



## Complete Summary

---

### GUIDELINE TITLE

Ataxia.

### BIBLIOGRAPHIC SOURCE(S)

Brunberg JA, Seidenwurm DJ, Davis PC, De La Paz RL, Dormont PD, Hackney DB, Jordan JE, Karis JP, Mukherji SK, Turski PA, Wippold FJ II, Zimmerman RD, McDermott MW, Sloan MA, Expert Panel on Neurologic Imaging. Ataxia. [online publication]. Reston (VA): American College of Radiology (ACR); 2006. 10 p. [80 references]

### GUIDELINE STATUS

This is the current release of the guideline.

This guideline updates a previous version: Johnson BA, Drayer BP, Anderson RE, Braffman B, Davis PC, Deck MD, Hasso AN, Masaryk T, Pomeranz SJ, Seidenwurm D, Tanenbaum L, Masdeu JC. Ataxia. American College of Radiology. ACR Appropriateness Criteria. Radiology 2000 Jun;215(Suppl):573-8.

The appropriateness criteria are reviewed annually and updated by the panels as needed, depending on introduction of new and highly significant scientific evidence.

## COMPLETE SUMMARY CONTENT

SCOPE  
METHODOLOGY - including Rating Scheme and Cost Analysis  
RECOMMENDATIONS  
EVIDENCE SUPPORTING THE RECOMMENDATIONS  
BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS  
QUALIFYING STATEMENTS  
IMPLEMENTATION OF THE GUIDELINE  
INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES  
IDENTIFYING INFORMATION AND AVAILABILITY  
DISCLAIMER

## SCOPE

### DISEASE/CONDITION(S)

Ataxia

**Note:** Vertigo is a separate American College of Radiology (ACR) Appropriateness Criteria® topic and will not be readdressed in this guideline.

## **GUIDELINE CATEGORY**

Diagnosis

## **CLINICAL SPECIALTY**

Family Practice  
Internal Medicine  
Medical Genetics  
Neurology  
Pediatrics  
Radiology

## **INTENDED USERS**

Health Plans  
Hospitals  
Managed Care Organizations  
Physicians  
Utilization Management

## **GUIDELINE OBJECTIVE(S)**

To categorize the diverse disorders that may present with ataxia and to suggest general imaging guidelines that may be useful for patients with the most common clinical presentations and underlying disorders

## **TARGET POPULATION**

Patients with ataxia

## **INTERVENTIONS AND PRACTICES CONSIDERED**

1. Magnetic resonance imaging (MRI)
  - Brain, without and with contrast
  - Cervical, thoracic and lumbar spine, without and with contrast
2. MR angiography (MRA), head and neck, without and with contrast
3. MR spectroscopy, (MRS), head
4. Computed tomography (CT)
  - Head, without and with contrast
  - Temporal bone
5. CT angiography (CTA), head and neck
6. Positron emission tomography with fluorodeoxyglucose (FDG-PET), brain

## **MAJOR OUTCOMES CONSIDERED**

Utility of radiologic examinations in differential diagnosis

## METHODOLOGY

### **METHODS USED TO COLLECT/SELECT EVIDENCE**

Searches of Electronic Databases

### **DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE**

The guideline developer performed literature searches of peer-reviewed medical journals and the major applicable articles were identified and collected.

### **NUMBER OF SOURCE DOCUMENTS**

The total number of source documents identified as the result of the literature search is not known.

### **METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE**

Weighting According to a Rating Scheme (Scheme Not Given)

### **RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE**

Not stated

### **METHODS USED TO ANALYZE THE EVIDENCE**

Systematic Review with Evidence Tables

### **DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE**

One or two topic leaders within a panel assume the responsibility of developing an evidence table for each clinical condition, based on analysis of the current literature. These tables serve as a basis for developing a narrative specific to each clinical condition.

### **METHODS USED TO FORMULATE THE RECOMMENDATIONS**

Expert Consensus (Delphi)

### **DESCRIPTION OF METHODS USED TO FORMULATE THE RECOMMENDATIONS**

Since data available from existing scientific studies are usually insufficient for meta-analysis, broad-based consensus techniques are needed to reach agreement in the formulation of the appropriateness criteria. The American College of Radiology (ACR) Appropriateness Criteria panels use a modified Delphi technique to arrive at consensus. Serial surveys are conducted by distributing questionnaires to consolidate expert opinions within each panel. These questionnaires are distributed to the participants along with the evidence table and narrative as

developed by the topic leader(s). Questionnaires are completed by the participants in their own professional setting without influence of the other members. Voting is conducted using a scoring system from 1-9, indicating the least to the most appropriate imaging examination or therapeutic procedure. The survey results are collected, tabulated in anonymous fashion, and redistributed after each round. A maximum of three rounds is conducted and opinions are unified to the highest degree possible. Eighty percent agreement is considered a consensus. This modified Delphi technique enables individual, unbiased expression, is economical, easy to understand, and relatively simple to conduct.

