

Autoimmune Pediatric CNS Disorders Panel, Serum and CSF

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Pediatric patients are susceptible to many of the same autoimmune syndromes that affect the central nervous system (CNS) of adults. However, the incidence and prevalence of specific antineural antibodies differ between pediatric and adult patients. ¹ In addition, diagnosis in children can be complicated by clinical overlap with other diseases (including genetic, infectious, metabolic, and psychiatric conditions), challenges in obtaining symptom history from very young patients, and the complexities of normal behavior changes during adolescence. ² Evaluation for the presence of antineural antibodies facilitates treatment, prognosis, and appropriate cancer screening. ³

Featured ARUP Testing

Autoimmune Pediatric CNS Disorders, Serum 3006210

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

Autoimmune Pediatric CNS Disorders, CSF 3006211

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

Disease Overview

Pediatric patients may develop autoimmune syndromes affecting the CNS that are characterized by diverse phenotypes including behavior changes, disseminated encephalomyelitis, encephalopathy, and epilepsy. Early recognition of possible autoimmune causes of neurologic changes in pediatric patients allows for appropriate diagnostic testing, rapid initiation of treatment, and improved outcomes.⁴

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

Test Description

These serum and CSF antineural antibody panel tests may be used for the evaluation of pediatric (<18 years of age) patients with subacute onset of encephalopathy, epilepsy, behavior changes, or movement disorders. Testing for the presence of antineural antibodies in both serum and CSF may improve diagnostic yield.⁵

These phenotype-targeted panels test for the presence of antibodies associated with pediatric autoimmune CNS syndromes. 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.⁵ For adult patients, other phenotype-specific panels are more appropriate:

ARUP Phenotype-Specific Panels to Consider for Adults With Autoimmune Neurologic Disease			
ARUP Panel	Test Code		
	Serum	CSF	
Autoimmune Encephalopathy/Dementia Panel	3006201	3006202	
Autoimmune Epilepsy Panel	3006204	3006205	
Autoimmune Movement Disorder Panel	3018964	3018966	
Autoimmune Myelopathy Panel	3006208	3006209	

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 antibodies is also available separately.

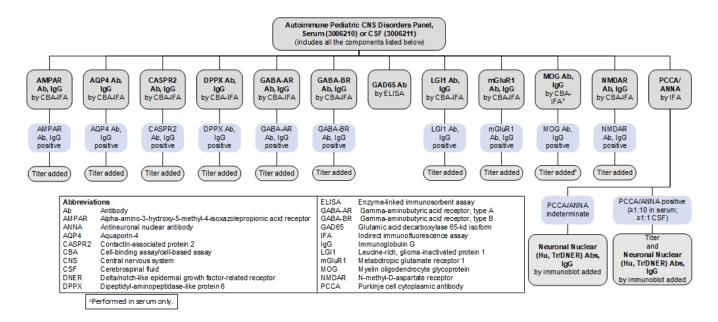
Antibodies Tested and Methodology

Autoimmune Pediatric CNS Disorders Panel, Serum (3006210) and CSF (3006211): Antibodies Tested and Methodology				
Autoantibody Markers	Methodology	Individu	Individual Autoantibody Test Code	
		Serum	CSF	
AMPAR Ab, IgG	CBA-IFA	3001260	3001257	
ANNA-1 (Hu)	IFA, reflex IB, reflex titer	2007961	2010841	
AQP4 Ab, IgG	CBA-IFA, reflex titer	2013320	2011699	
CASPR2 Ab, IgG	CBA-IFA, reflex titer	2009452	3001986	
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	
LGI1 Ab, IgG	CBA-IFA, reflex titer	2009456	3001992	
mGluR1 Ab, IgG	CBA-IFA, reflex titer	3006044	3006039	
MOG Ab, IgG	CBA-IFA, reflex titer	3001277	_	
NMDAR Ab, IgG	CBA-IFA, reflex titer	2004221	2005164	
PCCA-Tr/DNER	IFA, reflex IB, reflex titer	2007961	2010841	

Ab, antibody; AMPAR, alpha-amino-3-hydroxy-5-methyl-4-isoxazolepropionic acid receptor; ANNA-1, antinuclear neuronal antibody type 1; AQP4, aquaporin 4; CASPR2, contactin-associated protein 2; CBA, cell-binding assay/cell-based assay; 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 B; GAD65, glutamic acid decarboxylase 65-kd isoform; IB, immunoblot; IFA, indirect immunofluorescence assay; LGI1, leucinerich, glioma-inactivated protein 1; mGluR1, metabotropic glutamate receptor 1; MOG, myelin oligodendrocyte glycoprotein; NMDAR, N-methyl-D-aspartate receptor; PCCA-Tr, Purkinje cell cytoplasmic antibody type Tr

Reflex Patterns

Autoimmune Pediatric CNS Disorders Panel, Serum (3006210) and CSF (3006211): Reflex Patterns



Limitations

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

- Glial fibrillary acidic protein (GFAP) and neurochondrin are not included in this panel 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.⁶
 - Neurochondrin has been reported in 0.002% of samples tested.⁷
- 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 Pediatric CNS Disorders Panel, Serum (<u>3006210</u>) and CSF (<u>3006211</u>): Results Interpretation			
Result	Interpretation		
Positive for ≥1 autoantibodies	Autoantibody(ies) detected Supports a clinical diagnosis of an autoimmune CNS disorder Consider a focused search for malignancy based on antibody-tumor associations		
Negative	No autoantibodies detected A diagnosis of an autoimmune CNS disorder is not excluded		

References

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- 4. Wright MA, Trandafir CC, Nelson GR, et al. Diagnosis and management of suspected pediatric autoimmune encephalitis: a comprehensive, multidisciplinary approach and review of literature. *J Child Neurol*. 2021;37(4):303-313.
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