Autoimmune Movement Disorder Panel, Serum and CSF

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Autoimmune movement disorders encompass a large, diverse group of neurologic disorders that can occur in isolation or in conjunction with other autoimmune encephalitides. Detection of antineural antibodies may help to establish a diagnosis, support treatment decisions, aid with prognostication, and guide the search for an associated malignancy.

Disease Overview

Autoimmune movement disorders may resemble genetic, metabolic, or neurodegenerative movement disorders. Importantly, autoimmune movement disorders may be treatable if identified in a timely manner; they may be the presenting symptom of an undiagnosed malignancy, and recognition of tumor-antibody associations may allow for cancer treatment at an early stage.[1] Signs and symptoms associated with these disorders are diverse and may include tremor, ataxia, chorea, neuromyotonia, myokymia, dystonia, myoclonus, abnormal eye movements, and/or parkinsonism.[1] Patients may also have symptoms such as headache, psychosis, hallucinations, and/or agitation. Subacute onset of symptoms, inflammatory cerebrospinal fluid (CSF) studies, and magnetic resonance imaging (MRI) findings consistent with inflammation or cerebellar degeneration may support this diagnosis.[2]

For more information about laboratory testing for autoimmune neurologic diseases, refer to the ARUP Consult Autoimmune Neurologic Diseases - Antineural Antibody Testing topic.

Featured ARUP Testing

Autoimmune Movement Disorder Panel, Serum 3006206

Method: Semi-Quantitative Cell-Based Indirect Fluorescent Antibody/Semi-Quantitative Indirect Fluorescent Antibody (IFA)/Qualitative Immunoblot/Semi-Quantitative Enzyme-Linked Immunosorbent Assay (ELISA)/Quantitative Radioimmunoassay (RIA)

Autoimmune Movement Disorder Panel, CSF 3006207

Method: Semi-Quantitative Cell-Based Indirect Fluorescent Antibody/Semi-Quantitative Indirect Fluorescent Antibody (IFA)/Qualitative Immunoblot/Semi-Quantitative Enzyme-Linked Immunosorbent Assay (ELISA)

Test Description

These serum and CSF antineural antibody panel tests may be used for the evaluation of patients with subacute onset of movement disorders. Testing for the presence of antineural antibodies in both serum and CSF may improve diagnostic yield.[3]

These phenotype-targeted panels test for the presence of antibodies associated with movement disorders. Clinical phenotypes for specific antineural antibody-associated syndromes often overlap, and phenotype-specific panels allow for rapid identification of associated antibodies, which may have implications for treatment, prognosis, and cancer screening.[3] Other panels may be more appropriate, depending on the patient's clinical phenotype:

ARUP Phenotype-Specific Panels to Consider for Autoimmune Neurologic Disease

ARUP Panel	Test Code	
	Serum	CSF
Autoimmune Encephalopathy/Dementia Panel	3006201	3006202
Autoimmune Epilepsy Panel	3006204	3006205
Autoimmune Pediatric CNS Disorders Panel	3006210	3006211

Regardless of the panel chosen, order only one panel for serum and/or one panel for CSF; many antineural antibodies are redundant between these panels, and choosing based on the predominant phenotype will provide the most meaningful results. To compare these panels and the antibodies included, refer to the ARUP Antineural Antibody Testing for Autoimmune Neurologic Disease page.

Testing for individual autoantibodies is also available separately.