If consensus cannot be reached by the Delphi technique, the panel is convened and group consensus techniques are utilized. The strengths and weaknesses of each test or procedure are discussed and consensus reached whenever possible. If "No consensus" appears in the rating column, reasons for this decision are added to the comment sections.

**RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS**

Not applicable

**COST ANALYSIS**

A formal cost analysis was not performed and published cost analyses were not reviewed.

**METHOD OF GUIDELINE VALIDATION**

Internal Peer Review

**DESCRIPTION OF METHOD OF GUIDELINE VALIDATION**

Criteria developed by the Expert Panels are reviewed by the American College of Radiology (ACR) Committee on Appropriateness Criteria.

**RECOMMENDATIONS**

**MAJOR RECOMMENDATIONS**

**ACR Appropriateness Criteria®**

**Clinical Condition: : Ataxia**

**Variant 1: Slowly progressive ataxia, or ataxia of long duration (adult or child).**

Radiologic Procedure	Appropriateness Rating	Comments
MRI, brain, without	8	

<b>Radiologic Procedure</b>	<b>Appropriateness Rating</b>	<b>Comments</b>
and with contrast		
MRI, brain, without contrast	7	
MRI, cervical, thoracic and lumbar spine, without and with contrast	7	Ataxia can be of spinal origin. Consider if brain imaging is negative or inconclusive.
MRI, cervical thoracic, and lumbar spine, without contrast	6	Ataxia can be of spinal origin. Consider if brain imaging is negative or inconclusive
CT, head, without and with contrast	5	
FDG-PET, brain	3	
MR spectroscopy (MRS), head	2	
<b><i>Appropriateness Criteria Scale</i></b> <b>1 2 3 4 5 6 7 8 9</b> <b>1 = Least appropriate 9 = Most appropriate</b>		

Note: Abbreviations used in the tables are listed at the end of the "Major Recommendations" field.

**VARIANT 2: Acute ataxia (<3 hours) as a manifestation of suspected stroke (adult or child). (Also see the ACR Appropriateness Criteria® topic for cerebrovascular disease).**

<b>Radiologic Procedure</b>	<b>Appropriateness Rating</b>	<b>Comments</b>
MRI, brain, with or without contrast	8	MR preferred if treatment is not unreasonably delayed. Combined vascular and cerebral evaluation should be considered. Fat saturated T1 axial images.
MRA, head and neck, with or without contrast	8	MR preferred if treatment is not unreasonably delayed. Combined vascular and cerebral evaluation should be considered.
CTA, head and neck	8	MR preferred if treatment is not unreasonably delayed. Combined vascular and cerebral evaluation should

<b>Radiologic Procedure</b>	<b>Appropriateness Rating</b>	<b>Comments</b>
		be considered.
CT, head, with or without contrast	8	CT perfusion is less accurate in the posterior fossa. MR preferred if treatment is not unreasonably delayed. Combined vascular and cerebral evaluation should be considered.
MRI, cervical spine, without and with contrast	5	Fat saturated T1 axial images.
MRI, cervical spine, without contrast	4	
MR spectroscopy (MRS), head	2	
<b><i>Appropriateness Criteria Scale</i></b> <b>1 2 3 4 5 6 7 8 9</b> <b>1 = Least appropriate 9 = Most appropriate</b>		

Note: Abbreviations used in the tables are listed at the end of the "Major Recommendations" field.

**Variant 3: Acute or subacute ataxia as a manifestation of suspected infection (adult or child).**

<b>Radiologic Procedure</b>	<b>Appropriateness Rating</b>	<b>Comments</b>
MRI, brain, without and with contrast	8	
MRI, brain, without contrast	7	
MRI, cervical spine, without and with contrast	6	Ataxia can be of spinal origin. Consider if brain imaging is negative or inconclusive.
MR spectroscopy (MRS), head	6	May help distinguish abscess from other masses.
MRI, cervical spine, without contrast	5	Ataxia can be of spinal origin. Consider if brain imaging is negative or inconclusive.
MRA, head, without and with contrast	5	

<b>Radiologic Procedure</b>	<b>Appropriateness Rating</b>	<b>Comments</b>
CTA, head	5	
CT, head, without and with contrast	5	
CT, temporal bone	5	Useful when skull-based or middle ear disease suspected.
MRA, head, without contrast	4	
CT, head without contrast	4	
<b><i>Appropriateness Criteria Scale</i></b> <b>1 2 3 4 5 6 7 8 9</b> <b>1 = Least appropriate 9 = Most appropriate</b>		

Note: Abbreviations used in the tables are listed at the end of the "Major Recommendations" field.

