

To submit an order via email, please send the completed test requisition form to [info@ambrygen.com](mailto:info@ambrygen.com)

**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**

Name (Last, First, MI)		Sex at Birth <input type="checkbox"/> F <input type="checkbox"/> M	Date of Birth (MM/DD/YY)	MRN
Ethnicity: <input type="checkbox"/> Asian <input type="checkbox"/> Ashkenazi Jewish <input type="checkbox"/> Black/African American <input type="checkbox"/> White <input type="checkbox"/> French Canadian/Cajun <input type="checkbox"/> Hispanic/Latino <input type="checkbox"/> Mediterranean <input type="checkbox"/> Middle Eastern <input type="checkbox"/> Native American <input type="checkbox"/> Pacific Islander <input type="checkbox"/> Portuguese <input type="checkbox"/> Unknown <input type="checkbox"/> Other:				
Address		City	State	Zip
Phone		Email		

**SPECIMEN INFORMATION** (Please see [ambrygen.com/specimen-requirements](http://ambrygen.com/specimen-requirements) for details)

Personal history of allogenic bone marrow or peripheral stem cell transplant

Specimen ID	Medical Record #
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Collection Assistance:  Phlebotomy draw\*    Send saliva kit to patient   |    Insurance preverification first (available for ExomeNext and SNP array only)  
*\* 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? <input type="checkbox"/> Yes <input type="checkbox"/> No
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**PATIENT HISTORY**    No personal history of neurological disease

**PLEASE SUPPLY CLINIC NOTES AND PEDIGREE**   If pregnant, due date: \_\_\_\_\_   Upcoming procedure date: \_\_\_\_\_

Reasons for Testing:

<p><b>Birth and Neonatal History</b>   <input type="checkbox"/> N/A</p> <p>Gestational age at birth: _____ Birth weight: _____</p> <p>Head circumference at birth (if available): _____</p> <p><b>Developmental History</b>   <input type="checkbox"/> N/A</p> <p>Developmental delay:   <input type="checkbox"/> Yes   <input type="checkbox"/> No   <input type="checkbox"/> Unknown</p> <p>Type of delay (choose all that apply):   <input type="checkbox"/> Motor   <input type="checkbox"/> Language   <input type="checkbox"/> Global</p> <p>Intellectual disability:   <input type="checkbox"/> Yes   <input type="checkbox"/> No   <input type="checkbox"/> Unknown</p> <p>Regression or plateau:   <input type="checkbox"/> Yes   <input type="checkbox"/> No   <input type="checkbox"/> Unknown</p> <p>Does patient meet DSM-V diagnostic criteria for an autism spectrum disorder?   <input type="checkbox"/> Yes   <input type="checkbox"/> No</p> <p><b>Seizure History</b>   <input type="checkbox"/> N/A   Age at first unprovoked seizure: _____</p> <p>Has this patient been diagnosed with an epilepsy syndrome?   <input type="checkbox"/> Yes   <input type="checkbox"/> No   <input type="checkbox"/> Unknown</p> <p>If yes, please specify: _____</p>	<p><b>Other History</b>   <input type="checkbox"/> N/A</p> <p>Hypo-/hyperpigmentation:   <input type="checkbox"/> Yes   <input type="checkbox"/> No   Telangiectasias:   <input type="checkbox"/> Yes   <input type="checkbox"/> No</p> <p>Other skin abnormality, type: _____</p> <p>Brain tumor, type: _____ Nerve tumor, type: _____</p> <p>Other tumor, type: _____</p> <p><b>Other Clinical Findings (choose all that apply)</b></p> <table border="0"> <tr> <td><input type="checkbox"/> Ataxia</td> <td><input type="checkbox"/> Macrocephaly</td> <td><input type="checkbox"/> Psychiatric disorder</td> </tr> <tr> <td><input type="checkbox"/> Dysmorphic features</td> <td><input type="checkbox"/> Microcephaly</td> <td><input type="checkbox"/> Spasticity</td> </tr> <tr> <td><input type="checkbox"/> Hearing disorder</td> <td><input type="checkbox"/> Migraine</td> <td><input type="checkbox"/> Vision disorder</td> </tr> <tr> <td><input type="checkbox"/> Hypotonia</td> <td><input type="checkbox"/> Movement disorder</td> <td></td> </tr> </table>	<input type="checkbox"/> Ataxia	<input type="checkbox"/> Macrocephaly	<input type="checkbox"/> Psychiatric disorder	<input type="checkbox"/> Dysmorphic features	<input type="checkbox"/> Microcephaly	<input type="checkbox"/> Spasticity	<input type="checkbox"/> Hearing disorder	<input type="checkbox"/> Migraine	<input type="checkbox"/> Vision disorder	<input type="checkbox"/> Hypotonia	<input type="checkbox"/> Movement disorder	
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Prior Testing:

