

Table S1 Classification of Rare Neurological Diseases diagnosed per disease category

DIAGNOSIS	n	%
<i>Neurodegenerative diseases with dementia</i>	80	23
PPA	29	8.4
FTD	34	9.9
LBD	4	1.1
FTD-MND	2	0.55
Leukodystrophies	2	0.55
RTLA (FTD variant)	4	1.1
CJD	5	1.4
<i>Neurodegenerative diseases with parkinsonism</i>	11	3.2
CBD	8	2.3
Parkinson plus syndrome	3	0.9
<i>Neurodegenerative diseases with chorea</i>	9	2.6
Huntington disease	9	2.6
<i>Other rare neurodegenerative diseases</i>	10	3
MSA	7	2.1
PCA	3	0.9
<i>Rare motor neuron diseases</i>	64	18.7
ALS - MND	57	16.6
Mills syndrome	1	0.3
MND- Flail arm syndrome	1	0.3
Brown- Vialetto -Van Laere syndrome	1	0.3
SMA	3	0.9
Progressive muscular atrophy	1	0.3
<i>Rare peripheral neuropathies</i>	30	8.8
CIDP	8	2.3
Guillain Barre syndrome	7	2
Charcot Marie Tooth disease	6	1.8
Isaacs syndrome	3	0.9
MADSAM	2	0.6
hATTR	2	0.6
HNPP	1	0.3
MMN	1	0.3
<i>Rare diseases of the neuromuscular junction</i>	28	8.2
Myasthenia gravis	27	7.9
Congenital myasthenic syndrome	1	0.3
<i>Genetic skeletal muscle diseases</i>	19	5.6
Muscular dystrophy	12	3.5
Mitochondrial myopathy	2	0.6
Congenital myotonia	3	0.9

Qualitative or quantitative defects of dystrophin	1	0.3
Henson-Heyck-Landahn myopathy	1	0.3
<i>Rare neuroinflammatory and neuro-immunological disease</i>	19	5.6
NMOSD	7	2
Autoimmune encephalitis	4	1.1
MOGAD	3	0.9
Paraneoplastic neurologic syndrome	2	0.6
ADEM	1	0.3
PML-IRIS	1	0.3
IgG4-related pachymeningitis	1	0.3
<i>Rare movement disorders</i>	10	3
PSP	8	2.3
PSP-FTD complex	1	0.3
Primary orthostatic tremor	1	0.3
<i>Rare systemic or rheumatologic disease with CNS involvement</i>	11	3.2
Behçet disease	4	1.1
Systemic lupus erythematosus with CNS involvement	3	0.9
Giant cell arteritis	2	0.6
Sarcoidosis	1	0.3
Sjögren syndrome	1	0.3
<i>Rare vascular diseases of the CNS</i>	8	2.3
CADASIL	4	1.1
Moyamoya disease	2	0.6
Primary angiitis of the central nervous system	1	0.3
Familial cerebral cavernous malformation	1	0.3
<i>Rare ataxias</i>	7	2
SCA	1	0.3
Friedreich ataxia	3	0.9
Sporadic adult-onset cerebellar ataxia	2	0.6
Paraneoplastic cerebellar degeneration	1	0.3
<i>Rare epilepsies</i>	5	1.5
Juvenile myoclonic epilepsy	1	0.3
Mesial temporal lobe epilepsy with hippocampal sclerosis	1	0.3
Cerebral malformation with epilepsy	1	0.3
Lafora disease	1	0.3
Inflammatory and autoimmune disease with epilepsy	1	0.3
<i>Neurocutaneous syndrome with epilepsy</i>	6	1.8

Tuberous sclerosis complex	3	0.9
NF1	2	0.6
Sturge Weber Syndrome	1	0.3
<i>Acquired skeletal muscle diseases</i>	4	1.2
Immune-mediated necrotizing myopathy	2	0.6
Necrotic myopathy unspecified	1	0.3
Polymyositis	1	0.3
<i>Rare inborn errors of metabolism</i>	4	1.2
MELAS	2	0.6
Pompe disease	1	0.3
Argininemia	1	0.3
<i>CNS malformations</i>	2	0.6
Congenital hydrocephalus	2	0.6
<i>Rare headache</i>	2	0.6
Sporadic hemiplegic migraine	1	0.3
Idiopathic intracranial hypotension	1	0.3
<i>Medullar disease</i>	2	0.6
Transverse myelitis	2	0.6
<i>Rare sleep disorder</i>	1	0.3
Narcolepsy	1	0.3
<i>Rare nervous system tumor</i>	1	0.3
Medulloblastoma	1	0.3
<i>Other rare diseases</i>	9	2.6
Idiopathic intracranial hypertension	3	0.9
Cerebellar ataxia and atrophy	1	0.3
Congenital cerebellar agenesis	1	0.3
Boucher-Näühauser syndrome and cerebellar ataxia	1	0.3
MEN-1 syndrome	1	0.3
Autoimmune autonomic ganglionopathy	1	0.3
Endocardial fibroelastosis - ischemic stroke	1	0.3

PPA: primary progressive aphasia; FTD: frontotemporal dementia; LBD: lewy-body dementia, MND: motor neuron disease; RTLA: right temporal lobe atrophy; CJD: Creutzfeldt-Jakob disease; CBD: corticobasal degeneration; MSA: multiple system atrophy; PCA: posterior cortical atrophy; CNS: Central Nervous System; ALS- MND: amyotrophic lateral sclerosis -motor neuron disease; SMA: spinal muscular atrophy; CIDP: chronic inflammatory demyelinating polyneuropathy; MADSAM: multifocal acquired demyelinating sensory and motor neuropathy; hATTR: hereditary transthyretin amyloidosis; HNPP: hereditary neuropathy with pressure palsies; MMN: multifocal motor neuropathy; NMOSD: neuromyelitis optica spectrum disorder; MOGAD: myelin oligodendrocyte glycoprotein antibody disease; ADEM: acute disseminated encephalomyelitis; PML-IRIS: progressive multifocal leukoencephalopathy - immune reconstitution inflammatory syndrome; PSP: progressive supranuclear palsy; CADASIL: cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy; SCA: spinocerebellar atrophy; NF1: neurofibromatosis type 1; MELAS: mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes; MEN-1 multiple endocrine neoplasia type 1; n: number of patients