**Variant 4: Acute ataxia following head trauma, less than 24 hours (adult or child).**

<b>Radiologic Procedure</b>	<b>Appropriateness Rating</b>	<b>Comments</b>
CT, head, without contrast	9	
MRI, head, without contrast	8	
CT, temporal bone	7	Useful when skull-based or middle ear disease suspected.
MRI, head, without and with contrast	7	
MRI, neck, without and with contrast	6	
CT, head, without and with contrast	6	
CTA, head and neck	6	
MRA, head and neck, with or without contrast	6	

Radiologic Procedure	Appropriateness Rating	Comments
<p><b><i>Appropriateness Criteria Scale</i></b>  <b>1 2 3 4 5 6 7 8 9</b>  <b>1 = Least appropriate 9 = Most appropriate</b></p>		

Note: Abbreviations used in the tables are listed at the end of the "Major Recommendations" field.

### **Summary of Literature Review**

Ataxia is a term that is used to describe abnormality in the coordination of movement. Manifestations commonly include a wide-based unsteady gait and poor coordination of the extremities. There can be associated abnormality in ocular motility and poor coordination of speech.

Ataxia can separately arise from disorders that involve the cerebellum, spinal cord, brainstem, vestibular nuclei, thalamic nuclei, white matter tracts of the brain stem and cerebral hemispheres, cerebral cortex (especially the frontal lobes), and peripheral sensory nerves. Because the anatomic regions responsible for ataxia are multiple, an effective imaging evaluation is often complex. Additionally, in patients with distinct clinical ataxia, appropriate magnetic resonance imaging (MRI) or computed tomography (CT) imaging of the cerebellum itself, or of the entire brain and spinal cord, may be entirely normal. This is most likely to occur early in the course of toxic, metabolic, or other progressive disorders associated with ataxia.

The medical disorders that cause ataxia are numerous and complex, and they are often individually quite uncommon. The development of prospective generalized recommendations for imaging, for the inclusion of anatomic regions to be studied, and for the use of specific protocols or pulse sequences is therefore imprecise. The purpose of this discussion, and of the guidelines presented below, is to categorize the diverse disorders that may present with ataxia and to suggest general imaging guidelines that may be useful for patients with the most common clinical presentations and underlying disorders. For individual patients, study design will be significantly compromised if the history that accompanies the patient is not precise or sufficient relative to the clinical and family history, to specific physical findings and/or to the results of relevant laboratory studies, many of which are quite complex.

Ataxia-associated disorders have been the subject of several classification schemes. For basic clinical and imaging purposes, however, these disorders can be approached on the basis of age at onset, potential disease mechanism, and anticipated clinical urgency for excluding a disorder that requires immediate diagnosis and management.

### **Classification of Disorders Causing Ataxia**

#### *Mass Lesions*

The exclusion of a posterior fossa mass lesion is often an important consideration in evaluating a patient with ataxia. The suspected mass can be primary or metastatic, and it can be intra-axial or extra-axial in location. In pediatric patients the most common intra-axial posterior fossa lesions are medulloblastoma, cystic astrocytoma, ependymoma, and brain stem glioma. In adults, intra-axial hemangioblastomas, choroid plexus papillomas, extra-axial meningiomas, and a host of intra-axial, extra-axial, or diffuse leptomeningeal metastatic processes become more prevalent. Isolated frontal lobe and thalamic mass lesions can also present with varying manifestations of gait and limb ataxia. Unless there is a contraindication, MRI, without and with contrast, is almost always superior to CT for the initial exclusion and characterization of a posterior fossa or other intracranial mass lesion.

Lhermitte-Duclos disease (dysplastic gangliocytoma) is a slowly growing benign cerebellar hamartoma or congenital malformation. Symptoms correlate with local mass effect. MRI demonstrates a cerebellar hemisphere mass that involves the cortex and folia and is generally of increased T2 signal intensity. There are also characteristic internal curvilinear bands that are isointense with cerebellar cortex. The mass does not enhance with contrast, and it may demonstrate restricted diffusion. Cowden syndrome, an autosomal dominant disorder characterized by cutaneous and noncutaneous hamartomas and by breast, thyroid, gastrointestinal and urologic gastric ulcer and genitourinary (GU) neoplasias, is frequently associated.

Paraneoplastic cerebellar degeneration is clinically characterized by the subacute or acute onset of gait and limb ataxia, dysarthria, and ocular dysmetria. It can occur in association with essentially any tumor type but most commonly occurs with breast, gynecologic, and lung tumors, and in association with Hodgkin's disease. Tumor seeding of brain parenchyma is not demonstrated with tissue sampling or with imaging. Several antineuronal antibodies have been identified in serum and in tissue, depending on the originating tumor cell type. MRI is generally normal until late stages of the simultaneously involved brain parenchymal areas. CT imaging and/or positron emission tomography (PET) imaging are indicated when an underlying primary is not evident.

