Athena Diagnostics Neurome™ Neurological Exome Requisition (July 2015)

athena diagnostics

Tests

	TEST CODE
☐ Neurome™ Neurological Exome (Proband)	1500
☐ Neurome™ Neurological Exome (Trio)+	1501
Specimen is: ☐ Proband ☐ Family Member	
☐ Family Testing Supporting Neurome™ Analysis+	1509

(If selected, please fill out "Variant Details" and Proband Information" sections)

Patient Information

Patient Name _____ (First Name / Middle Initial / Last Name)

Date of Birth ____ / ____ / ____ (Month / Day / Year)

Address _____ (Street)

City _____ State _____ Zip _____

Phone _____ _____ (Alternate Phone)

MRN # _____

Gender ☐ Male ☐ Female ☐ Unknown

Ethnicity ☐ African American/Black ☐ Jewish (Ashkenazi) ☐ Asian ☐ Middle Eastern ☐ Caucasian ☐ Native American ☐ Hispanic ☐ _____ (Other, please specify)

Known Consanguinity ☐ No ☐ Yes

History of Bone Marrow Transplant? ☐ No ☐ Yes

(Please contact Athena Diagnostics if yes prior to submission)

Ordering Physician

Physician/Genetic Counselor _____

Institution _____

Address _____ (Street)

City _____ State _____ Zip _____

NPI # _____ Email _____

Phone _____ Fax _____

Patient Specimen

Specimen ID _____ Specimen Type* ☐ Blood

Specimen Collection Date ____ / ____ / ____ (Month / Day / Year)

Authorization to Use De-identified Specimen for Research. To promote medical understanding and develop better health insights, Athena Diagnostics requests your permission to use your specimen in a de-identified way (without identifying information) for research, educational studies, commercial purposes and/or publication, if appropriate. Your name or other personal identifying information will not be used in or linked to the results of any studies and publications. Your refusal to have your specimen used or not used for research purposes will not affect processing or testing of your specimen, your test results or the service support provided by Athena Diagnostics to your physician. Please indicate your approval by checking the box next to **Yes** or denial by checking the box next to **No**.

I consent to the use of my de-identified specimen for research: ☐ **Yes** ☐ **No**

_____ Signature of Patient, Parent or Legally Authorized Representative — Date

_____ Printed Name of Patient, Parent or Legally Authorized Representative — Date

_____ Relationship to Patient if Signatory is Someone Other than Patient

Additional Contacts

Physician/Genetic Counselor 1 _____

Phone _____

Address _____ (Street)

City _____ State _____ Zip _____

Physician/Genetic Counselor 2 _____

Phone _____

Address _____ (Street)

City _____ State _____ Zip _____

Institutional Billing

Facility Name _____

Contact Name _____ Phone _____

Address _____ (Street)

City _____ State _____ Zip _____

+Family Members Submitted for Testing

Please complete a separate requisition form for each family member submitted.

Full Name _____ Full Name _____

Relationship to Proband _____ Relationship to Proband _____

Specimen Type ☐ Blood Specimen Type ☐ Blood

Affected ☐ No ☐ Yes Affected ☐ No ☐ Yes

+If you selected the Trio or Family Testing, please fill out the above section "Family Members Submitted for Testing," and provide the proband's previous test results on page 3.

*8mL (6mL minimum) whole blood collected in an EDTA (lavender top) tube. Pediatric: 4mL (2mL minimum).

Variant Details

Gene _____ Nucleotide Coordinate _____

Gene _____ Nucleotide Coordinate _____

Gene _____ Nucleotide Coordinate _____

Additional _____

Proband Information

Proband Name _____

Date of Birth ____ / ____ / ____ (Month / Day / Year)

Personalis Accession # _____

Relationship of Patient to Proband _____

Indications for Testing (Check One) ☐ Diagnostic (symptomatic) ☐ Family Testing

Physician Attestation of Informed Consent

Verification of patient informed consent is required for genetic testing. Informed consent applies to physician practices in 12 states (AK, AZ, DE, GA, MA, MN, NV, NH, NJ, NM, SD, VT).

Note: The signed acknowledgement below is only required to complete the genetic testing ordered if you have not previously signed a blanket Physician Attestation of Informed Consent (PAIC) at any Quest lab. The company offers a blanket PAIC that can be signed for all future orders.

