

COLLECTION DATE (REQUIRED)

If date of collection is not provided, three calendar days before specimen receipt will be used (for specimens stored longer than 30 days, the day of archive retrieval will be used as the date of service)

PLEASE SUBMIT THE FOLLOWING WITH THE TRF:

1. Clinic Notes 2. Pedigree 3. Insurance Card

PATIENT INFORMATION

Legal Name (Last, First, MI)		Date of Birth (MM/DD/YY)	Sex Assigned at Birth ☐ F ☐ M	Gender (optional) ☐ Man ☐ Woman ☐ Nonbinary ☐ Self-described
Genetic Ancestry: ☐ Ashkenazi Jewish ☐ Asian ☐ Black/African American ☐ French Canadian/Cajun ☐ Hispanic/Latino ☐ Mediterranean ☐ Middle Eastern ☐ Native American ☐ Pacific Islander ☐ Portuguese ☐ White ☐ Unknown ☐ Other:				MRN
Address		City	State	Zip
Mobile #		Email		

SPECIMEN INFORMATION (Please see ambrygen.com/specimen-requirements for details)

☐ Personal history of allogenic bone marrow or peripheral stem cell transplant

Specimen ID	Medical Record #
Collection Assistance: ☐ Phlebotomy draw* ☐ Send saliva kit to patient ☐ Send buccal kit to patient ☐ Insurance preverification first (available for ExomeNext® and GenomeNext™)	
* As the patient's clinician, I am unaware of any potential for complication or difficulty in drawing blood for the listed patient(s). I understand that the phlebotomist has full authority to refuse to draw any patient if the safety of the phlebotomist and/or patient(s) are in question.	

INDICATION(S) FOR TESTING

ICD-10 code(s):	Will medical management change depending upon the results of the test? ☐ Yes ☐ No
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ORDERING PHYSICIAN/SENDING FACILITY (Each listed person will receive a copy of the report)

Facility Name (Facility Code)	Address	City	State /Country	Zip	Phone
Ordering Licensed Provider Name (Last, First)(Code)	NPI#	Phone	Fax/Email		
Genetic Counselor or Other Medical Provider Name (Last, First) (Code)		Phone/Fax/Email			

CONFIRMATION OF INFORMED CONSENT, PRE-TEST GENETIC COUNSELING, AND MEDICAL NECESSITY FOR GENETIC TESTING

I confirm that the genetic test ordered is medically appropriate. All information on this TRF is true to the best of my knowledge. I also confirm that the patient has consented to proceed with genetic testing, including the transfer and processing of their sample and personal/sensitive information in the United States. I agree to allow Ambry Genetics to facilitate the provision of pre-test genetic counseling services by a third-party service, as required by the patient's insurance provider.

Signature Required for Processing Medical Professional Signature: _____ **Date:** _____

INSURANCE BILLING (Include copy of both sides of insurance card)

☐ **INSTITUTIONAL BILLING**

Patient Relation to Policy Holder? ☐ Self ☐ Spouse ☐ Child	Name and DOB of Policy Holder (if not self)	Facility Name	☐ Send invoice to facility address above
Insurance Company	Policy #	HMO Auth #	Address
Special Billing Notes:		Contact Name	
		Phone Number	E-mail/Fax
		☐ PATIENT PAYMENT ☐ Check (Payable to Ambry Genetics) ☐ Credit Card (Call 949-900-5795)	

Patient Acknowledgement: I acknowledge that the information provided by me is true and correct. For direct insurance billing: I authorize my insurance benefits to be paid directly to Ambry Genetics Corporation (Ambry), authorize Ambry to release medical information concerning my testing to my insurer, to be my designated representative for purposes of appealing any denial of benefits as needed and to request additional medical records for this purpose. I understand that I am financially responsible for any amounts not covered by my insurer and responsible for sending Ambry money received from my health insurance company.

☐ I agree to be contacted regarding future research studies for which I may be a candidate. Any future research projects will be subject to a separate informed consent process and participation is voluntary. Learn more about Ambry's privacy practices at <https://www.ambrygen.com/legal/notice-of-privacy-practices>.

For patient payment by credit card: I hereby authorize Ambry Genetics Corporation to bill my credit card as indicated above.

For NY Residents: ☐ I understand that New York State law requires Ambry Genetics to destroy my sample at the end of the testing process or not more than sixty days after the sample was taken. By checking this box, I agree that Ambry Genetics will instead retain my sample for at least 6 months after the testing above has been completed, and may (a) retain and use samples and health information for an indefinite period of time in accordance with applicable law; and (b) de-identify such samples and information and use and share the resulting de-identified samples and information in accordance with applicable law.

