

PDR	U DMR	Aandoening	Orphacode	Patiëntenorganisatie
P 296	U 21	Anterior cutaneous nerve entrapment syndrome	ORPHA:51890	ACNES Foundation
P 296	U 22	Anterior cutaneous nerve entrapment syndrome	ORPHA:51890	ACNES Foundation
P 1	U 1837	Rare ataxia	ORPHA:102002	ADCA-Ataxie Vereniging
P 1	U 1838	Rare ataxia	ORPHA:102002	ADCA-Ataxie Vereniging
P 1	U 1839	Autosomal recessive cerebellar ataxia	ORPHA:1172	ADCA-Ataxie Vereniging
P 1	U 1840	Epilepsy and/or ataxia with myoclonus as major feature	ORPHA:306756	ADCA-Ataxie Vereniging
P 1	U 1841	Autosomal dominant cerebellar ataxia	ORPHA:99	ADCA-Ataxie Vereniging
P 1	U 1842	Autosomal dominant cerebellar ataxia	ORPHA:99	ADCA-Ataxie Vereniging
P 1	U 1907	CLIPPERS	ORPHA:284448	ADCA-Ataxie vereniging
P 268	U 90	Amyotrophic lateral sclerosis	ORPHA:803	ALS Patients Connected
P 268	U 91	Amyotrophic lateral sclerosis	ORPHA:803	ALS Patients Connected
P 268	U 92	amyotrophic lateral sclerosis-parkinsonism-dementia complex	ORPHA:90020	ALS Patients Connected
P 2	U 1821	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136	Alzheimer NL
P 2	U 1822	Pantothenate kinase-associated neurodegeneration	ORPHA:157850	Alzheimer NL
P 2	U 1823	Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:199354	Alzheimer NL
P 2	U 1827	neuronal intranuclear inclusion disease	ORPHA:2289	Alzheimer NL
P 2	U 1828	Frontotemporal dementia with motor neuron disease	ORPHA:275872	Alzheimer NL
P 2	U 1829	Frontotemporal dementia	ORPHA:282	Alzheimer NL
P 2	U 1830	Neurodegeneration with brain iron accumulation	ORPHA:385	Alzheimer NL
P 2	U 1831	PRKAR1B-related neurodegenerative dementia with intermediate filaments	ORPHA:412066	Alzheimer NL
P 2	U 1832	corticobasal syndrome	ORPHA:454887	Alzheimer NL
P 2	U 1833	HTRA1-related autosomal dominant cerebral small vessel disease	ORPHA:482077	Alzheimer NL
P 2	U 1834	Cathepsin A-related arteriopathy-strokes-leukoencephalopathy	ORPHA:575553	Alzheimer NL
P 2	U 1835	Juvenile neuronal ceroid lipofuscinosis	ORPHA:79264	Alzheimer NL
P 2	U 1836	Primary progressive aphasia	ORPHA:95432	Alzheimer NL
P 2	U 1861	primary progressive apraxia of speech	ORPHA:314566	Alzheimer NL
P 68	U 1212	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	ORPHA:420789	Apneuvereniging
P 109	U 633	Malignant peritoneal mesothelioma	ORPHA:168811	Asbest VN
P 109	U 634	Pleural mesothelioma	ORPHA:50251	Asbest VN
P 109	U 635	Pleural mesothelioma	ORPHA:50251	Asbest VN
P 87	U 886	Rare pancreatic disease	ORPHA:101937	AVKV - Alveeskliervereniging
P 87	U 887	Rare pancreatic disease	ORPHA:101937	AVKV - Alveeskliervereniging
P 87	U 888	Autoimmune pancreatitis	ORPHA:103919	AVKV - Alveeskliervereniging
P 87	U 889	Rare tumor of pancreas	ORPHA:180824	AVKV - Alveeskliervereniging
P 87	U 890	Rare tumor of pancreas	ORPHA:180824	AVKV - Alveeskliervereniging
P 87	U 891	Rare tumor of Pancreas	ORPHA:180824	AVKV - Alveeskliervereniging
P 87	U 892	Neuroendocrine neoplasms of the pancreas	ORPHA:506052	AVKV - Alveeskliervereniging
P 87	U 893	Rare digestive tract tumor	ORPHA:98059	AVKV - Alveeskliervereniging
P 316	U 1900	Rare arteriovenous malformation	ORPHA:211266	AVM in de hersenen
P 316	U 1901	rare arteriovenous malformation	ORPHA:211266	AVM in de hersenen
P 316	U 1902	Rare arteriovenous malformation	ORPHA:211266	AVM in de hersenen
P 273	U 87	Bardet-Biedl syndrome	ORPHA:110	Bardet Biedl syndroom ST
