Ambry Genetics®
A Konica Minolta Company

Hereditary Neuro-Oncology Cancer Testing

REFERENCE GUIDE

November 2017

There is no room for doubt when it comes to making important life impacting healthcare decisions. By providing advanced confirmation genetic testing for hereditary brain and nervous system tumors, we can help you make more informed and reliable healthcare decisions with your patients.

The more accurate the results, the more insight you have to better treat your patients.

Why Is Genetic Testing Important?

KEY BENEFITS

Knowing if your patient has hereditary brain and/or nervous system tumors can help you determine their future risks and guide your medical management recommendations. Some patients with brain and/or nervous system tumors have an underlying genetic syndrome that can cause additional medical complications. Some key benefits of genetic testing include:

- Ability to modify surveillance options and age of initial screening
- Consideration of risk-reduction measures for your patient and/or their family members
- Referral to other specialists for screening/management of non-nervous system symptoms, as needed
- Identification of at-risk family members
- Availability of tailored treatment options (e.g. avoid radiation-based therapy for *TP53* mutation carriers)

Identify Patients Who May Need Genetic Testing

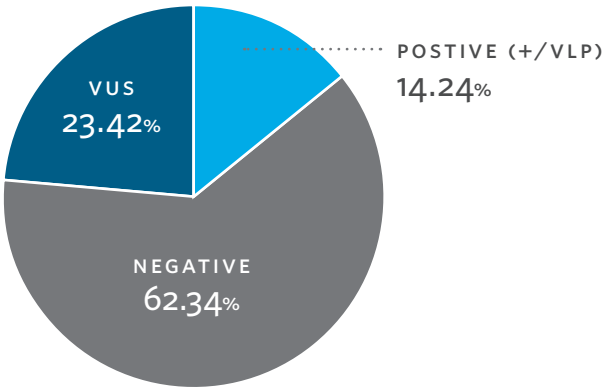
If your patient or their family members have any of the following, genetic testing may be considered:

MULTIPLE	**3 OR MORE** tumors/cancers on the same side of the family	**2 OR MORE** tumors/cancers in the same person
YOUNG	**BRAIN AND/OR NERVOUS SYSTEM TUMORS DIAGNOSED ≤50 YEARS**	
RARE	**PERSONAL OR FAMILY HISTORY OF ANY OF THE FOLLOWING TUMORS/CANCERS:** Atypical teratoid/rhabdoid tumors, choroid plexus carcinoma, ganglioglioma, pineoblastoma, pituitary blastoma	

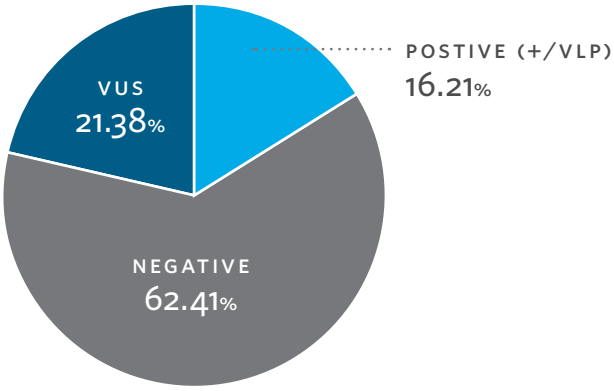
Genetic Testing Outcomes for Hereditary Brain Tumors

Over 79,000 cases of brain tumors are diagnosed each year, and over 2/3 are benign. Approximately 14-16% of brain tumors are due to a variety of hereditary causes, although this can vary widely by tumor type.[1]

Detection Rate for Benign Brain Tumors



Detection Rate for Malignant Brain Tumors



Ambry Genetics studied >600 patients with primary brain tumors, demonstrating a mutation detection rate of 14% and 16% for benign and malignant brain tumors, respectively, in 25 different genes. The genes identified in these patients are available on BrainTumorNext and CancerNext-*Expanded*.[2]