Signature Required For Insurance/Self-Pay Patients and NY Sample Storage Consent: Patient Signature _____ **Date:** _____

Neurology Test Requisition Form

PATIENT HISTORY ☐ No personal history of neurological disease		
PLEASE SUPPLY CLINIC NOTES AND PEDIGREE If pregnant, due date: _____ Upcoming procedure date: _____		
Reasons for Testing:		
Birth and Neonatal History ☐ N/A Gestational age at birth: _____ Birth weight: _____ Head circumference at birth (if available): _____ Developmental History ☐ N/A Developmental delay: ☐ Yes ☐ No ☐ Unknown Type of delay (choose all that apply): ☐ Motor ☐ Language ☐ Global Intellectual disability: ☐ Yes ☐ No ☐ Unknown Regression or plateau: ☐ Yes ☐ No ☐ Unknown Does patient meet DSM-V diagnostic criteria for an autism spectrum disorder? ☐ Yes ☐ No Seizure History ☐ N/A Age at first unprovoked seizure: _____ Has this patient been diagnosed with an epilepsy syndrome? ☐ Yes ☐ No ☐ Unknown If yes, please specify: _____	Other History ☐ N/A Hypo-/hyperpigmentation: ☐ Yes ☐ No Telangiectasias: ☐ Yes ☐ No Other skin abnormality, type: _____ Brain tumor, type: _____ Nerve tumor, type: _____ Other tumor, type: _____ Other Clinical Findings (choose all that apply) ☐ Ataxia ☐ Macrocephaly ☐ Psychiatric disorder ☐ Dysmorphic features ☐ Microcephaly ☐ Spasticity ☐ Hearing disorder ☐ Migraine ☐ Vision disorder ☐ Hypotonia ☐ Movement disorder Prior Testing: _____	
Previously Reported Variants: See instructions for reporting of Previously Reported Variants on the Supplemental Information Page		
Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____		
Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____		
Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____		
Known Familial Variant: Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____		
PROBAND'S CLINICAL OVERVIEW (Check yes for all that apply)		
☐ Yes ☐ No Audiologic/Otolaryngologic ☐ Yes ☐ No Cardiovascular ☐ Yes ☐ No Craniofacial ☐ Yes ☐ No Dental ☐ Yes ☐ No Dysmorphic Features ☐ Yes ☐ No Dermatologic ☐ Yes ☐ No Endocrine ☐ Yes ☐ No Fetal (Please complete and attach "ExomeNext Prenatal Form") ☐ Yes ☐ No Gastrointestinal ☐ Yes ☐ No Genitourinary ☐ Yes ☐ No Growth Disorders: ☐ Yes ☐ No Undergrowth ☐ Yes ☐ No Overgrowth ☐ Yes ☐ No Failure to thrive	☐ Yes ☐ No Hematologic ☐ Yes ☐ No Immunologic/Infectious/Allergy ☐ Yes ☐ No Metabolic/Biochemical ☐ Yes ☐ No Movement Disorder ☐ Yes ☐ No Musculoskeletal/Structural ☐ Yes ☐ No Multiple Congenital Anomalies ☐ Yes ☐ No Neurologic ☐ Yes ☐ No Seizures/Epilepsy ☐ Yes ☐ No Autism Spectrum Disorder ☐ Yes ☐ No Developmental Delay/Intellectual disability ☐ Yes ☐ No Ataxia/Spasticity ☐ Yes ☐ No Psychiatric ☐ Yes ☐ No Abnormal brain MRI ☐ Yes ☐ No Obstetric ☐ Yes ☐ No Oncologic	☐ Yes ☐ No Ophthalmologic ☐ Yes ☐ No Pulmonary ☐ Yes ☐ No Renal ☐ Yes ☐ No Tone abnormalities ☐ Yes ☐ No Hypotonia ☐ Yes ☐ No Hypertonia

Neurology Test Requisition Form

For Reflex or Concurrent Testing: See Reflex or Concurrent Testing section of the Supplemental Information page.

