

## Supporting information

**S2 Table:** Table showing the complete list of included RD in the study

<b>Rare disease name</b>	<b>ORPHA code</b>		
		Congenital glaucoma	98976
Acromegaly	963	Congenital pulmonary valvar stenosis	3189
Acute intermittent porphyria	79276	Cowden syndrome	201
Addison disease	85138	CREST syndrome	90290
Adult-onset cervical dystonia	420492	Dermatomyositis	221
Alpha-1-antitrypsin deficiency	60	Disorder of fructose metabolism	308463
Amyotrophic lateral sclerosis	803	Distal myotilinopathy	98911
Antiphospholipid syndrome	80	Dural sinus malformation	97339
Antisynthetase syndrome	81	Dystrophic epidermolysis bullosa	303
Arachnoid cyst	2356	Ehlers-Danlos syndrome	98249
Arachnoiditis	137817	Eosinophilic granulomatosis with polyangiitis	183
Arnold-Chiari malformation type I	268882	Epidermolysis bullosa simplex	304
Autosomal dominant Charcot-Marie-Tooth disease type 2K	99944	Epithelioid hemangioendothelioma	157791
Autosomal erythropoietic protoporphyria	79278	Essential thrombocythemia	3318
Autosomal recessive cerebelloparenchymal disorder type 3	1170	Facioscapulohumeral dystrophy	269
Autosomal recessive spastic paraplegia type 76	488594	Familial drusen	75376
Behçet disease	117	Familial paroxysmal ataxia	97
Benign concentric annular macular dystrophy	251287	Focal, segmental or multifocal dystonia	1866
Bullous pemphigoid	703	Fragile X syndrome	908
Calpain-3-related limb-girdle muscular dystrophy R1	267	Friedreich ataxia	95
Caroli disease	53035	Gaucher disease type 1	77259
Catecholaminergic polymorphic ventricular tachycardia	3286	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	368
Chronic inflammatory demyelinating polyneuropathy	2932	Gyrate atrophy of choroid and retina	414
Classic mycosis fungoides	2584	Hereditary angioedema type 1	100050
Clippers	284448	Hereditary fructose intolerance	469
Combined dystonia	98203	Hereditary spastic paraplegia	685
Congenital fibrosis of extraocular muscles	45358	Hyperlipoproteinemia type 1	411
		Hypermobile Ehlers-Danlos syndrome	285

Idiopathic achalasia	930	Neuralgic amyotrophy	2901
Idiopathic hypersomnia	33208	Neurofibromatosis type 1	636
Idiopathic intracranial hypertension	238624	Neurofibromatosis type 2	637
Idiopathic multicentric Castleman disease	570431	Non-acquired panhypopituitarism	90695
Idiopathic pleuroparenchymal fibroelastosis	494428	Oculopharyngeal muscular dystrophy	270
Idiopathic pulmonary arterial hypertension	275766	Oligodontia	99798
Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	52430	Osteochondritis dissecans	2764
Indolent systemic mastocytosis	98848	Osteogenesis imperfecta	666
Interstitial cystitis	37202	Perineural cyst	65250
Isolated complex I deficiency	2609	Poland syndrome	2911
Isolated cytochrome C oxidase deficiency	254905	Polycythemia vera	729
Isolated microphthalmia-anophthalmia-coloboma	2542	Postpoliomyelitis syndrome	2942
Isolated optic neuritis	499096	Primary progressive aphasia	95432
Langerhans cell histiocytosis	389	Progressive cone dystrophy	1871
Leber hereditary optic neuropathy	104	Progressive hemifacial atrophy	1214
Legg-Calvé-Perthes disease	2380	Pseudoxanthoma elasticum	758
Limb-girdle muscular dystrophy	263	Pudendal neuralgia	60039
Lymphangioliomyomatosis	538	Pulmonary arterial hypertension associated with another disease	275791
Lynch syndrome	144	Rare dystonia	68363
Marfan syndrome	558	Rare pulmonary hypertension	71198
Melkersson-Rosenthal syndrome	2483	Renal nutcracker syndrome	71273
Mesial temporal lobe epilepsy with hippocampal sclerosis	99701	Retinitis pigmentosa	791
Mitochondrial myopathy	206966	Sarcoidosis	797
Monoclonal mast cell activation syndrome	529468	Selective IgM deficiency	331235
Monosomy 9p	261112	Sjögren-Larsson syndrome	816
Multiminicore myopathy	598	Spinocerebellar ataxia type 3	98757
Multiple endocrine neoplasia type 2A	247698	Stargardt disease	827
Multiple endocrine neoplasia type 2B	247709	Steinert myotonic dystrophy	273
Myasthenia gravis	589	Sturge-Weber syndrome	3205
Narcolepsy	619284	STXBP1-related encephalopathy	599373
Narcolepsy type 1	2073	Sympathetic ophthalmia	79098
		Systemic capillary leak syndrome	188
		Systemic mastocytosis	2467
		Systemic sclerosis	90291
		Thalidomide embryopathy	3312
		Thomsen and Becker disease	614
		Toxic oil syndrome	227972
		Trigeminal neuralgia	221091

Tumor necrosis factor receptor 1 associated periodic syndrome	32960	Vascular Ehlers-Danlos syndrome	286
Turner syndrome	881	Vestibular schwannoma	252175
Usher syndrome	886	Von Willebrand disease type 1	166078
Usher syndrome type 1	231169	X-linked adrenoleukodystrophy	43
Usher syndrome type 2	231178	X-linked Charcot-Marie-Tooth disease	64747