		Yes	No	Unknown	
1. Mother	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
2. Father	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____

#### AFFECTED SIBLINGS

	a. Sibling's birth mo / yr	b. Neurological problem*	c. Primary DX**	d. Method of evaluation***	e. Age of onset
3a.	____/____	____	_____	____	_____
3b.	____/____	____	_____	____	_____
3c.	____/____	____	_____	____	_____
3d.	____/____	____	_____	____	_____
3e.	____/____	____	_____	____	_____
3f.	____/____	____	_____	____	_____

Center: \_\_\_\_\_ ADC Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

**NOTE:** This form is to be completed by intake interviewer per subject/informant report. For additional clarification and examples, see UDS Coding Guidebook for Initial Visit Packet, Form A3..

ADC Visit #: \_\_\_\_

**SIBLINGS:**  
(continued)

	5a. Year of birth  (9999=unknown)	5b. Is the sibling still living?			5c. If deceased, indicate year of death  (9999=unknown)	5d. Does/did this sibling have dementia (defined above), as indicated by symptoms, history or diagnosis?			5e. If yes, indicate age at onset  (999=unknown)
		Yes	No	Unknown		Yes	No	Unknown	
Sibling 4	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 5	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 6	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 7	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 8	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 10	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 11	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 12	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 13	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 14	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 15	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 16	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 17	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 18	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 19	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Sibling 20	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____

Center: \_\_\_\_\_ ADC Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

**NOTE: This form is to be completed by intake interviewer per subject/informant report. For additional clarification and examples, see UDS Coding Guidebook for Initial Visit Packet, Form A3..**

ADC Visit #: \_\_\_\_

CHILDREN:									
6. How many biological children did the subject have? (99 = Unknown) ____									
7. For all biological children, indicate the following:									
	7a. Year of birth (9999=unknown)	7b. Is the child still living?			7c. If deceased, indicate year of death (9999=unknown)	7d. Does/did this child have dementia (defined above), as indicated by symptoms, history or diagnosis?			7e. If yes, indicate age at onset (999=unknown)
		Yes	No	Unknown		Yes	No	Unknown	
Child 1	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 2	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 3	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 4	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 5	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 6	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 7	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 8	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 10	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 11	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 12	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 13	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 14	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____
Child 15	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____	<input type="checkbox"/> 1	<input type="checkbox"/> 0	<input type="checkbox"/> 9	_____

**INITIAL VISIT PACKET** NACC UNIFORM DATA SET (UDS) — FTLD MODULE  
**Form A3F: Family History: Affected Family Members**

Center: \_\_\_\_\_ Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

**NOTE:** This form is to be completed by a clinician with experience in evaluating patients with frontotemporal lobar degeneration. For additional clarification and examples, see FTLD Coding Guidebook for Initial Visit Packet, Form A3F.

Visit #: \_\_\_\_\_

Examiner's initials: \_\_\_\_\_

**"AFFECTED FAMILY MEMBERS"** — For the purposes of Form A3F, "affected" means affected by dementia **OR** by any of the non-normal clinical diagnoses listed in Appendix 1 on page 4 of this form.

AFFECTED FAMILY MEMBERS	
1. Are there affected family members? (See box above for definition of "affected.")	<input type="checkbox"/> 0 No
If the answer is "No" or "Unknown," please skip the rest of this form.	<input type="checkbox"/> 1 Yes
	<input type="checkbox"/> 9 Unknown

**\*Codes for neurological problems and psychiatric conditions**

- 1 Cognitive impairment/behavior change
- 2 Parkinsonism
- 3 ALS
- 4 Other neurologic condition such as multiple sclerosis or stroke
- 5 Psychiatric condition such as schizophrenia, bipolar disorder, or alcoholism
- 9 Unknown

**\*\*Codes for primary diagnosis**

See Appendix 1 on page 4 of this form

**\*\*\*Codes for method of evaluation**

For descriptions, see Appendix 2 on page 4 of this form

- 1 Autopsy
- 2 Examination
- 3 Medical record review from formal dementia evaluation
- 4 Review of general medical records AND informant and/or subject telephone interview
- 5 Review of general medical records only
- 6 Subject and/or informant telephone interview
- 7 Family report

**AFFECTED PARENTS** — Use the form below to provide information on affected parents only (see definition of "affected" in the box above).