### *Vascular Disorders*

Strategically located ischemic or hemorrhagic insults can result in distinct and often prominent ataxia. Infarction in the distribution of the posterior inferior cerebellar artery (lateral medullary syndrome or Wallenberg syndrome) is the most common pattern of brainstem infarction that is associated with a specific syndrome of ataxia. Symptoms include ipsilateral hemiataxia, vertigo, dysarthria, ptosis, and miosis. While brainstem and cerebellar infarctions are predominately arterial in origin, venous infarction should also be considered. The radiologic evaluation of ataxia generally requires MRI, with water diffusion characterization and with time-of-flight MR angiography. Use is also made of neck vessel MRI, using T1-weighted images, without and with fat saturation, to exclude the possibility of dissection. MR venography should be accomplished if there is consideration of central or dural venous thrombosis. Catheter-based diagnostic angiography and/or CT angiography (CTA) is occasionally necessary.

When the posterior fossa or supratentorial brain parenchyma is involved, vascular malformations, angiopathy, or aneurysm rupture can each lead to the acute development of ataxia. When there is consideration of acute or subacute hemorrhage, CT imaging of the brain and CTA may replace or supplement MRI.

MRI is the definitive diagnostic procedure for characterizing superficial siderosis. In this disorder hemosiderin accumulates in subpial layers of brain and spinal cord as the result of recurrent, often silent, subarachnoid hemorrhage. The most prominent symptoms are slowly progressive ataxia and hearing loss. With MRI there is low superficial T2 signal intensity over cortex, brain stem, and/or spinal cord with usual cerebellar atrophy.

### *Infectious and Postinfectious Processes*

A number of infectious and postinfectious processes can be responsible for the development of ataxia. To detect infectious-process-related alterations in the cerebellum, MRI without and with contrast administration, provides a distinct advantage over CT. This advantage is due to superior contrast resolution of MRI, and to the absence of CT-related artifacts that can occur in association with bone at the margins of the posterior fossa.

Bacterial cerebellitis can occur in association with meningitis or with cerebritis involving the cerebral hemispheres. It can also occur independently. Penetrating trauma or transdural extension of an epidural process, most commonly from the temporal bone, also needs to be considered. Diffusion imaging and MR spectroscopy can narrow the differential diagnosis. Multiple viral processes, including herpes and arbovirus, can also be associated with brainstem or cerebellar involvement.

Variant Creutzfeldt-Jakob disease (vCJD), also known as bovine spongiform encephalopathy (BSE), familial Creutzfeldt-Jakob disease (fCJD), and sporadic Creutzfeldt-Jakob disease (sCJD) are the most common prion-associated spongiform encephalopathies. Both vCJD and sCJD present with behavioral, emotional, and intellectual deterioration, followed by development of ataxia and dysarthria. Progression is to stupor and coma, with associated myoclonus being prominent in sCJD. The clinical course is more prolonged in vCJD. MRI demonstrates increased T2 signal intensity, and increased signal intensity on inversion recovery and diffusion-weighted sequences, in the heads of the caudate nuclei, the putamen, and regions of frontal, parietal, and occipital cortex. These alterations in signal intensity can initially be asymmetric. There is eventual diffuse volume loss. While all forms of CJD can have increased T2 signal intensity and restricted diffusion in the thalamic nuclei and in the pulvinar bilaterally, this focal alteration is especially prominent in vCJD.

Acute cerebellitis, also called acute cerebellar ataxia, is a para-infectious disorder that predominately, but not exclusively, occurs in childhood. Symptoms include headache, ataxia, photophobia, and varying findings associated with potential increased intracranial pressure or brainstem involvement. MRI demonstrates increased T2 signal intensity in the cerebellar hemispheres with associated mass effect. Bilateral abnormality is more common than unilateral abnormality. There may be obstruction of cerebrospinal fluid (CSF) flow with enlargement of the lateral ventricles and upward herniation of posterior fossa structures. There may

also be cerebellar meningeal enhancement following contrast administration. Surgical decompression of the posterior fossa may be necessary. Follow-up imaging may demonstrate cerebellar atrophy.

Bickerstaff encephalitis is a brainstem and cerebellar inflammatory disorder that most commonly follows a viral illness. It is characterized by ataxia and ophthalmoplegia, with MRI demonstrating mass effect, increased T2 signal intensity, and restricted diffusion within portions of the pons, medulla, and cerebellum.

Fisher syndrome, a variant of the Guillain-Barré syndrome, involves the peripheral and central nervous system. It is clinically characterized by ophthalmoplegia and cerebellar ataxia, and it is associated with transient high T2 signal intensity within the cerebellum and/or brainstem. Enhancement of cranial nerves and spinal nerve roots can be demonstrated, and there may be increased T2 signal intensity within the posterior portions of the spinal cord. Fluid attenuated inversion recovery (FLAIR) images often demonstrate this alteration to greatest advantage in the acute phase. Cerebellar atrophy is generally demonstrated during the convalescent phase.