I warrant that I have obtained both oral and written consent using the **Patient Informed Consent Form for Genetic Testing** with the **Neurome Neurological Exome Test and/or Targeted Site-Specific Genetic Test** provided by Athena Diagnostics or another patient informed consent form. This written consent was signed by the person who is the subject of the test (or if that person lacks capacity to consent, signed by the person authorized to consent for that person).

_____ Medical Practitioner Signature — Date

_____ Printed Name of Medical Practitioner — NPI #

NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, last four digits of SS#, patient ID no. These same two forms of ID must be indicated on the test requisition.

Note: Testing performed at Personalis, Inc. 1330 O'Brien Drive, Menlo Park, CA 94025

Athena Diagnostics, Inc., 200 Forest Street, 2nd Floor, Marlborough, MA 01752 • 800-394-4493 • Fax 610-271-6085 • AthenaDiagnostics.com

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Clinical Features for Patient

Please fill in as completely as possible to aid diagnosis. Please provide available clinic notes and pedigree information along with this requisition form.

☐ Environmental History
- ☐ Exposure to lead
- ☐ Exposure to viral agents, specify:
- ☐ History of trauma, specify:
- ☐ Other:

☐ Prenatal/Perinatal History
- ☐ Cystic hygroma/increased NT
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Cerebral palsy
- ☐ Complicated delivery, specify:
- ☐ Prematurity, specify:
- ☐ Hypoxic ischemic encephalopathy
- ☐ Other:

☐ CNS/Neurological
- ☐ Abnormal nerve conduction velocity, specify:
- ☐ Ataxia
- ☐ Chorea
- ☐ Congenital neuropathy
- ☐ Cranial nerve abnormalities, specify:
- ☐ Dysmyelination, specify:
- ☐ Dystonia
- ☐ Foot drop
- ☐ Motor neuropathy
  - ☐ Proximal
  - ☐ Distal
- ☐ Pes cavus
- ☐ Pressure palsy
- ☐ Recurrent headache/migraine
- ☐ Reduced/absent deep tendon reflexes
- ☐ Sensory neuropathy
- ☐ Sleep apnea
- ☐ Spasticity
- ☐ Stroke/stroke-like episodes
- ☐ Tremor
- ☐ Vocal cord paresis
- ☐ Other:

☐ Seizures/Epilepsy
- ☐ Epileptic encephalopathy
- ☐ Febrile seizures
  - ☐ Dravet syndrome
- ☐ Focal seizures
- ☐ Generalized seizures
  - ☐ Absence
  - ☐ Clonic
  - ☐ Myoclonic
  - ☐ Tonic-clonic
- ☐ Infantile/epileptic spasms
  - ☐ Ohtahara syndrome
  - ☐ West syndrome
- ☐ Status epilepticus
- ☐ Other:

☐ Major Brain Anomalies
- ☐ Agenesis of the corpus callosum
- ☐ Basal ganglia abnormality
- ☐ Brain atrophy
- ☐ Cortical dysplasia
- ☐ Dandy-Walker malformation
- ☐ Encephalocele
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Lissencephaly
- ☐ Molar tooth sign
- ☐ Periventricular leukomalacia
- ☐ Periventricular nodular heterotopia
- ☐ Polymicrogyria
- ☐ Pontocerebellar hypoplasia
- ☐ Subcortical band heterotopia
- ☐ Other:

☐ Developmental Delay
- ☐ Intellectual disability/mental retardation
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe
- ☐ Developmental regression
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Speech delay
- ☐ Speech articulation difficulties
- ☐ Other:

☐ Behavioral
- ☐ Autism spectrum disorder/autistic features
- ☐ Self-injurious behavior
- ☐ Stereotypic behaviors
- ☐ Other:

☐ Muscular
- ☐ CPK abnormalities, specify:
- ☐ Dysphagia
- ☐ Easy fatigue
- ☐ Exercise intolerance
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Mobility limitations, specify:
- ☐ Muscle fasciculations
- ☐ Muscle wasting
- ☐ Muscle weakness, specify location:
- ☐ Myotonia

☐ Growth
- ☐ Microcephaly
  - ☐ Std Dev
  - ☐ Progressive/acquired
  - ☐ Congenital
- ☐ Macrocephaly
  - ☐ Std Dev
  - ☐ Progressive/acquired
  - ☐ Congenital
- ☐ Failure to thrive
- ☐ IUGR
- ☐ Obesity
- ☐ Overgrowth
- ☐ Short stature
  - ☐ Std Dev
- ☐ Tall stature
  - ☐ Std Dev
- ☐ Other:

☐ Craniofacial/Dysmorphology
- ☐ Abnormal hair, specify:
- ☐ Cleft lip +/- cleft palate, specify (unit/bilat):
- ☐ Craniosynostosis, specify:
- ☐ Deafness
  - ☐ Congenital
  - ☐ Progressive/acquired
  - ☐ Conductive
  - ☐ Sensorineural
  - ☐ Mixed
  - ☐ Unilateral
  - ☐ Bilateral
- ☐ External ear malformation, specify:
- ☐ Dysmorphic facies, describe:
- ☐ Other:

☐ Skeletal/Limb Abnormalities
- ☐ Contractures
- ☐ Polydactyly
- ☐ Syndactyly
- ☐ Scoliosis
- ☐ Vertebral anomaly
- ☐ Other:

☐ Ophthalmologic
- ☐ Blindness
- ☐ Cataracts
- ☐ Chronic progressive external ophthalmoplegia
- ☐ Coloboma
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinitis pigmentosa
- ☐ Visual impairment
- ☐ Other:

☐ Cardiac
- ☐ Arrhythmia, specify:
- ☐ Cardiomyopathy, specify:
- ☐ Structural heart malformation, specify:
- ☐ Syncope
- ☐ Other:

☐ Genitourinary Abnormalities
- ☐ Renal abnormality, specify:
- ☐ Other genitourinary abnormality:

☐ Abdominal/Gastrointestinal
- ☐ Abdominal wall defect
- ☐ Chronic diarrhea
- ☐ Constipation
- ☐ Pyloric stenosis
- ☐ Recurrent vomiting
- ☐ TE fistula
- ☐ Other:

☐ Skin/Autonomic
- ☐ Abnormal temperature regulation
- ☐ Altered sweating
- ☐ Abnormal nails, specify:
- ☐ Altered skin pigmentation, specify:
- ☐ Other:

☐ Hematologic/Immunologic
- ☐ Specify:

☐ Tumor/Growths:
- ☐ Medullary carcinoma of the thyroid
- ☐ Pheochromocytoma
- ☐ Other tumor type(s) and location(s):

☐ Endocrine/Metabolic
- ☐ Abnormal metabolic testing, specify:
- ☐ Abnormal newborn screen, specify:
- ☐ Adrenal insufficiency
- ☐ Adrenal hyperplasia
- ☐ Cushing syndrome
- ☐ Other abnormal testing:

☐ Test Results
- ☐ Abnormal CT, specify:
- ☐ Abnormal electromyography (EMG), specify:
- ☐ Abnormal electroencephalography (EEG), specify:
- ☐ Abnormal magnetic resonance imaging (MRI), specify:
- ☐ Muscle biopsy, specify results:
- ☐ Nerve biopsy, specify results:

Other Patient History

Other Clinical Features

Pertinent Negatives

Other Clinical Details
(Clarification of above features: age of onset, unilateral/bilateral, etc)

Family History

Clinical/Suspected Diagnoses

Clinical Diagnosis

Additional Suspected Genes

Additional Considered Diagnosis

ICD-9 Codes

†Previous Test Results

Summarize below or attach results

Karyotype/FISH

Genetic Testing

Array CGH

Other

Form Submission Instructions

When the form is complete please include a printed copy with each sample. Additionally, please e-mail the form to genetics@athenadiagnostics.com and fax the form to 508-802-5912. Samples must arrive in a kit with a printed copy of each form.

Ordering Checklist

☐ Completed requisition form

☐ Institutional billing account is established

☐ Included medical records, clinic note(s), and/or pedigree

☐ Retain a copy of all paperwork in the office

☐ Patient sample and requisition form submitted in appropriate shipping container(s) with two unique identifiers matching between the form and sample.

☐ Family member requisition form(s) submitted each in a separate container/box with the applicable sample and two unique identifiers matching between each form and sample.

Athena Diagnostics Client Service Representatives are available from 8:30am to 6:30pm Eastern Time (U.S.). Customers in the U.S. and Canada please call toll-free

800-394-4493

(Non-U.S. customers please call 508-756-2886 or fax 610-271-6085.)

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