Test 1: _____ ☐ Reflex to _____ Test 2: _____ ☐ Reflex to _____ Test 3: _____
☐ Concurrent with _____ ☐ Concurrent with _____

Check	Test Name	Test Code	Description
Exome			
! REQUIRED: Select a Primary Test Order			
☐	ExomeNext® ☐ Proband only ☐ Duo ☐ Trio	9900	Exome sequencing
☐	ExomeNext-Rapid®	9999-R	Rapid trio exome sequencing including a defined list of established disease-causing variants in the mitochondrial DNA (mtDNA)
ExomeNext Supplemental Test Options (Primary test order required. See descriptions for details.)			
☐	ACMG Secondary Findings* ☐ Decline	9920	Analysis of genes included in the ACMG Recommended List of secondary findings. Secondary findings results are available free of charge for the proband and each family member who is fully sequenced as part of the duo/trio.
☐	ExomeReveal® RNA Analysis	9990	RNA analysis is available with all ExomeNext orders except for ExomeNext-Rapid. EDTA and PAX-gene RNA tubes are required.
☐	Mito DNA	9900-M	Analysis of a defined list of established disease-causing variants in the mitochondrial DNA (mtDNA)

Check	Test Name	Test Code	Description
Genome			
! REQUIRED: Select a Primary Test Order			
☐	GenomeNext™ ☐ Proband only ☐ Duo ☐ Trio	7000	Genome sequencing including sequencing of mitochondrial DNA (mtDNA)
GenomeNext Supplemental Test Options (Primary test order required.)			
☐	ACMG Secondary Findings* ☐ Decline	7002	Analysis of genes included in the ACMG Recommended List of secondary findings. Secondary findings results are available free of charge for the proband and each family member who is fully sequenced as part of the duo/trio.
☐	GenomeReveal™ RNA Analysis	7004	RNA analysis is available with all GenomeNext orders. EDTA and PAX-gene RNA tubes are required. Genome Reveal will be added automatically if a PAX tube is provided.

Check	Test Name	Test Code	Description
Fragile X syndrome and Chromosomal Microarray			
☐	Fragile X syndrome	4544	FMR1 repeat expansion analysis and methylation studies
☐	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)

*Secondary Findings: Check "decline" to opt-out of the ACMG Recommended List of secondary findings. If left unchecked, secondary findings will be reported.

FAMILY MEMBER INFORMATION (Completion of this section is required for order including parental samples. If available, please also submit a 3-generation pedigree)				
Relative	Name	DOB	Affected status**	Samples included?
			☐ Yes ☐ No	☐
			☐ Yes ☐ No	☐

** If affected, please list symptoms and age at diagnosis:

Check	Test Name	Test Code	Description
Epilepsy			
☐	EpilepsyNext®	6864	Multigene panel test for epilepsy
☐	EpilepsyNext-Expanded™	6865	Multigene panel test for genes associated with seizures, primarily with neonatal to childhood onset
Neurodevelopmental Disorders			
☐	AutismNext®	6863	Multigene panel test for non-syndromic autism spectrum disorders and/or intellectual disability
☐	Autism, macrocephaly	2106	PTEN
☐	NeurodevelopmentNext™	6861	Multigene panel test for genes known to cause developmental delay, intellectual disability, and/or autism spectrum disorders
Hereditary Neuropathy			
☐	Familial transthyretin amyloidosis	1560	TTR
Neurocutaneous/Neuro-Oncology Disorders			
Testing is available for Neurocutaneous and Neuro-Oncology disorders (such as neurofibromatosis and tuberous sclerosis) using our Cancer or Comprehensive requisition forms available at: www.ambrygen.com/providers/forms			
KNOWN VARIANT ANALYSIS (Please include a copy of relative's report)			
Gene(s): _____ Variant(s) (c. and/or p.): _____			
Relative Name: _____			
Relationship to Relative: _____ Accession # (If tested at Ambry): _____			
Positive control sample: ☐ Will be provided ☐ Already at Ambry ☐ Not available			
FOR PRENATAL SPECIMENS, POC OR CORD BLOOD: MATERNAL CELL CONTAMINATION ANALYSIS REQUIRED			
Both test codes required for fetal specimens.c			
☐ 1260 MCC for fetal specimen or cord blood			
☐ 1262 MCC Reference for maternal blood sample (No Charge)			

☐ **Opt-in to Reporting of Variants of Unknown Significance (VUS)**

For patients undergoing an epilepsy or neurodevelopmental disorder panel, checking this box indicates that VUS identified on the test(s) ordered above will be reported for this patient. If you do not check this box, VUS will not be reported.

☐ **Parental samples provided for cosegregation**

Cosegregation testing of family members is available for the following panels: EpilepsyNext, EpilepsyNext-Expanded, AutismNext, NeurodevelopmentNext

Supplemental Information

Electronic ordering is also available via AmbryPort®. Please visit <https://portal.ambrygen.com/login> to log in to the online portal.

