

## PANEL NMG11: Trastornos del movimiento

### DISTONIAS

DISTONIA POR DEFICIENCIA DE SEPIAPTERINA REDUCTASA CON RESPUESTA A DOPA	SPR
SÍNDROME DE SEGAWA (DÉFICIT SÍNTESIS DOPAMINA)	TH
	GCH1
	SLC6A3
DEFICIENCIA DE DOPAMINA BETA-HIDROXILASA : DEFICIENCIA DE NORADRENALINA	DBH
DÉFICIT DE DECARBOXILASA DE AMINOÁCIDO AROMÁTICO DECARBOXILASA	DDC
DÉFICIT DE ÁCIDO GAMMA AMINOBUTÍRICO (GABA) TRANSAMINASA	ABAT
DISTONIA, CON RESPUESTA A DOPA	GCH1
PARKINSONISMO-DISTONIA, INFANTIL; PKDYS	SLC6A3
DYSTONIA 1, TORSION, AUTOSOMAL DOMINANT; DYT1	TOR1A
DYSTONIA 11 MIOCLONICA	SGCE
MYOCLONIC DYSTONIA	DRD2
DYSTONIA 6, TORSION; DYT6	THAP1
DYSTONIA 12; DYT12	ATP1A3
DYSTONIA 18; DYT18	SLC2A1
DYSTONIA 10; DYT10	PRRT2
PARKINSONISM-DYSTONIA, INFANTILE; PKDYS	SLC6A3
DYSTONIA 16; DYT16	PRKRA
DYSTONIA, JUVENILE-ONSET	ACTB
DYSTONIA 8; DYT8	MR1
<b>ATAXIAS (NO INCLUYE AQUELLAS ASOCIADAS A EXPANSIONES TRINUCLEOTÍDICAS)</b>	
ANIRIDIA, CEREBELLAR ATAXIA , AND MENTAL RETARDATION	PAX6
ATAXIA CEREBELLAR, CAYMAN TYPE	ATCAY
ATAXIA WITH ISOLATED VITAMIN E DEFICIENCY	TTPA
ATAXIA, EARLY-ONSET, WITH OCULOMOTOR APRAXIA AND HYPOALBUMINEMIA	APTX
ATAXIA, SENSORY, 1, AUTOSOMAL DOMINANT	RNF170
ATAXIA, SPASTIC, 4	MTPAP
ATAXIA, SPASTIC, 5, AUTOSOMAL RECESSIVE	AFG3L2
ATAXIA-OCULAR APRAXIA-2	SETX
ATAXIA-OCULOMOTOR APRAXIA 3	PIK3R5
ATAXIA-TELANGIECTASIA	ATM
ATAXIA-TELANGIECTASIA-LIKE DISORDER	MRE11A
CEREBELLAR ATAXIA AND MENTAL RETARDATION WITH OR WITHOUT QUADRUPEDAL LOCOMOTION 3	CA8
CEREBELLAR ATAXIA, DEAFNESS, AND NARCOLEPSY, AUTOSOMAL DOMINANT	DNMT1
CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 2	WDR81
CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 4	ATP8A2

CEREBELLAR ATAXIA, NONPROGRESSIVE, WITH MENTAL RETARDATION	CAMTA1
CEREBELLAR ATAXIA, PROGRESSIVE DEMENTIA AND AMYLOID DEPOSITS IN CNS	PRNP
COGNITIVE IMPAIRMENT WITH OR WITHOUT CEREBELLAR ATAXIA	SCN8A
EPISODIC ATAXIA, TYPE 2/ MIGRAINE, FAMILIAL HEMIPLEGIC, 1/ SPINOCEREBELLAR ATAXIA 6	CACNA1A
EPISODIC ATAXIA, TYPE 5	CACNB4
EPISODIC ATAXIA, TYPE 6	SLC1A3
EPISODIC ATAXIA/MYOKYMIA SYNDROME	KCNA1
MITOCHONDRIAL DNA DEPLETION SYNDROME 7 (HEPATOCEREBRAL TYPE)	C10ORF2
MITOCHONDRIAL RECESSIVE ATAXIA SYNDROME (INCLUDES SANDO AND SCAE)	POLG
NIJMEGEN BREAKAGE SYNDROME	NBN
SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE	SACS
SPINOCEREBELLAR ATAXIA 5, SCA5	SPTBN2
SPINOCEREBELLAR ATAXIA 11, SCA11	TTBK2
SPINOCEREBELLAR ATAXIA 13, DOMINANT, SCA13	KCNC3
SPINOCEREBELLAR ATAXIA 14, SCA14	PRKCG
SPINOCEREBELLAR ATAXIA 15, SCA15, SCA16, SCA29	ITPR1
SPINOCEREBELLAR ATAXIA 20, SCA20	11Q12 DUP
SPINOCEREBELLAR ATAXIA 23, SCA23	PDYN
SPINOCEREBELLAR ATAXIA 27, SCA27	FGF14
SPINOCEREBELLAR ATAXIA 28, SCA28/ATAXIA, SPASTIC, 5, AUTOSOMAL RECESSIVE	AFG3L2
SPINOCEREBELLAR ATAXIA 35, SCA35	TGM6
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 5; SCAR5	ZNF592
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 8; SCAR8	SYNE1
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 9; SCAR9	CABC1:ADCK3
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10; SCAR10	ANO10
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 11; SCAR11	SYT14
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 13; SCAR13	GRM1
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 14; SCAR14	SPTBN2
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE WITH AXONAL NEUROPATHY	TDP1
SPINOCEREBELLAR ATAXIA, X-LINKED 1	ATP2B3
<b>Nuevos genes asociados a Ataxia</b>	
Identification of CHIP as a Novel Causative Gene for Autosomal Recessive Cerebellar Ataxia.	STUB1 (CHIP)
Spinocerebellar ataxia, autosomal recessive 12; SCAR12	WWOX
A mitochondrial origin for frontotemporal dementia and amyotrophic lateral sclerosis through CHCHD10 involvement.	CHCHD10
TMEM240 mutations cause spinocerebellar ataxia 21 with mental retardation and severe cognitive impairment	TMEM240
Mutations in KCND3 cause spinocerebellar ataxia type 22	KCND3
Recessive loss of function of the neuronal ubiquitin hydrolase UCHL1 leads to early-onset progressive neurodegeneration	UCHL1
Autosomal Recessive Spinocerebellar Ataxia 7 (SCAR7) is Caused by Variants inTPP1	TPP1
A novel locus for episodic ataxia:UBR4 the likely candidate	UBR4
Ataxia, Dementia, and Hypogonadotropism Caused by Disordered Ubiquitination	RNF216
De novo partial deletion in GRID2 presenting with complicated spastic paraplegia.	GRID2