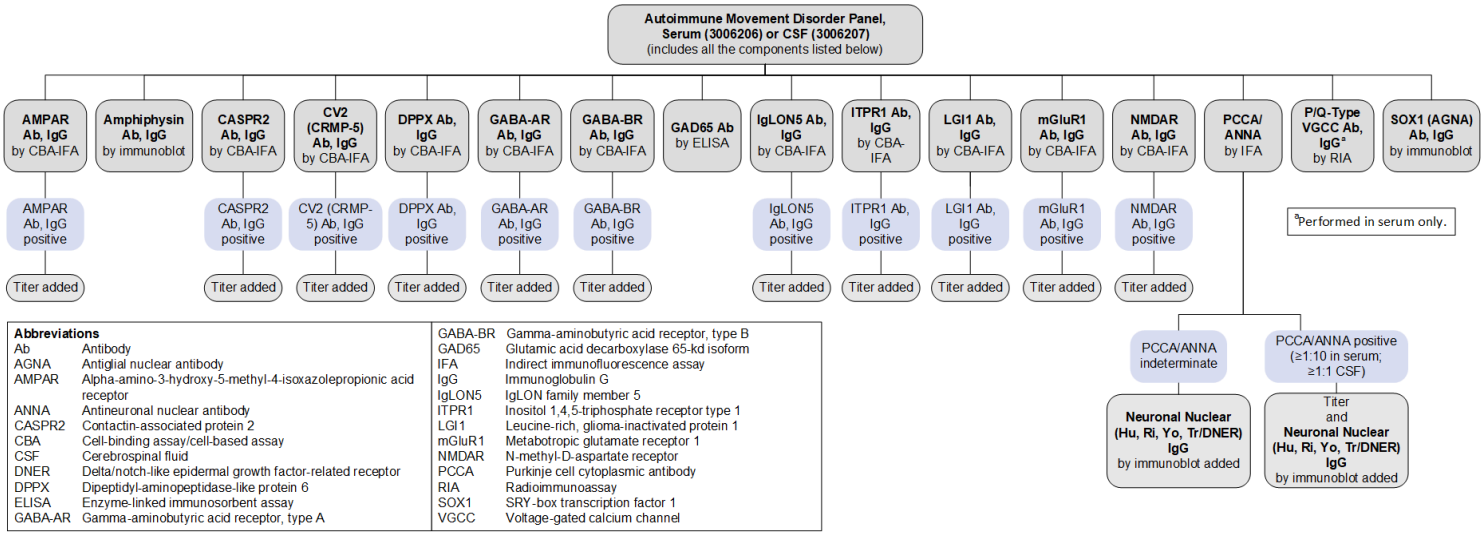
Antibodies Tested and Methodology

Autoimmune Movement Disorder Panel, Serum (3006206) and CSF (3006207): Antibodies Tested and Methodology			
Autoantibody Markers	Methodology	Individual Autoantibody Test Code	
		Serum	CSF
AMPAR Ab, IgG	CBA-IFA, reflex titer	3001260	3001257
Amphiphysin Ab, IgG	IB	2008893	3004510
ANNA-1 (Hu)	IFA, reflex IB, reflex titer	2007961	2010841
ANNA-2 (Ri)	IFA, reflex IB, reflex titer	2007961	2010841
CASPR2 Ab, IgG	CBA-IFA, reflex titer	2009452	3001986
CV2 (CRMP-5) Ab, IgG	CBA-IFA, reflex titer	3016999	3017001
DPPX Ab, IgG	CBA-IFA, reflex titer	3004359	3004512
GABA-AR Ab, IgG	CBA-IFA, reflex titer	3006008	3006003
GABA-BR Ab, IgG	CBA-IFA, reflex titer	3001270	3001267
GAD65 Ab	ELISA	2001771	3002788
IgLON5 Ab, IgG	CBA-IFA, reflex titer	3006018	3006013
ITPR1 Ab, IgG	CBA-IFA, reflex titer	3006031	3006023
LGI1 Ab, IgG	CBA-IFA, reflex titer	2009456	3001992
mGluR1 Ab, IgG	CBA-IFA, reflex titer	3006044	3006039
NMDAR Ab, IgG	CBA-IFA	2004221	2005164
PCCA-1 (Yo)	IFA, reflex IB, reflex titer	2007961	2010841
PCCA-Tr/DNER	IFA, reflex IB, reflex titer	2007961	2010841
P/Q-type VGCC Ab, IgG	RIA	0092628	—
SOX1 (AGNA) Ab, IgG	IB	3002885	3002886

Ab, antibody; AGNA, antiglial nuclear antibody; AMPAR, alpha-amino-3-hydroxy-5-methyl-4-isoxazolepropionic acid receptor; ANNA-1, antineuronal nuclear antibody type 1; ANNA-2, antineuronal nuclear antibody type 2; CASPR2, contactin-associated protein 2; CBA, cell-binding assay/cell-based assay; CRMP-5, collapsin response-mediator protein 5; DNER, Delta/notch-like epidermal growth factor-related receptor; DPPX, dipeptidyl-aminopeptidase-like protein 6; ELISA, enzyme-linked immunosorbent assay; GABA-AR, gamma-aminobutyric acid receptor, type A; GABA-BR, gamma-aminobutyric acid receptor, type B; GAD65, glutamic acid decarboxylase 65-kd isoform; IB, immunoblot; IFA, indirect immunofluorescence assay; IgLON5, IgLON family member 5; ITPR1, inositol 1,4,5-trisphosphate receptor type 1; LGI1, leucine-rich, glioma-inactivated protein 1; mGluR1, metabotropic glutamate receptor 1; NMDAR, N-methyl-D-aspartate receptor; PCCA-1, Purkinje cell cytoplasmic antibody type 1; PCCA-Tr, Purkinje cell cytoplasmic antibody type Tr; RIA, radioimmunoassay; SOX1, SRY-box transcription factor 1; VGCC, voltage-gated calcium channel

Reflex Patterns

Autoimmune Movement Disorder Panel, Serum (3006206) and CSF (3006207): Reflex Patterns



Limitations

These panels do not include every antibody that has been associated with autoimmune movement disorders:

- ANNA-3 and PCCA-2 are not included because they are extremely rare (present in approximately 0.0001% of specimens submitted for evaluation using a paraneoplastic antibody panel), and commercial assays to confirm the specificity of these antibodies are not currently available.[4]
- Adaptor protein 3 subunit B2 (AP3B2), glial fibrillary acidic protein (GFAP), GTPase regulator associated with focal adhesion kinase 1 (GRAF1), neuronal intermediate filament (NIF) and its associated reflexes (NIF heavy and light chain, alpha internexin), neurochondrin, septin 5, and septin 7 antibodies are not included because they have been only recently identified and their prevalence is currently not well established.
  - GFAP has been reported in 0.17% of samples screened, often co-occurring with other antineural antibodies.[5]
  - GRAF1 has only been described in rare case reports; its prevalence remains unknown.[6]
  - NIF has been reported in 0.014% of samples screened; NIF heavy and light chain and alpha internexin were reflexed in samples that were positive for NIF to further identify the associated antibody.[7]
  - Neurochondrin has been reported in 0.002% of samples tested.[8]
  - Septin 5 has been reported in <0.001% of samples screened.[9]
  - Septin 7 has been reported in 0.002% of samples screened.[9]
- Kelch-like protein 11 (KLHL11) antibody testing is available separately.[10]
- As testing for newly described antibodies becomes available and their clinical relevance is established, these panels will evolve to reflect these discoveries.

Test Interpretation

Results

Results must be interpreted in the clinical context of the individual patient; test results (positive or negative) should not supersede clinical judgment.

Autoimmune Movement Disorder Panel, Serum (3006206) and CSF (3006207): Results Interpretation	
Result	**Interpretation**
Positive for ≥1 autoantibodies	Autoantibody(ies) detected Supports a clinical diagnosis of an autoimmune movement disorder Consider a focused search for malignancy based on antibody-tumor associations
Negative	No autoantibodies detected A diagnosis of an autoimmune movement disorder is not excluded

References

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