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**LETTER OF MEDICAL NECESSITY**

**Ataxia Complete Recessive Evaluation**

**Instructions for Healthcare Provider:**

1. This letter template is being provided as a tool for clinicians to assist communication with payers.
2. Include specific patient information in the letter for this tool to be effective. The areas that must be edited/deleted are indicated in gray on the template
3. Print the template on the physician’s letterhead, **NOT** Athena letterhead. There should be no Athena branding on the letter.

[LMN: Ataxia Complete Recessive Evaluation (11.9.2021)]

<Date>

ATTN: <Medical Director/ Physician Name>, MD

<Institution/Insurance Company>

<Street Address>

<City>**,** <State> <zip code>

RE: <Patient Name>

DOB: <MM/DD/YYYY>

Member ID: <Insurance ID Number>

Group #: <Enter Group #>

Dear Doctor <Medical Director/ Physician Name>:

I am writing this letter on behalf of my patient <Patient Name> to request coverage for the Ataxia, Complete Recessive Evaluation offered by Athena Diagnostics®. This test analyzes 18 genes for pathogenic variants or repeat expansions associated with ataxias exhibiting an autosomal recessive (AR; 2 copies of a pathogenic alteration are required for disease) mode of inheritance. This letter documents the medical necessity for Ataxia, Complete Recessive Evaluation, in light of my patient’s medical history. Results from the test will be used to guide appropriate medical care for my patient.

I have determined that this test is medically necessary because of the following aspects of this patient’s history:

<Patient name> is a <age>-year-old <gender> with a suspected diagnosis of hereditary ataxia. Symptoms and clinical findings are consistent with this diagnosis:

1. <Symptom #1 with ICD-10 code>

2. <Symptom #2 with ICD-10 code>

<Symptoms should support diagnosis or risk of genetic disease. Relevant information may include results of prior testing, such as an MRI scan and tests for hypothyroidism, and results of a physical examination and patient consultation.>

<Note family or other personal history, or lack therof, if relevant. Consider including information on both neurological and non-neurological problems, such as movement disorders, spasticity, peripheral neuropathy, intellectual impairment, etc.>

**Rationale for testing**

Hereditary ataxias compose a group of diseases characterized by incoordination of speech and movement.1{Jayadev, 2013 #942} Obtaining a specific diagnosis is complex because genetic causes are highly heterogeneous and clinical symptoms frequently overlap among these diseases.1-3 Hereditary ataxias are broadly classified by mode of inheritance, predominantly as AD or AR ataxias, but confirming a diagnosis for a specific type of ataxia is difficult without a molecular diagnosis obtained through genetic testing.1 AR ataxias occur more frequently than AD ataxias, although many remain genetically undiagnosed since oftentimes only 1 individual in a family presents with an ataxia and/or the patient’s family history is unknown; these cases may appear sporadic or idiopathic.3,4

Obtaining a genetic diagnosis for a specific type of AR ataxia is important because certain types have treatments available, such as coenzyme Q10 (CoQ10) for ataxia with CoQ10 deficiency and high-dose vitamin E for ataxia with vitamin E deficiency (AVED).4,5 A genetic diagnosis can prevent misdiagnosis and subsequent mistreatment. For example, AVED and Friedreich’s ataxia (FRDA), the most common AR ataxia in the United States and Europe, can have similar clinical presentations. Thus, a misdiagnosis of FRDA instead of AVED could prevent patients from receiving appropriate treatment (high-dose vitamin E).3-5 A genetic diagnosis also allows for the prevention or treatment of potential complications (eg, cardiac, neurologic, or ocular) that are associated with various AR ataxias.1,6

Diagnostic rate can vary widely because commercially available tests differ in the genes tested and testing methods. This places the onus on the clinician to identify the most suitable test to obtain a diagnosis, which may contribute to delays in diagnosis and burden to the patient.7,8 The diagnostic odyssey can significantly decrease a patient’s psychological well-being and quality of life since it generally entails many visits to a succession of physicians and other health professionals, during which time the patient is suffering from distressing and disabling symptoms without knowing the cause.9 A study of patients with hereditary ataxia reported a mean delay of 18.1 years (range 3-35 years) from disease onset to molecular diagnosis.7 Prompt diagnosis can provide resolution and allow for genetic counseling, determination of prognosis, life and family planning, and enrollment in support groups and research activities.9,10 A panel specifically designed to detect the most common AR ataxias, such as the 18-gene Ataxia, Complete Recessive Evaluation, increases the likelihood that a patient with ataxia will receive a prompt, specific diagnosis instead of a prolonged and/or unsuccessful diagnostic odyssey that often results from a tiered testing approach.