P 310	U 1824	Neuronal ceroid lipofuscinosis	ORPHA:216	Beat Batten
P 310	U 1825	Neuronal ceroid lipofuscinosis	ORPHA:216	Beat Batten
P 310	U 1826	CLN3-ziekte	ORPHA:228346	Beat Batten

P 205	U 189	Pudendal neuralgia	ORPHA:60039	Bekkenbodem4all
P 69	U 1115	Multiple endocrine neoplasia type 1	ORPHA:652	Belangengroep MEN
P 69	U 1116	Multiple endocrine neoplasia type 1	ORPHA:652	Belangengroep MEN
P 69	U 1117	Multiple endocrine neoplasia type 1	ORPHA:652	Belangengroep MEN
P 69	U 1118	Multiple endocrine neoplasia type 2	ORPHA:653	Belangengroep MEN
P 69	U 1119	Multiple endocrine neoplasia type 2	ORPHA:653	Belangengroep MEN
P 274	U 83	Leber hereditary optic neuropathy	ORPHA:104	Belangenvereniging LOA/LHON
P 274	U 84	Leber hereditary optic neuropathy	ORPHA:104	Belangenvereniging LOA/LHON
P 274	U 85	Hereditary optic neuropathy	ORPHA:98671	Belangenvereniging LOA/LHON
P 274	U 86	leber plus disease	ORPHA:99718	Belangenvereniging LOA/LHON
P 274	U 1942	Mitochondrial disease	ORPHA:68380	Belangenvereniging LOA/LHON
P 274	U 1943	Mitochondrial disease	ORPHA:68380	Belangenvereniging LOA/LHON
P 152	U 336	Interstitial lung disease	ORPHA:182095	Belangenvereniging Longfibrosepatiënten NL
P 152	U 337	Interstitial lung disease	ORPHA:182095	Belangenvereniging Longfibrosepatiënten NL
P 152	U 338	Interstitial lung disease	ORPHA:182095	Belangenvereniging Longfibrosepatiënten NL
P 152	U 339	Interstitial lung disease	ORPHA:182095	Belangenvereniging Longfibrosepatiënten NL
P 152	U 340	Idiopathic pulmonary fibrosis	ORPHA:2032	Belangenvereniging Longfibrosepatiënten NL
P 152	U 341	Idiopathic pulmonary fibrosis	ORPHA:2032	Belangenvereniging Longfibrosepatiënten NL
P 152	U 342	Interstitial lung disease specific to adulthood	ORPHA:264735	Belangenvereniging Longfibrosepatiënten NL
P 152	U 344	Exposure-related interstitial lung disease	ORPHA:264984	Belangenvereniging Longfibrosepatiënten NL
P 152	U 345	Hypersensitivity pneumonitis	ORPHA:31740	Belangenvereniging Longfibrosepatiënten NL
P 152	U 346	Rare disorder potentially indicated for lung transplant	ORPHA:506222	Belangenvereniging Longfibrosepatiënten NL
P 152	U 347	Idiopathic interstitial pneumonia	ORPHA:98300	Belangenvereniging Longfibrosepatiënten NL
P 152	U 348	Idiopathic interstitial pneumonia	ORPHA:98300	Belangenvereniging Longfibrosepatiënten NL
P 66	U 1120	Polyendocrinopathy	ORPHA:101956	Belangenvereniging Von Hippel-Lindau
P 66	U 1121	Von Hippel-Lindau disease	ORPHA:892	Belangenvereniging Von Hippel-Lindau
P 66	U 1122	Von Hippel-Lindau disease	ORPHA:892	Belangenvereniging Von Hippel-Lindau
P 66	U 1123	Von Hippel-Lindau disease	ORPHA:892	Belangenvereniging Von Hippel-Lindau
P 108	U 636	Isolated epispadias	ORPHA:93928	Blaasextrophie NL (BEN)
P 108	U 637	Bladder exstrophy	ORPHA:93930	Blaasextrophie NL (BEN)
P 4	U 1810	Malformation syndrome with short stature	ORPHA:139021	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1811	Growth hormone insensitivity syndrome	ORPHA:181393	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1812	SHOX-related short stature	ORPHA:314795	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1813	Primary bone dysplasia	ORPHA:364526	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1814	Primary Bone Dysplasia	ORPHA:364526	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1815	Menke-Hennekam syndrome	ORPHA:592574	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1816	Non-acquired isolated growth hormone deficiency	ORPHA:631	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1817	Silver-Russell syndrome	ORPHA:813	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1818	Rare Endocrine Growth Disease	ORPHA:90692	BVKM - Belangenvereniging van Kleine Mensen