**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**

The undersigned person (or representative thereof) ensures he/she is a licensed medical professional authorized to order genetic testing and confirms that the patient has given appropriate consent. I confirm that testing is medically necessary and that test results may impact medical management for the patient. I agree to allow Ambyr Genetics to facilitate the provision of pre-test genetic counseling services by a third-party service, as required by the patient's insurance provider. Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the attached letter of medical necessity.

Signature Required for Processing   Medical Professional Signature: \_\_\_\_\_   Date: \_\_\_\_\_

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

Patient Relation to Policy Holder? <input type="checkbox"/> Self <input type="checkbox"/> Spouse <input type="checkbox"/> Child	Name and DOB of Policy Holder (if not self)
Insurance Company	Policy #      HMO Auth #

**INSTITUTIONAL BILLING**

Facility Name	<input type="checkbox"/> Send invoice to facility address above
Address	

Special Billing Notes:

Contact Name	
Phone Number	E-mail/Fax
<input type="checkbox"/> <b>PATIENT PAYMENT</b>	<input type="checkbox"/> Check (Payable to Ambyr Genetics) <input type="checkbox"/> 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 Ambyr Genetics Corporation (Ambyr), authorize Ambyr 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 Ambyr money received from my health insurance company.  
**For patient payment by credit card:** I hereby authorize Ambyr Genetics Corporation to bill my credit card as indicated above. In order to expedite consideration for eligibility for Ambyr's Patient Assistance Program, please provide the total annual gross household income: \$ \_\_\_\_\_ and the number of family members in the household supported by the listed income: \_\_\_\_\_. I authorize Ambyr Genetics Corporation to verify the above information for the sole purpose of assessing financial need, including the right to seek supporting documentation.

**For NY Residents:**  By checking this box, I agree that Ambyr Genetics will retain my sample for 6 months after the testing above has been completed. By not checking this box, I understand that under New York State law, Ambyr Genetics must discard my sample after the longer of (a) testing completion and (b) 60 days after the Date of Collection above.

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

# Neurology Test Requisition Form

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

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

**For Reflex or Concurrent Testing:**

Test 1: \_\_\_\_\_  Reflex to Test 2: \_\_\_\_\_  
 Concurrent with

Check	Test Name	Test Code	Description
<b>Comprehensive Testing</b>			
Must order through Ambry-Port®	CustomNext-Neuro™	9545	Customizable test of up to 500 genes from neuro menu
<b>Epilepsy</b>			
<input type="checkbox"/>	EpiRapid®	6862	22 epilepsy genes with treatment associations <input type="checkbox"/> Check if parental samples are included
<input type="checkbox"/>	EpilepsyNext®	6864	124 genes for epilepsy <input type="checkbox"/> Check if parental samples are included
<input type="checkbox"/>	EpilepsyNext-Expanded™	6865	>890 genes associated with seizures, primarily with neonatal to childhood onset <input type="checkbox"/> Check if parental samples are included
<b>Clinical Genomics</b>			
<input type="checkbox"/>	SNP Array <i>Buccal swab accepted</i>	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)
<input type="checkbox"/>	Familial targeted microarray <i>Buccal swab accepted</i>	5495	Paid option. Only available following SNP Array (5490) completed at Ambry. Incidental findings unrelated to the variant(s) detected in the proband, will NOT be reported. Name of proband tested at Ambry: _____
<input type="checkbox"/>	ExomeNext®-Proband	9993	Proband only exome sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
<input type="checkbox"/>	ExomeNext®-Proband plus mtDNA	9994	Proband only exome sequencing plus mtDNA sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
<input type="checkbox"/>	ExomeNext®-Duo	9991	Duo exome sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
<input type="checkbox"/>	ExomeNext®-Duo plus mtDNA	9992	Duo exome sequencing plus mtDNA sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
<input type="checkbox"/>	ExomeNext®-Trio	9995	Trio exome sequencing plus mtDNA sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
<input type="checkbox"/>	ExomeNext®-Trio plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
<input type="checkbox"/>	ExomeNext®-Rapid® <i>(Institutional billing or patient payment only)</i>	9999R	Rapid Trio exome sequencing plus mtDNA sequencing Secondary Findings <sup>†</sup> : <input type="checkbox"/> Opt-out
Order through Ambry-Port	ExomeNext®-Select	9500	Up to 500 gene custom exome sequencing test