### *Trauma*

Gait instability is a frequent component of concussion syndrome, and it may persist in association with a posttraumatic encephalopathy. Symptoms may be due to damage to the posterior fossa, vestibular, or brain stem structures. Persisting ataxia can also relate to frontal lobe injury. One possible explanation for the association of ataxia with a frontal lobe lesion of any origin is interruption of the frontopontocerebellar tract (Arnold's bundle). This tract originates in Brodmann's area 10 and carries information regarding intentional movement to the contralateral cerebellum via the middle cerebellar peduncle. Interruption of this tract along its course, or at its origin, deprives the cerebellum of frontal cortical input, resulting in impaired coordination and locomotion. In the presence of acute trauma, or in subjects with progressive post-traumatic ataxia, an expanding cyst or extra-axial hematoma should separately be considered.

### *Demyelinating Disorders*

Multiple sclerosis patients commonly present with or subsequently manifest persisting ataxia. In these patients, MRI (without and with contrast), diffusion imaging, spectroscopy, perfusion imaging, and magnetization transfer imaging can each support but not establish the diagnosis. The presence of multiple oval periventricular regions of increased T2 signal intensity, generalized cerebral volume loss, callosal and optic nerve involvement, and ring enhancement of active lesions is typical. MRI findings, and the utility of advanced MRI techniques for patient evaluation and management, have recently been reviewed.

### *Congenital Disorders*

A large number of ataxia-associated disorders occur on a congenital basis. In each of these processes MRI is preferred to CT. All of these disorders will most commonly manifest ataxia during early childhood development. Some of them are sporadic, while others have a known or an apparent genetic basis. Clinical

abnormalities that occur in association with congenital ataxia can include mild to severe mental retardation, hearing loss, optic atrophy, cataract, growth retardation, seizures, cleft palate, and either spasticity or diminished muscle tone. In these well-characterized congenital ataxia-associated disorders, imaging findings generally include nonspecific hypoplasia of the cerebellar vermis, hypoplasia of the entire cerebellum, or congenital cerebellar developmental dysplasia. Additional associated imaging alterations can include brain stem hypoplasia, lissencephaly, aprosencephaly, microcephaly, or variable and less prominent cerebral developmental alteration.

Though the disorders that can present as congenital ataxia are individually uncommon, a small number are more frequently recognized. Dandy Walker Syndrome, with ataxia, nystagmus, cranial nerve palsies, apneic episodes, hydrocephalus, and cognitive dysfunction, is characterized by hypoplasia of the cerebellar vermis with an associated CSF collection that is predominately posterior to the cerebellum but continuous with the fourth ventricle. The torcula is usually elevated and the posterior fossa usually enlarged. Hydrocephalus is frequently associated, and there may be anomalies of cerebral development that involve the cerebral cortex and corpus callosum. Differentiation from other congenital or acquired posterior fossa cysts is essential.

Joubert syndrome, with congenital ataxia, hypotonia, and oculomotor ataxia, has unique imaging alterations that involve the midbrain and cerebellum ("molar tooth" contour of brainstem or "bat wing" configuration of fourth ventricle). Four types have been identified, each with somewhat variable clinical and imaging features, and with genetic alterations that involve different loci.

Rhombencephalosynapsis is a rare cerebellar dysplastic process that can occur alone or in association with other developmental anomalies. In rhombencephalosynapsis there is vermian agenesis with fusion of the cerebellar hemispheres, apposition or fusion of the dentate nuclei, and fusion of the superior cerebellar peduncles. There is usually enlargement of the lateral ventricles, and there may be fusion of the thalamic nuclei. There is a wide spectrum of clinical symptomatology, and some patients are clinically normal.

Congenital ataxia can also occur in association with perinatal cerebral infarction and in association with congenital cytomegalovirus or other infectious processes of the central nervous system. Though uncommon relative to other forms of cerebral palsy, the imaging correlates of ataxic cerebral palsy have not been well defined.

#### *Hereditary and Idiopathic Degenerative Processes*

The hereditary ataxias are classified on the basis of their causative gene (when known) and their pattern of inheritance (autosomal dominant, autosomal recessive, x-linked, or mitochondrial). In each of these disorders MRI is the preferred imaging modality. Among this group of patients, a broad range of potential diagnostic considerations is often suggested by family history, by findings on physical examination, and, in symptomatic patients, by MRI evidence of atrophy involving the cerebellum and varying combinations of the pons, medulla, spinal cord, cerebral cortex and optic nerves. Dentate calcification may also be identified on CT imaging. Definitive diagnosis, however, relies on molecular genetic testing. While cerebellar ataxia is the dominant and occasionally

the only clinical finding, spasticity, neuropathy, seizures, extrapyramidal symptomatology, mental retardation, cognitive decline, nystagmus, visual loss, spasmodic cough, and migraine-like episodes may also be associated.