AFFECTED PARENTS				
	a. Neurological problem*	b. Primary DX**	c. Method of evaluation***	d. Age at onset
2a. Mother	_____	_____	_____	_____
2b. Father	_____	_____	_____	_____

## **A3F: Family History**

***“Affected” parents, sibs, and children only***

- **Use UDS Family Hx to determine Birth year—**
  - **If Module birth yr is more valid then edit UDS to match and re-submit it**
- **“Affected”: dementia or non-normal diagnosis from Appendix 1 of the form**
- **Select most specific Dx known**
- **Age of Onset: age at which Sx began that developed into the primary Dx**
  - **“Oh, he’s been forgetful most of his life.”**
  - **Not good enough!**

Center: \_\_\_\_\_ Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

Visit #: \_\_\_\_\_

## \*\*\*APPENDIX 1: PRIMARY DIAGNOSIS CODES

CODE	DIAGNOSIS
040	Mild cognitive impairment (MCI), not otherwise specified
041	MCI — amnesic
042	MCI — multiple domain with amnesia
043	MCI — single domain nonamnesic
044	MCI — multiple domain nonamnesic
045	Impaired, but not MCI
050	Alzheimer disease
070	Dementia with Lewy bodies
080	Vascular dementia
100	Alcohol-related dementia
110	Dementia of undetermined etiology
120	Behavioral variant frontotemporal dementia
130	Primary progressive aphasia, semantic variant
131	Primary progressive aphasia, nonfluent/agrammatic variant
132	Primary progressive aphasia, logopenic variant
133	Primary progressive aphasia, not otherwise specified

CODE	DIAGNOSIS
140	Progressive supranuclear palsy
150	Corticobasal syndrome/corticobasal degeneration
160	Huntington's disease
170	Prion disease
180	Cognitive dysfunction from medications
190	Cognitive dysfunction from medical illness
200	Depression
210	Other major psychiatric illness
220	Down syndrome
230	Parkinson disease
240	Stroke
250	Hydrocephalus
260	Traumatic brain injury
270	CNS neoplasm
280	Other
310	Amyotrophic lateral sclerosis
320	Multiple sclerosis

## \*\*\*APPENDIX 2: METHOD OF EVALUATION

- Autopsy** — If the autopsy was performed at an outside institution, **you must have the report** to code as diagnosis by autopsy.
- Examination** — The subject must have been examined in person at your ADC/institution or by genetic studies staff associated with your ADC/institution to code as diagnosis by examination. Medical records may or may not have been used when assigning diagnosis.
- Medical record review from formal dementia evaluation** — Medical records should be from an examination that focused specifically on dementia; that was performed by a neurologist, geriatrician, or psychiatrist; that includes a neurologic examination, an imaging study, and cognitive testing (e.g., MMSE, Blessed, or more formal tests). A telephone interview may also be used to collect additional information.
- Review of general medical records AND informant and/or subject telephone interview** — **General medical records** can be of various types, including those from a primary-care physician's office, hospitalization records, nursing home records, etc. They may include a neurologic exam and a cognitive test such as the MMSE along with a medical history. **The telephone interview** with the subject and/or the informant should include a medical history to capture the nature and presentation of cognitive deficits, if present, and age of onset if symptomatic. If the subject is normal or is in the early stages of dementia, brief formal cognitive testing should be included in the interview. Unless an affected subject is in the early stages of dementia, the interview should be conducted with an informant.
- Review of general medical records ONLY** — See definition No. 4 above. If general medical records are used to diagnose a subject as demented or not demented, they should include a medical history, neurologic exam, and a cognitive test such as an MMSE. In most cases, general medical records alone should not be used to assign a diagnosis of mild cognitive impairment, or of any of the FTLD spectrum subtypes, or of parkinsonian disorders other than Parkinson disease.
- Subject and/or informant telephone interview** — See definition No. 4 above.
- Family report** — Family report should be coded when the informant for the family reports a subject as having been diagnosed with a particular disorder. In most cases, family report alone should not be used to assign a diagnosis of mild cognitive impairment, or of any of the FTLD spectrum subtypes, or of parkinsonian disorders other than Parkinson disease.