1. Ambry internal data
2. Jackson *et al.* Society of Neuro-Oncology Meeting Abstract. 2017.

Finding Answers Through Quality Genetic Testing

Ambry offers genetic testing for a broad range of hereditary brain and/or nervous system tumor conditions including the following:

CONDITION NAME	ASSOCIATED BRAIN/NERVOUS SYSTEM TUMORS	GENE(S)*
Li-Fraumeni syndrome (LFS)	Astrocytomas, glioblastomas, medulloblastomas, choroid plexus carcinomas, other	*TP53*
Neurofibromatosis Type 1 (NF1)	Neurofibromas, malignant nerve sheath tumors, optic gliomas, astrocytomas	*NF1*
Neurofibromatosis Type 2 (NF2)	Vestibular and other cranial nerve schwannomas, intracranial meningiomas, spinal tumors	*NF2*
Nevoid basal cell carcinoma (Gorlin syndrome)	Medulloblastomas, meningiomas	*PTCH1, SUFU*
Schwannomatosis	Schwannomas along the spinal and peripheral nerves	*SMARCB1*
Tuberous sclerosis complex (TSC)	Astrocytomas, cortical tubers/subependymal nodules	*TSC1, TSC2*
von Hippel-Lindau (VHL) disease	Hemangioblastomas of the brain and spine	*VHL*

*14-21 day turnaround time

Comprehensive Testing for Hereditary Brain/Nervous System Tumors

TEST NAME	DESCRIPTION	AVERAGE TURNAROUND TIME^
BrainTumorNext	Includes 27 high and moderate risk genes associated with increased risk for brain/nervous system tumors and other cancers/tumors, such as breast, colorectal, kidney, and neuroendocrine	15.8 calendar days
CancerNext-*Expanded*	Includes 67 genes associated with increased risks for brain, breast, colon, ovarian, pancreatic, prostate, renal, uterine, and many other cancers	14.9 calendar days

^Ambry's turnaround time represents the time that it takes for Ambry to perform the requested testing. When all necessary clinical and family history information is provided with the sample, results are typically completed within 14 days. We will notify you in the unusual event that results will take longer than 21 days.

Ambry continually participates in important game-changing studies to expand our knowledge of hereditary cancers. Please visit our website to see the most updated lists of genes included on our panels and additional testing options available: ambrygen.com/hereditary-cancer-panels

Over 1 Million Tests Completed

MOVING SCIENCE FORWARD

Purposeful Confirmatory Testing

Many labs validate their tests based on certain limited studies. That's why we participated and led the largest study of its kind (20,000 cases) guiding us to utilize confirmatory testing when we see specific well-defined thresholds. Our mission is to get it right the first time.

Understanding Disease Better Through Free Data Sharing

Identifying an individual's genetic information is nothing new—it's what we do with it that is unique. When labs share genomic information, we can together accelerate the understanding of human disease. Through AmbryShare, we leverage de-identified information to collaborate with others and help people everywhere find answers.

SuperLab

Our 65,000 square foot highly-automated CLIA/CAP certified lab produces some of the fastest turnaround times in the industry, without compromising testing accuracy or specificity.

Ambry's Translational Genomics (ATG) Lab

As an advanced diagnostic lab, it's our responsibility to ensure the results you get from us are accurate and that classification is as complete and robust as possible. Our ATG lab is a unique laboratory that provides an additional service at no additional cost for you and your patients to generate more precise data potentially bringing clarity to some variants of unknown significance (VUS). This helps to actively drive down the rate of VUS results and can give you an increased understanding of your patient's results, so you can better provide medical management recommendations and improve health outcomes.

About Ambry

Just as no two fingerprints are alike, the way disease presents itself in every individual is different. Since 1999, our mission has always been about understanding disease better, so treatments and cures can be found faster. Every sample that arrives in our lab is viewed as a person with a life and a story that is unique to only them. By providing advanced confirmation genetic testing for inherited and non-inherited diseases, we can help you make more informed and responsible treatment decisions with your patients.

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