Among the identified autosomal dominant spinocerebellar ataxias (AD-SCAs), specific diagnostic nomenclature is replacing less specific terms such as "spinocerebellar degeneration," "Marie's ataxia," and olivopontocerebellar atrophy (OPCA). Among the AD-SCA disorders, 22 separate and distinct genetic abnormalities have now been identified. The term OPCA continues to be used only as a label for cases that have a clinical and pathology-related combination of "cerebellar-plus" symptomatology, have imaging correlates of cerebellar and brainstem atrophy, and have an (as yet) unidentified genetic basis. The designation of "idiopathic late onset cerebellar ataxia" is separately used to describe a different and significantly large group of adult patients with predominant cerebellar symptomatology, absence of a family history, and absence of an identified genetic marker. In these patients MRI generally demonstrates cerebellar and pontine volume loss.

SCA2 is one form of AD-SCA. It is caused by the presence of 32 or more CAG trinucleotide repeats on the ATXN2 gene. Symptoms include slowly progressive ataxia, dysarthria, nystagmus and initially brisk but later absent tendon reflexes, with associated peripheral neuropathy. Dystonia, Parkinsonism, and dementia may also be present in SCA2. Symptoms are more rapidly progressive when they have their onset before 20 years of age. Imaging findings include cerebellar and pontine volume loss and deep white matter alterations that may also involve the cerebral hemispheres. Although SCA2 is a relatively common form of AD-SCA (13% of AD-SCA cases in one study), clinical and imaging features do not allow a definitive diagnosis. Molecular genetic analysis is necessary. Many, but not all cases as Marie's ataxia and AD-OPCA are thought to represent SCA2.

SCA3 (Machado-Joseph disease or MJD) accounts for 23% of patients with AD-SCA. Symptoms most commonly include an onset in the second to fourth decade of cerebella ataxia, spasticity, peripheral neuropathy, bulbar dysfunction with facial and tongue atrophy, and occasional myoclonus or intellectual impairment. Subtypes have been described. MRI alterations include volume loss involving the cerebellum, pons and medulla, and a linear region of high T2 signal intensity along the posterior and medial margins of the globus pallidus.

Dentatorubral-pallidoluysian (DRPLA) is characterized by progressive ataxia, choreoathetosis, and dementia or character changes when the disorder occurs in adults. In children it is characterized by ataxia, myoclonus, epilepsy, and progressive intellectual deterioration. MRI demonstrates cerebellar and brainstem volume loss with cerebral cortical atrophy. Less frequently there is increased T2 signal intensity in deep white matter of the cerebral hemispheres, in the thalamus, and in the brainstem. Diagnosis is established through the identification of >48 CAG repeats in the DRPLA gene.

Autosomal recessive hereditary ataxias can be associated with multiple underlying genetic disorders. Refer to the original guideline document for a description of ataxia-telangiectasia, fragile X tremor/ataxia syndrome, multiple system atrophy, and mitochondrial disorders.

### *Paroxysmal Disorders*

Paroxysmal ataxia has been associated with several disorders, including epilepsy and migraine, as well as with a transient limb and trunk ataxia that can occur with high systemic fever in otherwise healthy children. These disorders can be idiopathic or can be associated with abnormalities in membrane calcium or potassium channel function, or with altered synaptic glutamate transport. MRI may be normal, may demonstrate cerebellar volume loss, or may demonstrate extensive areas of cortical increased T2 signal intensity that may correlate with the possible simultaneous occurrence of hemiplegic migraine or recent seizure activity. MRI is the imaging modality of choice.

### *Spinal Cord and Peripheral Nerve-Related Ataxia*

Ataxia that is potentially due to pathologic processes that originate within the spinal cord, or within the roots/nerves that originate from the spinal cord, requires high resolution T1 and T2-weighted axial and sagittal imaging without and with contrast that focuses on the posterior columns and on the nerve roots. Findings in pernicious anemia depend on the duration and severity of the disorder.

There may be early localized or relatively diffuse cord swelling with increased T2 signal intensity that is usually most evident in the posterior columns. Late atrophy and persistent gliosis may develop, or all findings may resolve with treatment. In the presence of hypertrophic, inflammatory, or postinfectious polyneuropathies, nerve root enhancement and enlargement may be demonstrated with MRI.

