

Ordering Testing for Known Variants

Ambry Genetics can perform testing for known variants, like those identified in a family member, across many applications. However, the technology used for analysis, the need for positive control, and reporting limitations differ depending on the type of testing requested.

Family Previously Tested at Ambry

KNOWN VARIANT TYPE	ORDERING	POSITIVE CONTROL?	REPORTING NOTES	BILLING
Known variant in a gene analyzed via targeted NGS panel, such as cancer or cardiology panels	<ul style="list-style-type: none"> • Test code: 5555 • Specify gene and variant • Provide proband report or internal Ambry identifiers 	No*	Full gene analyzed. Will report presence/absence of known variant as well as any pathogenic, likely pathogenic, or VUS identified in gene.	Eligible for family testing for pathogenic and likely pathogenic variants within 180 days of original report.
Known variant from a neurology panel, exome testing, or whole genome testing	<ul style="list-style-type: none"> • Test code: 5555 • Specify gene(s) and variant(s) • Provide proband report or internal Ambry identifiers 	No*	Will report presence or absence of only the specific variant(s)	Otherwise, indicate billing information.
Known variant testing on a prenatal specimen	<ul style="list-style-type: none"> • Test code: 5555 • Test code for maternal cell contamination (MCC): 1260 • Specify gene(s) and variant(s) • Provide proband report or internal Ambry identifiers 	Yes, plus maternal specimen for MCC	Will report presence or absence of only the specific variant(s)*	Paid testing, including MCC
Known variant identified via SNP array performed at Ambry	<ul style="list-style-type: none"> • Test code: 5555 • Specify region of interest • Provide proband report or internal Ambry identifiers 	No*	Will report presence or absence of only the specific variant(s)	Paid testing

*Unless specifically requested by laboratory for quality assurance reasons.

Family Previously Tested at Another Lab

KNOWN VARIANT TYPE	ORDERING	POSITIVE CONTROL?	REPORTING NOTES	BILLING
Known variant in a gene analyzed via targeted NGS panel, such as cancer or cardiology panels	<ul style="list-style-type: none"> • Test code: 5555 • Specify gene and variant • Proband report required 	No*	Full gene analyzed. Will report presence/absence of known variant as well as any pathogenic, likely pathogenic, or VUS identified in gene.	Paid testing
Known variant from a neurology panel, exome testing, or whole genome testing (prenatal, routine exome, or routine whole genome)	Ambry does not perform known variant analysis for variants detected on an external lab's test.	N/A	See below**	N/A
Known variant testing on a prenatal specimen*	<ul style="list-style-type: none"> • Test code: 5555 • MCC (1260) • Specify gene(s) and variant(s) • Proband report required • Exome, whole genome, and neurology variants excluded 	Yes, plus maternal sample for MCC	Will report presence or absence of only the specific variant(s)	Paid testing including MCC
Known variant identified via SNP array	Ambry does not perform known variant analysis for variants detected on an external lab's test.	N/A	See below**	N/A

*Please review information about prenatal testing [here](#). Please contact Ambry directly if you have questions about accessing prenatal testing.

**Ambry reserves discretion in determining which platform/assay and test code are best suited to fulfill the known variant analysis order. We will communicate with you if requests cannot be fulfilled for technical reasons or platform limitations. Some genes have specific restrictions on their reported analytical ranges. Please review detailed assay information at ambrygen.com. If you are concerned about the reporting range of a specific gene, please contact us directly. These restrictions are subject to change at any time.

How to Order Known Variant Analysis Testing

When testing was performed at Ambry, please provide one of the following:

- The accession number (e.g., 25-112233) for the tested family member; or
- The tested family member's full name and date of birth; or
- The tested family member's report

When testing was performed at an external lab, a proband report is required for review of variant nomenclature and to ensure the appropriate testing strategy.

- Testing can often still be performed in the absence of a proband report, but reporting comments may be limited, or an alternative testing strategy may be recommended.
- However, known variant testing is not available for variants detected on external lab exome, exome backbone tests, whole genome, whole genome backbone tests, or chromosomal microarray.

Billing Information

For family members tested at Ambry:

Ambry offers family variant testing at no additional cost for blood relatives of patients (up to 4th degree) who undergo full single gene sequencing, multigene panel testing, exome sequencing, or whole genome sequencing at Ambry Genetics and are found to have a pathogenic or likely pathogenic variant. Testing must be completed within 180 days of the original report date. Whenever possible, more closely related relatives should be tested before more distant relatives. Note that Ambry can only provide such family testing services to patients receiving medical care in the US or US territories.

Outside of the family variant testing program, tests will be billed to the patient's insurance or cash pay pricing may be elected. See our [patient billing page](#) for more information.

Limitations

Deletion / duplication analysis is not available for some genes.

- Cardiology: *ABCG5, ABCG8, ALMS1, APOB, APOC2, APOC3, APOE, BGN, CALM2, CALM3, CBL, CBS, CHST14, CYP27A1, DOLK, FHL1, FKBP14, FLNA, FOXE3, GPIHBP1, LCAT, LDLRAP1, LIPA, LMF1, MAP2K1, MAP2K2, MAT2A, MFAP5, PPP1CB, PRDM5, RIT1, SCN10A, SHOC2, SLCO1B1, SOS1, SOS2, TECRL,* and *ZNF469*.
 - Only full gene deletion/duplications are reported in *ALPK3, FLNC, LOX*.
- Oncology: *ATRIP, AXIN2, CFTR, CPA1, CTNNA1, CTRC, DDX41, EGFR, EGLN1, HOXB13, KIT, MBD4, MITF, MLH3, MSH3, PALLD, PDGFRA, POLD1, POLE, PRSS1, RAD51B, RNF43, SPINK1,* and *TERT*.
 - *EPCAM*: only gross deletions limited to the 3' end of the gene are reported.
 - *GREM1*: only the status of the 40kb 5'UTR gross duplication is routinely analyzed and reported.
 - *NTHL1*: only full-gene gross deletions and duplications are detected.

Targeted analysis is not available for:

- BRCA Ashkenazi Jewish 3-mutation panel. If you desire analysis for the common Ashkenazi Jewish 3-site mutations, please use *BRCA1/BRCA2* gene sequence and deletion/duplication analysis (test code 8838).
- Genes from tests that have been discontinued, including: *BLM, CASR, FAM175A, FANCC, GALNT12, MRE11A, NBN, RAD50, RECQL, RINT1,* and *XRCC2*.