The importance of obtaining a specific diagnosis for AR ataxia is reflected in professional organization clinical practice guidelines. In the case of a family history compatible with an AR cerebellar ataxia, the European Federation of Neurological Societies (EFNS) recommends expanded genetic testing after standard testing for the most common AR ataxias.4 For patients without known family history, a full AR ataxia diagnostic workup is recommended if the age of onset is below 45 years and after testing negative for FRDA.4 Ataxia UK, a patient support organization that developed guidelines in consultation with >30 health professionals, recommends expanded gene panels as second-line tests for patients who remain without a diagnosis after initial testing for FRDA.5 Therefore, current guidelines recommend expanded genetic testing within a tiered diagnostic approach, but extended gene panels are acknowledged as new tools that are expected to further improve diagnosis of AR ataxia.4,5

In summary, extended genetic testing with Ataxia, Complete Recessive Evaluation can improve the diagnostic yield for AR ataxia patients, which provides valuable information to both the patient and clinician for appropriate clinical management. I am requesting that <Patient Name> be approved for the Ataxia, Complete Recessive Evaluation (test code 6910; CPT codes 81286, 81284, 81404, 81405[2], 81406[3], 81408, 81479) offered by Athena Diagnostics.

I hope you will support this letter of medical necessity for <Patient Name>. Please feel free to contact me at <Physician Phone> if you have additional questions.

Sincerely,

<Physician Name>, MD

NPI #: <Physician NPI#>

Contact information:

< Address>

<City>**,** <State>, <zip code>

Contact phone: <phone number>

The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.

**References**

**1.** Jayadev S, Bird TD. Hereditary ataxias: overview. *Genet Med*. 2013;15(9):673-683. doi:10.1038/gim.2013.28

**2.** Sailer A, Houlden H. Recent advances in the genetics of cerebellar ataxias. *Curr Neurol Neurosci Rep*. 2012;12(3):227-236. doi:10.1007/s11910-012-0267-6

**3.** Sandford E, Burmeister M. Genes and genetic testing in hereditary ataxias. *Genes (Basel)*. 2014;5(3):586-603. doi:10.3390/genes5030586

**4.** van de Warrenburg BP, van Gaalen J, Boesch S, et al. EFNS/ENS Consensus on the diagnosis and management of chronic ataxias in adulthood. *Eur J Neurol*. 2014;21(4):552-562. doi:10.1111/ene.12341

**5.** de Silva R, Greenfield J, Cook A, et al. Guidelines on the diagnosis and management of the progressive ataxias. *Orphanet J Rare Dis*. 2019;14(1):51. doi:10.1186/s13023-019-1013-9

**6.** Beaudin M, Klein CJ, Rouleau GA, et al. Systematic review of autosomal recessive ataxias and proposal for a classification. *Cerebellum Ataxias*. 2017;4:3. doi:10.1186/s40673-017-0061-y

**7.** Németh AH, Kwasniewska AC, Lise S, et al. Next generation sequencing for molecular diagnosis of neurological disorders using ataxias as a model. *Brain*. 2013;136(Pt 10):3106-3118. doi:10.1093/brain/awt236

**8.** Wallace SE, Bird TD. Molecular genetic testing for hereditary ataxia: what every neurologist should know. *Neurol Clin Pract*. 2018;8(1):27-32. doi:10.1212/CPJ.0000000000000421

**9.** Orengo JP, Murdock DR. Genetic testing in neuromuscular disorders. *Pract Neurol*. 2019;July/August:35-41.

**10.** Ashizawa T, Xia G. Ataxia. *Continuum (Minneap Minn)*. 2016;22(4 Movement Disorders):1208-1226. doi:10.1212/CON.0000000000000362