

**Required for Review:**

3-generation pedigree (or table completed below)

**Preferred:**

Clinic notes/pathology records for proband or affected relatives, if available

## Cancer Application for Family Studies

Please complete the following information and fax to Ambry Genetics with the required documents at **949-271-5621 (ATTN: Family Studies)** or email an attachment to **FamilyStudies@ambrygen.com**. Please allow 7-10 business days for a response from one of our genetic counselors. Incomplete applications will not be reviewed until all required information is received.

### Proband Information

Ordering Physician/GC:

Name: \_\_\_\_\_ Accession number: \_\_\_\_\_ Phone number: \_\_\_\_\_

DOB: \_\_\_\_\_ Ethnicity: \_\_\_\_\_ Email: \_\_\_\_\_

Proband Clinical Cancer History (please attach relevant clinic/pathology notes if available):

1. Which VUS are you interested in testing? (Please refer to exclusions below) \_\_\_\_\_

2. Which of the proband's relatives are available for testing? A detailed 3-generation pedigree is preferred, but the table below may be used in lieu of pedigree. Please see "considerations for selection" below.

NAME (FIRST NAME ONLY IS ACCEPTABLE)	GENDER	RELATIONSHIP TO PROBAND (INDICATE MATERNAL OR PATERNAL)	HEALTHY (CURRENT AGE)	AFFECTED	CANCER TYPE (DIAGNOSIS AGE)	OUTSIDE US?*
			<input type="checkbox"/>	<input type="checkbox"/>		
			<input type="checkbox"/>	<input type="checkbox"/>		
			<input type="checkbox"/>	<input type="checkbox"/>		
			<input type="checkbox"/>	<input type="checkbox"/>		
			<input type="checkbox"/>	<input type="checkbox"/>		
			<input type="checkbox"/>	<input type="checkbox"/>		
			<input type="checkbox"/>	<input type="checkbox"/>		

\*Please indicate if any of the proband's relatives live outside the United States as special considerations may apply.

### Considerations for selection of families/individuals for enrollment:

- For common cancers (i.e., breast and prostate), younger affected individuals are more likely to be selected (dx <60y).
  - In order to be informative, Gleason scores are required for prostate cancer diagnoses.
- Families with cancers unrelated to the gene in question are unlikely to be selected.
- Occasionally, older (>70) unaffected individuals may be approved (may vary due to gene-specific penetrance differences).
- Generally, younger unaffected individuals for adult-onset cancer syndromes are not informative and will not be approved.
- For genes with significant de novo rates (i.e., *APC*, *TP53*, *NF1*), testing of parents may be approved regardless of age or affected status to assess for de novo mutation status.

**Common exclusions for complimentary family studies:** 1. VUS in moderate penetrance genes, unless for potential phase determination (i.e., *ATM*, *BARD1*, *BRIP1*, *CHEK2*, *MRE11A*, *NBN*, *RAD50*, *RAD51C*, *RAD51D*, *BLM*, *XRCC2*, *FANCC*). 2. Family history is not consistent with the gene in question (no gene-associated cancers reported in relatives). 3. High frequency VUS.

Please be aware that results for family studies have a longer turnaround time than our clinical testing (2-3 months) since priority is given to clinical specimens. If you have any questions, please contact Ambry Genetics and ask to speak with one of our family study specialists at 949-900-5500 or email FamilyStudies@ambrygen.com.