<sup>†</sup> Secondary Findings: If box is left unchecked, the ACMG recommended list of Secondary Findings will be reported. Secondary Findings are not available for ExomeNext-Select orders.

Check	Test Name	Test Code	Description
<b>Migraine</b>			
<input type="checkbox"/>	Familial hemiplegic migraine	6866	ATP1A2, CACNA1A, PRRT2, SCN1A <input type="checkbox"/> Check if parental samples are included
<b>Neurodevelopmental Disorders</b>			
<input type="checkbox"/>	AutismNext®	6863	72 genes for non-syndromic autism spectrum disorders and/or intellectual disability <input type="checkbox"/> Check if parental samples are included
<input type="checkbox"/>	Autism, macrocephaly	2106	PTEN
<input type="checkbox"/>	Fragile X syndrome	4544	FMR1 repeat expansion analysis and methylation studies
<input type="checkbox"/>	NeurodevelopmentNext™	6861	202 genes known to cause developmental delay, intellectual disability, and/or autism spectrum disorders <input type="checkbox"/> Check if parental samples are included
<b>Hereditary Neuropathy</b>			
<input type="checkbox"/>	Familial transthyretin amyloidosis	1560	TTR
<b>Neurocutaneous/Neuro-Oncology Disorders</b>			
<input type="checkbox"/>	Ataxia-telangiectasia	9014	ATM
<input type="checkbox"/>	BrainTumorNext®	8847	27 genes for brain tumors
<input type="checkbox"/>	HHTNext®	8672	ACVRL1, ENG, EPHB4, SMAD4, GDF2, RASA1
<input type="checkbox"/>	Legius syndrome	5724	SPRED1
<input type="checkbox"/>	Li-Fraumeni syndrome	2866	TP53
<input type="checkbox"/>	Neurofibromatosis 1	5704	NF1
<input type="checkbox"/>	Neurofibromatosis 2	9024	NF2
<input type="checkbox"/>	Nevoid basal cell carcinoma syndrome/ Gorlin syndrome	5684	PTCH1
<input type="checkbox"/>	Schwannomatosis	7180	SMARCB1
<input type="checkbox"/>	Tuberous sclerosis	5904	TSC1, TSC2
<input type="checkbox"/>	von Hippel-Lindau	2606	VHL

**SINGLE SITE ANALYSIS (Please include a copy of relative's report)**

Gene(s): \_\_\_\_\_ Mutation(s): \_\_\_\_\_  
 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.

1260 MCC for fetal specimen or cord blood  
 1262 MCC Reference for maternal blood sample (No Charge)

**FAMILY HISTORY (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 <sup>††</sup>	Samples included?
			<input type="checkbox"/> Yes <input type="checkbox"/> No	<input type="checkbox"/>
			<input type="checkbox"/> Yes <input type="checkbox"/> No	<input type="checkbox"/>

<sup>††</sup> If affected, please list symptoms and age at diagnosis:

**Note:** Gene lists for EpilepsyNext-Expanded are updated annually due to proactive review of current literature using an internal, peer-reviewed clinical validity scheme (Smith ED, Radtke K, Rossi M, et al. 2017 Human mutation 38(5):600-608). The patient's test report will include a list of genes evaluated. For up-to-date gene lists, visit ambrygen.com

## Supplemental Information

### 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](http://ambrygen.com/specimen-requirements) for details

Buccal swab sample available for chromosomal microarray (SNP array, familial targeted microarray), CustomNext-*Neuro*, epilepsy, ExomeNext, fragile X syndrome, hereditary neuropathy (familial transthyretin amyloidosis), HHTNext, migraine (familial hemiplegic migraine), and neurodevelopmental disorder tests. 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](http://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 2 for MCC test codes.

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