Center: \_\_\_\_\_ Subject ID: \_\_\_\_\_ Form Date: \_\_\_\_/\_\_\_\_/\_\_\_\_  
Visit #: \_\_\_\_\_

**AFFECTED CHILDREN** — Use the form below to provide information on affected children only (see definition of “affected” in the box on page 1 of this form).

“Child’s birth year” on this form MUST agree with the birth year listed for that child on UDS Initial Visit Form A3 and FTL Module Form A3aF (if applicable).

“Unknown” (9999) is not a permissible value. If birth year is unknown, please provide an approximate year on UDS Initial Visit Form A3 so that the child with unknown birth year ends up in correct birth order relative to the other children. (EXAMPLE: Suppose a subject has three children. The oldest is a son born in 1960, the youngest a son born in 1964, and the middle child a girl whose birth year is unknown. The girl should be assigned an approximate birth year of 1962 or 1963.) Use that same birth year from UDS Initial Visit Form A3 on FTL Module Forms A3F and A3aF.

If an affected child has already been listed on UDS Initial Visit Form A3 with a birth year of 9999, then UDS Initial Visit Form A3 must be edited so that an approximate birth year is entered, as described in the paragraph above. That same birth year should be entered below.

AFFECTED CHILDREN					
	a. Child's birth year	b. Neurological problem*	c. Primary DX**	d. Method of evaluation***	e. Age of onset
4a.	_____	_____	_____	_____	_____
4b.	_____	_____	_____	_____	_____
4c.	_____	_____	_____	_____	_____
4d.	_____	_____	_____	_____	_____
4e.	_____	_____	_____	_____	_____
4f.	_____	_____	_____	_____	_____
4g.	_____	_____	_____	_____	_____
4h.	_____	_____	_____	_____	_____
4i.	_____	_____	_____	_____	_____
4j.	_____	_____	_____	_____	_____
4k.	_____	_____	_____	_____	_____
4l.	_____	_____	_____	_____	_____
4m.	_____	_____	_____	_____	_____
4n.	_____	_____	_____	_____	_____
4o.	_____	_____	_____	_____	_____

**\*Codes for neurological problems and psychiatric conditions**

- 1 Cognitive impairment/behavior change
- 2 Parkinsonism
- 3 ALS
- 4 Other neurologic condition such as multiple sclerosis or stroke
- 5 Psychiatric condition such as schizophrenia, bipolar disorder, or alcoholism
- 9 Unknown

**\*\*Codes for primary diagnosis**

See Appendix 1 on page 4 of this form

**\*\*\*Codes for method of evaluation**

For descriptions, see Appendix 2 on page 4 of this form

- 1 Autopsy
- 2 Examination
- 3 Medical record review from formal dementia evaluation
- 4 Review of general medical records AND informant and/or subject telephone interview
- 5 Review of general medical records only
- 6 Subject and/or informant telephone interview
- 7 Family report



# Issues you may have thought of...

---

- What to do about twins ?
- What to do about sibs or children born in the same year?
- How can the correct one be identified from UDS family history?
  - Birth month of relatives in not collected on UDS or FTLD Module
- ...*stay tuned, no good answer for these but:*
- Identification would be completed by actual Family-based study

# Summary of A3F forms

- A3F records Affected relatives only
- Use UDS Family to obtain birth year and verify match—estimates are preferred to “unknown”
- Choose most specific primary diagnosis from Appendix 1
- Data can be used to formulate Family-based studies and files can be made for use by “Progeny” software