Autoantikörper bei Erkrankungen der neuromuskulären Transmission

home

- Diagnostischer Krankheitsmarker von pathogener Bedeutung
- Diagnostischer Krankheitsmarker der Grunderkrankung
- hinweisender Autoantikörper
- Autoantikörper auch mit anderen Krankheiten assoziiert
- Bei dem betreffenden Krankheitsbild ebenfalls anzutreffende, aber nicht krankheitsspezifische Autoantikörper. Keine diagnostische Bedeutung für die beschriebene Erkrankung.

- **Literaturzitate** sind mit roten Zahlen markiert und auf die Autoren am Ende des Dokuments verlinkt. Durch Anklicken des Handsymbols (☞) gelangt man an den Tabellenanfang zurück.
- Die in der Tabelle aufgeführten Autoantikörper sind mit ihrer Beschreibung verlinkt.

Autoantikörper bei Erkrankungen der neuromuskulären Transmission

Myasthenia gravis

Autoantikörper gegen	Sens [%]	Spez [%]	Krankheitsassoziation
Acetylcholinrezeptor (AChRm)	50 - 90	hoch	
MuSK *	< 10	hoch	15
Lrp4	< 10	hoch	
Agrin	< 10	hoch	26, 27
Kaliumkanal Kv1.4	12 - 15	hoch	
Kollagen Q	3	hoch	28
Ryanodinrezeptor (RyR)	15 - 20	hoch	Thymom
Titin	30	hoch	Thymom
Dihydropyridinrezeptor ($Ca_v1.1$)	37	hoch	Thymom
TRPC3 Kanäle	36	hoch	Thymom
Gravin (AKAP-12)	31	hoch	4
Rapsyn	nieder	mittel	1
Skelettmuskel (IIFT)	80	mittel	
Aquaporin-4	nieder	nieder	NMO 3, 7, 8, 13, 19
CASPR2	nieder	nieder	NM, MS 19, 20
Actin	< 5	nieder	AH 14, 17, 21, 22
α-Actinin	< 25	nieder	AH 14, 17, 21, 22
Filamin	< 10	nieder	9, 23
Heat shock Protein 70	< 20	nieder	6, 12
Heat shock Protein HSC71	< 20	nieder	12
Interferon-α2	< 20	nieder	2, 5, 10, 11, 16
Interferon-α8	< 20	nieder	2, 5, 10, 11, 16
Interferon-ω	< 20	nieder	2, 5, 10, 11
Interleukin-12	< 20	nieder	5, 10, 11, 16, 18, 24, 25
Myosin	< 50	nieder	14, 17, 21, 22
Tropomyosin	< 30	nieder	17, 23
Troponin	< 30	nieder	17
Vinculin	< 10	nieder	23

AH: autoimmune Hepatitis MS: Morvan-Syndrome NM: Neuromyotonie
NMO: Neuromyelitis optica

* ein mit MuSK identisches Antigen wurde unter der Bezeichnung Protein p110 beschrieben 15

☞ Krankheitsverzeichnis ☞ Abkürzungen

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Lambert-Eaton myasthenisches Syndrom

Autoantikörper gegen	Sens [%]	Spez [%]	Krankheitsassoziation
Calciumkanäle (VGCC)	90 - 100	hoch	SCLC (60%), LE, CD, SN, DA
Calciumkanal $Ca_v2.1$ (P/Q-Typ)	90 - 100 *1	hoch	SN, DA 8, 10
Calciumkanal $Ca_v2.2$ (N-Typ)	33 - 49 *1	hoch	DA 7, 8, 10
Calciumkanal $Ca_v1.1$ (L-Typ)	Kasuistiken	unbekannt	1,5
Calciumkanal ß-Untereinheit	23 - 55	unbekannt	14, 15, 24
SOX1 *2	bis 67	hoch (SCLC)	Frühmarker für SCLC [21] 16, 20
Synaptotagmin 1	30 *3	unbekannt	2, 17
Acetylcholinrezeptor (AChRm)	Kasuistiken		MG/LEMS overlap 6, 11, 12
M1 mAcetylcholinrezeptor	70 *4	unbekannt	18, 19
Glutamatdecarboxylase (GAD)	35	unbekannt	4
IA-2 (Thyrosinphosphatase)	21	unbekannt	4
HuD	Kasuistiken *5		3, 9, 13, 22
CV2/CRMP5	5	unbekannt	16
PCA-2	Kasuistiken		23

CD: cerebellare Degeneration DA: Dysautonomie LE: limbische Enzephalitis
LEMS: Lambert-Eaten myastenisches Syndrom MG: Myasthenia gravis
SCLC: kleinzelliges Lungenkarzinom SN: sensorische Neuropathie