### *Nutritional Deficiency, Toxins and Drugs*

In each of these disorders MRI is the preferred imaging modality. Solvent abuse or toxic exposure to solvents can result in gait impairment and in encephalopathy. MRI abnormalities are characterized by diffuse cortical atrophic changes and by hyperintensity on T2-weighted images in the white matter, basal ganglia, and thalami.

Methyl-mercury poisoning (Minamata disease) is a neurological illness caused by the ingestion of contaminated seafood and characterized by ataxia, visual loss, and sensory disturbance. MRI in affected patients demonstrates atrophy of the cerebellar vermis and hemispheres, as well as the calcarine cortex.

Metronidazole (Flagyl)-induced cerebellar toxicity is associated with symptoms of ataxia, with MRI findings of increased T2 signal intensity and restricted diffusion in the dentate nuclei. With symptom resolution MRI becomes normal.

In central pontine myelinolysis (osmotic demyelination syndrome), cerebellar or extrapyramidal symptoms have been observed. More typically, patients demonstrate coma, locked-in syndrome, or quadriparesis. This disorder is typically seen in chronic alcoholics or malnourished patients following the rapid correction of hyponatremia. Increased T2 signal intensity in the pons is the characteristic finding.

A leukoencephalopathy with initial symptoms of ataxia has also been reported to occur in association with the chronic inhalation of heroin vapors.

Vitamin E deficiency can occur in association with several acquired gastrointestinal disorders or with autosomal recessive defects in vitamin E transport. Symptoms include ataxia with associated weakness, areflexia, and retinal degeneration. Imaging has demonstrated cerebellar atrophy and increased T2 signal intensity in the posterior columns of the spinal cord.

Chronic ethanol abuse is associated with ataxia and multiple other symptoms of neurologic dysfunction. These symptoms result from the neurotoxicity of ethanol and its metabolic products, from associated chronic liver disease, from secondary nutritional deficiencies, and from the effect of other toxins that are simultaneously ingested. MRI demonstrates atrophy of the cerebellar vermis, especially superiorly, as well as volume loss involving pons, medulla, and cerebral hemispheres.

Wernicke encephalopathy is due to thiamine deficiency and classically presents with ataxia, altered mental status, and abnormality of ocular motility. MRI demonstrates increased T2 signal intensity, reversible contrast enhancement, and reversible restricted diffusion in multiple areas including mammillary bodies, hypothalamus adjacent to the third ventricle, periaqueductal gray and white matter, pulvinar, and dorsomedial portions of the thalamic nuclei. Small hemorrhagic foci can also be demonstrated in these regions.

Reversible posterior leukoencephalopathy is most commonly characterized by headache, altered consciousness, visual disturbance, and seizures. Ataxia can be a component, especially when there is brainstem or cerebellar involvement. Antecedent clinical conditions include hypertension, eclampsia, renal disease, and the use of cytotoxic or immunosuppressant drugs. MRI findings include the presence of bilateral and generally symmetrically increased T2 signal intensity in the posterior parietal and occipital lobes, with apparent early restricted and subsequent increased diffusion in these same areas.

### **Abbreviations**

- CT, computed tomography
- CTA, computed tomographic angiography
- FDG, fluorodeoxyglucose
- MRA, magnetic resonance angiography
- MRI, magnetic resonance imaging
- PET, positron emission tomography

### **CLINICAL ALGORITHM(S)**

Algorithms were not developed from criteria guidelines.

## **EVIDENCE SUPPORTING THE RECOMMENDATIONS**

### **TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS**

The recommendations are based on analysis of the current literature and expert panel consensus.

## **BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS**

### **POTENTIAL BENEFITS**

Selection of appropriate radiologic imaging procedures for evaluation of patients with ataxia

### **POTENTIAL HARMS**

Not stated

## **QUALIFYING STATEMENTS**

### **QUALIFYING STATEMENTS**

An American College of Radiology (ACR) Committee on Appropriateness Criteria and its expert panels have developed criteria for determining appropriate imaging examinations for diagnosis and treatment of specified medical condition(s). These criteria are intended to guide radiologists, radiation oncologists and referring physicians in making decisions regarding radiologic imaging and treatment. Generally, the complexity and severity of a patient's clinical condition should dictate the selection of appropriate imaging procedures or treatments. Only those exams generally used for evaluation of the patient's condition are ranked. Other imaging studies necessary to evaluate other co-existent diseases or other medical consequences of this condition are not considered in this document. The availability of equipment or personnel may influence the selection of appropriate imaging procedures or treatments. Imaging techniques classified as investigational by the U.S. Food and Drug Administration (FDA) have not been considered in developing these criteria; however, study of new equipment and applications should be encouraged. The ultimate decision regarding the appropriateness of any specific radiologic examination or treatment must be made by the referring physician and radiologist in light of all the circumstances presented in an individual examination.

## **IMPLEMENTATION OF THE GUIDELINE**

### **DESCRIPTION OF IMPLEMENTATION STRATEGY**

An implementation strategy was not provided.