Neurology Multi-Gene Tests

For current neurology panel gene content, please visit www.ambrygen.com/providers/neurology/test-menu (linked to QR code below)



Scan for current neurology
panel gene content

Sample Requirements

Blood/saliva from patients with a history of allogenic bone marrow or stem cell transplant cannot be used for genetic testing. Blood/saliva from patients with active hematological disease is not recommended. An alternative specimen may be needed. Please see ambrygen.com/specimen-requirements for details

Buccal swab samples from patients with a history of allogenic bone marrow or stem cell transplant should not be used for genetic testing. For these patients, an alternative specimen (e.g. cultured fibroblasts) is required. Testing on buccal swab samples from patients with active hematological disease is not recommended. An alternative specimen (e.g. cultured fibroblasts) is recommended. Please see ambrygen.com/specimen-requirements for details.

Fetal specimens, cord blood and POC will have maternal cell contamination (MCC) studies added for a charge. Maternal and fetal specimen required. Please see page 3 for MCC test codes. Fetal specimens, cord blood, and POC are not accepted sample types for GenomeNext™.

Known variant analysis should be accompanied by a copy of the original testing report or internal Ambry testing information (internal Ambry ID, Name/DOB). Please review information about positive controls and other specifics at ambrygen.com/knownvariantanalysis.

Previously Reported Variants

ExomeNext® or GenomeNext™ report comments about the presence or absence of a variant previously reported **in the patient** require the “Previously Reported Variant” section of this form to be completed accurately, including an internal Ambry reference ID and/or a copy of the positive family member’s lab report. Acceptable types of Ambry identifiers include:

- Accession number
- Order number
- Name and date of birth

Previously reported variant requests without an internal Ambry reference ID or positive lab report will not receive a variant-specific report comment.

Variant-specific report comments about the presence or absence of a variant previously reported **in a family member** are not included in ExomeNext, GenomeNext, or Neurology panel reports.

Reflex or Concurrent Testing

Concurrent testing is when multiple tests are initiated at the same time. When multiple tests are ordered on the same test requisition form, testing will be run concurrently unless otherwise specified.

Reflex testing is when a subsequent test is initiated pending the outcome of the initial test. Reflex testing may result in delayed reporting of results.

For reflex test orders:

- Any diagnostic finding at any step will result in cancellation of any subsequent reflex tests.
- Non-diagnostic findings (including VUS or Uncertain results) will automatically reflex to the subsequent test.
- Secondary findings results do not impact whether a subsequent test is initiated or canceled.

ExomeNext Medical Necessity Form

REQUIRED FOR INSURANCE ORDERS ONLY (NOT REQUIRED FOR CIGNA MEMBERS)

This form is ONLY required if you are requesting reflex to Exome sequencing and wish to have the patient's insurance billed. Please complete and submit with the TRF and a copy of clinical notes. This form replaces the Letter of Medical Necessity.

1. Has the patient had previous Whole Exome Sequencing (WES) performed?

☐ Yes, date performed: _____

☐ No

2. Does this patient have a clinical presentation consistent with the following (select all that apply):

☐ Multiple abnormalities affecting unrelated organ systems (please specify): _____

OR two of the following:

☐ Abnormality affecting a single organ system(specify): _____

☐ Significant intellectual disability, symptoms of a complex neurodevelopmental disorder (i.e. self-injurious behavior, reverse sleep-wake cycle, or seizure/epilepsy), or severe neuropsychiatric condition (e.g. schizophrenia, bipolar, Tourette syndrome)

☐ Family history strongly implicating a genetic etiology (please specify findings and relationships)

☐ Period of unexplained developmental regression (unrelated to autism or epilepsy)

3. Are the results of this WES test expected to directly influence this patient's medical management recommendations and clinical outcome?

☐ Yes (please describe): _____

☐ No

4. Please describe the genetic tests that would be indicated if WES were NOT performed (i.e., single gene tests, gene panels, etc.):

☐ Chromosomal microarray

☐ Single gene test(s): _____

☐ Multigene panel(s): _____

☐ Other genetic test(s): _____

5. Please describe follow-up procedures & frequency that would be needed if WES were NOT performed (i.e., lumbar puncture, imaging studies, brain MRI, etc.):

☐ Imaging study: _____

☐ Surgery: _____

☐ Biopsy: _____

☐ Other: _____