*1 weniger häufig bei paraneoplastischen Formen mit SCLC
*2 Kreuzreaktion mit SOX2, SOX3, SOX21
*3 wurde nicht bei allen beschriebenen Patientenkollektiven gefunden [2]
*4 Vorkommen auch bei anti-VGCC negativen LEMS-Patienten beschrieben [18, 19]
*5 9 % siehe Mason et al. (1997), [9]

siehe: Paraneoplastische Neuropathien

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Neuromyotonie

Autoantikörper gegen	Sens [%]	Spez [%]	Krankheitsassoziation	
Kaliumkanalkomplex *1	54 [13] - 95	hoch *2	Thymom , SCLC, DA	3, 4, 5
Kaliumkanal Kv1.1 *3	< 3			4, 7, 8
Kaliumkanal Kv1.2 *3	< 3			4, 7, 8
Kaliumkanal Kv1.6 *3	< 3			4, 7, 8
CASPR2	hoch	hoch *2		7, 9, 14
LGI1	nieder	hoch *2		2, 7
Tag-1/Contactin 2	nieder	hoch *2		7
Acetylcholinrezeptor (muskulärer)	14 [13]		Thymom	6, 13
Acetylcholinrezeptor (ganglionärer)	14 [13]		assozierte Tumore *4	12, 13
MuSK	Kasuistiken			11a
Ryanodinrezeptor (RyR)	Kasuistiken		Thymom	10
Amphiphysin	Kasuistiken		Paraneoplasie	11
Glutamatdecarboxylase (GAD)	Kasuistiken		Thymom	1

DA: Dysautonomie SCLC: kleinzelliges Lungenkarzinom

*1 Die Antikörper richten sich in den meisten Fällen nicht gegen die Kanalporen-bildenden (Kv1.1, 1.2, 1.6) sondern gegen die assoziierten Proteine CASPR2, LGI-1, Tag-1 (Kanal-Komplex).

*2 Gültig für Neuromyotonie, Morvan-Syndrom und limbische Enzephalitis.

*3 Autoantikörper nachgewiesen mittels IIFT an transfizierten, das entsprechende Kanalprotein exprimierenden Kulturzellen (4, 8); die Ergebnisse ließen sich nicht regelmäßig bestätigen (7).

*4 Thymom (anti-CRMP5 positiv), SCLC (anti-CRMP5 positiv), Lungenkarzinom (anti-Amphiphysin positiv), SCLC (ANNA positiv) (13).

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Morvan-Syndrom

Autoantikörper gegen	Sens [%]	Spez [%]	Krankheitsassoziation	
Kaliumkanalkomplex *1	40 - 80	hoch *3	LE, NM, EP, Thymom	1, 6, 7
Kaliumkanal Kv1.1 *2	< 3			4
Kaliumkanal Kv1.2 *2	< 3			4
Kaliumkanal Kv1.6 *2	< 3			4
CASPR2	hoch	hoch *3	LE, NM, EP	9
LGI1	nieder	hoch *3	LE, NM, EP	5
Tag-1/Contactin 2	nieder	hoch *3		8
Acetylcholinrezeptor (AChRm)	Kasuistik		Thymom, MG/MS	2, 3, 6
Titin	Kasuistik		Thymom	6
Skelettmuskel (IIFT)	Kasuistik		Thymom	6
Calciumkanal $Ca_v2.2$ (N-Typ)	Kasuistik			6

LE: limbische Enzephalitis NM Neuromyotonie EP Epilepsie, Krampfanfälle
MG/MS: Überlappungssyndrom mit Myasthenia gravis und anti-AChR, anti-MuSK und anti-VGKC

*1 Die Antikörper richten sich in den meisten Fällen nicht gegen die Kanalporen-bildenden (Kv1.1, 1.2, 1.6) sondern gegen die assoziierten Proteine CASPR2, LGI-1, Tag-1 (Kanal-Komplex).

*2 Autoantikörper nachgewiesen mittels IIFT an transfizierten, das entsprechende Kanalprotein exprimierenden Kulturzellen (4); die Ergebnisse ließen sich nicht regelmäßig bestätigen (vergl. Literatur: Neuromytonie 7).

*3 Gültig für Neuromyotonie, Morvan-Syndrom und limbische Enzephalitis.

siehe: Autoantikörper bei paraneoplastische Neuropathien

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Autoantikörper bei Erkrankungen der neuromuskulären Transmission

Abkürzungen

Sens	Sensitivität
Spez	Spezifität
CASPR2	contactin-assozioated protein-2
CV2/CRMP5	collapsing response mediator protein 5
Hu	Abkürzung eines Patientennamens
IA2	islet cell antigen 2
IIFT	indirekter Immunfluoreszenztest
GAD	Glutamatdecarboxylase
LGI1	leucine-rich glioma inactivated 1
Lrp4	low density lipoprotein receptor-related protein 4
M1 mAChR	M1 muskarinischer Acetylcholinrezeptor
MuSK	mukelspezifische Tyrosinkinase
PCA-2	pukinje cell antibody Typ 2
SOX1	sex determining region y-box 1
Tag-1/Contactin 2	transient axonal glycoprotein 1
TRPC3	transient receptor potential canonical type 3
VGCC	voltage gated calcium channel (Calciumkanal)
VGKC	voltage gated potassium channel (Kaliumkanal)

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