### **IMPLEMENTATION TOOLS**

Personal Digital Assistant (PDA) Downloads

For information about [availability](#), see the "Availability of Companion Documents" and "Patient Resources" fields below.

## INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

### IOM CARE NEED

Living with Illness

### IOM DOMAIN

Effectiveness

## IDENTIFYING INFORMATION AND AVAILABILITY

### BIBLIOGRAPHIC SOURCE(S)

Brunberg JA, Seidenwurm DJ, Davis PC, De La Paz RL, Dormont PD, Hackney DB, Jordan JE, Karis JP, Mukherji SK, Turski PA, Wippold FJ II, Zimmerman RD, McDermott MW, Sloan MA, Expert Panel on Neurologic Imaging. Ataxia. [online publication]. Reston (VA): American College of Radiology (ACR); 2006. 10 p. [80 references]

### ADAPTATION

Not applicable: The guideline was not adapted from another source.

### DATE RELEASED

1999 (revised 2006)

### GUIDELINE DEVELOPER(S)

American College of Radiology - Medical Specialty Society

### SOURCE(S) OF FUNDING

The American College of Radiology (ACR) provided the funding and the resources for these ACR Appropriateness Criteria®.

### GUIDELINE COMMITTEE

Committee on Appropriateness Criteria, Expert Panel on Neurologic Imaging

### COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

*Panel Members:* James A. Brunberg, MD; David J. Seidenwurm, MD; Patricia C. Davis, MD; Robert L. DeLaPaz, MD; Pr. Didier Dormont; David B. Hackney, MD; John E. Jordan, MD; John P. Karis, MD; Suresh Kumar Mukherji, MD; Patrick A. Turski, MD; Franz J. Wippold II, MD; Robert D. Zimmerman, MD; Michael W. McDermott; Michael A. Sloan, MD, MS

## **FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST**

Not stated

## **GUIDELINE STATUS**

This is the current release of the guideline.

This guideline updates a previous version: Johnson BA, Drayer BP, Anderson RE, Braffman B, Davis PC, Deck MD, Hasso AN, Masaryk T, Pomeranz SJ, Seidenwurm D, Tanenbaum L, Masdeu JC. Ataxia. American College of Radiology. ACR Appropriateness Criteria. Radiology 2000 Jun;215(Suppl):573-8.

The appropriateness criteria are reviewed annually and updated by the panels as needed, depending on introduction of new and highly significant scientific evidence.

## **GUIDELINE AVAILABILITY**

Electronic copies: Available in Portable Document Format (PDF) from the [American College of Radiology \(ACR\) Web site](#).

ACR Appropriateness Criteria® *Anytime, Anywhere*™ (PDA application). Available from the [ACR Web site](#).

Print copies: Available from the American College of Radiology, 1891 Preston White Drive, Reston, VA 20191. Telephone: (703) 648-8900.

## **AVAILABILITY OF COMPANION DOCUMENTS**

The following is available:

- ACR Appropriateness Criteria®. Background and development. Reston (VA): American College of Radiology; 2 p. Electronic copies: Available in Portable Document Format (PDF) from the [American College of Radiology \(ACR\) Web site](#).

## **PATIENT RESOURCES**

None available

## **NGC STATUS**

This summary was completed by ECRI on July 31, 2001. The information was verified by the guideline developer as of August 24, 2001. This NGC summary was updated by ECRI Institute on April 27, 2007.

## **COPYRIGHT STATEMENT**

Instructions for downloading, use, and reproduction of the American College of Radiology (ACR) Appropriateness Criteria® may be found on the [ACR Web site](#).

## DISCLAIMER

### NGC DISCLAIMER

The National Guideline Clearinghouse™ (NGC) does not develop, produce, approve, or endorse the guidelines represented on this site.

All guidelines summarized by NGC and hosted on our site are produced under the auspices of medical specialty societies, relevant professional associations, public or private organizations, other government agencies, health care organizations or plans, and similar entities.

Guidelines represented on the NGC Web site are submitted by guideline developers, and are screened solely to determine that they meet the NGC Inclusion Criteria which may be found at <http://www.guideline.gov/about/inclusion.aspx>.

NGC, AHRQ, and its contractor ECRI Institute make no warranties concerning the content or clinical efficacy or effectiveness of the clinical practice guidelines and related materials represented on this site. Moreover, the views and opinions of developers or authors of guidelines represented on this site do not necessarily state or reflect those of NGC, AHRQ, or its contractor ECRI Institute, and inclusion or hosting of guidelines in NGC may not be used for advertising or commercial endorsement purposes.

Readers with questions regarding guideline content are directed to contact the guideline developer.

© 1998-2008 National Guideline Clearinghouse

Date Modified: 9/22/2008

