신경근육질환 자가항체 검사

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Serologic tests for neuromuscular disorders

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질병특이적 자가항체가 발견된 자가면역 신경근육질환들에는 중증근무력증(myasthenic gravis, MG)과 Lambert Eaton myasthenic syndrome (LEMS) 등 신경근접합부 질환(Gilhus & Verschuuren 2015), 만성염증성탈수초다발신경병증(Latov 2014)과 길랑바레증후군(Yuki & Hartung 2012), 자율신경병증(Koike et al. 2013) 등 말초신경질환, 다발성근염과 피부근염 등 염증성근육병증(Simon et al. 2016) 등이 있다 (Table). 각 질환의 역학, 병인과 기전, 진단 및 치료에 대한 자세한 내용은 최근에 발표된 종설들을 참고하기 바라며 (참고문헌), 본 강의는 자가면역 신경근육질환의 진단 및 치료 과정을 중심으로 해당 자가항체들의 임상적 의의를 살펴보고자 한다.

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Table. Auto-antibodies in neuromuscular disorders.

Auto-antibodies	Disease	Note
Neuromuscular junction disorders		
Anti-AchR	MG	Sensitivity -50% in ocular MG, 70-80% in generalized MG
Anti-MuSK	MG	Variable sensitivity (0-70%) in anti-AchR-Ab negative generalized MG, but rarely detected in ocular MG, IgG subclass (IgG4), often young female, bulbar dominant phenotype, facial muscles/tongue atrophy, intolerance to ACEI
Anti-LRP4	MG	7-33% of double seronegative MG (Anti-AchR and anti-MuSK), clinical characteristics similar to anti-AchR rather than anti-MUSK MG, IgG1 (and G2) subclass
Anti-agrin	MG	Pathgenecity not established
Anti-titin/RyR/Kv1.?	MG	Common in thymomatous MG and late-onset MG
Anti-VGCC (P/Q type)	LEMS	Associated with malignancy in -50% of patients (most commonly associated with small cell lung cancer)
Anti-VGKC	Neuromyotonia, Morvan's syndrome	Mostly autoimmune, but some cases with tumor (thymus and lung), peripheral nerve hyperexcitability, limbic encephalitis
Peripheral neuropathies		
Anti-gangliosides	GBS	GM1/GD1a in AMAN/AMSAN, GQ1b/GT1a in MFS/Bickerstaff's brainstem encephalitis/Pharyngeal-cervical-brachial variant of GBS
Anti-GM1	Multifocal motor neuropathy	IgM
Anti-MAG	Anti-MAG peripheral neuropathy	-50% in polyneuropathy with IgM monoclonal gammopathy, paresthesia and sensory ataxia followed by a varying degree of sensorimotor deficits
Anti-contactin 1	CIDP	-3% In CIDP, IgG4, subacute sensory ataxia, poor response to IVIG
Anti-NF155	CIDP	3-7% in CIDP, IgG4, younger age of onset, associated with ataxia, CNS demyelination, disabling tremor, poor response to IVIG
Anti-AchR (ganglionic)	Autonomic neuropathy	idiopathic autoimmune autonomic ganglionopathy
Myopathies		
Anti-tRNA synthetase	Dermatomyositis or overlap myositis	Anti-tRNA synthetase syndrome (myositis, interstitial lung disease, arthritis, Raynaud phenomenon, mechanic's hands), anti-Jo1 (histidyl-tRNA synthetase)
Anti-HMGCR, anti-SRP	Autoimmune necrotizing myopathy	-60% in autoimmune necrotizing myopathy, statin exposure in -50% of anti-HMGCR positive patients