P 4	U 1819	Primary bone dysplasia with decreased bone density	ORPHA:93446	BVKM - Belangenvereniging van Kleine Mensen
P 89	U 825	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL
P 89	U 826	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL
P 89	U 827	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL
P 89	U 828	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL
P 89	U 829	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL
P 89	U 830	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL

P 89	U 831	Hereditary breast and ovarian cancer syndrome	ORPHA:145	BVN - Borstkankervereniging NL
P 89	U 832	Rare malignant breast tumor	ORPHA:180257	BVN - Borstkankervereniging NL
P 89	U 836	Hereditary breast cancer	ORPHA:227535	BVN - Borstkankervereniging NL
P 89	U 837	Hereditary breast cancer	ORPHA:227535	BVN - Borstkankervereniging NL
P 89	U 838	Hereditary breast cancer	ORPHA:227535	BVN - Borstkankervereniging NL
P 89	U 839	Hereditary Breast Cancer	ORPHA:227535	BVN - Borstkankervereniging NL
P 89	U 840	Extramammary Paget disease	ORPHA:2800	BVN - Borstkankervereniging NL
P 114	U 601	Hydrops fetalis	ORPHA:1041	Care4Neo
P 114	U 602	Syndrome with a central nervous system malformation as a major feature	ORPHA:108991	Care4Neo
P 114	U 603	Non-syndromic respiratory or mediastinal malformation	ORPHA:108993	Care4Neo
P 114	U 604	Rare disorder related with pregnancy, childbirth and puerperium	ORPHA:163637	Care4Neo
P 114	U 605	Rare disorder related with pregnancy, childbirth and puerperium	ORPHA:163637	Care4Neo
P 114	U 606	HELLP syndrome	ORPHA:244242	Care4Neo
P 114	U 607	HELLP syndrome	ORPHA:244242	Care4Neo
P 114	U 608	Preeclampsia	ORPHA:275555	Care4Neo
P 114	U 609	Preeclampsia	ORPHA:275555	Care4Neo
P 114	U 610	Hemolytic disease due to fetomaternal alloimmunization	ORPHA:275938	Care4Neo
P 114	U 611	Fetal cytomegalovirus syndrome	ORPHA:294	Care4Neo
P 114	U 612	Fetal parvovirus syndrome	ORPHA:295	Care4Neo
P 114	U 613	Placental insufficiency	ORPHA:439167	Care4Neo
P 114	U 614	Placental insufficiency	ORPHA:439167	Care4Neo
P 114	U 615	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853	Care4Neo
P 114	U 617	Selective IUGR	Pending	Care4Neo
P 114	U 618	Twin anemia polycythemia sequence	Pending	Care4Neo
P 114	U 619	Twin reversed arterial perfusion sequence	Pending	Care4Neo
P 114	U 1918	Necrotizing enterocolitis	ORPHA:391673	Care4neo
P 114	U 1919	Necrotizing enterocolitis	ORPHA:391673	Care4neo
P 114	U 1920	Necrotizing enterocolitis	ORPHA:391673	Care4neo
P 114	U 616	Sepsis in premature infants	ORPHA:90051	Care4Neo
P 92	U 788	Rare arteriovenous malformation	ORPHA:211266	CMTC-OVM
P 92	U 789	rare arteriovenous malformation	ORPHA:211266	CMTC-OVM
P 92	U 790	Rare arteriovenous malformation	ORPHA:211266	CMTC-OVM
P 92	U 791	Rare arteriovenous malformation	ORPHA:211266	CMTC-OVM
P 92	U 798	Congenital primary lymphedema without systemic or visceral involvement	ORPHA:2416	CMTC-OVM
P 92	U 799	Late-onset primary lymphedema without systemic or visceral involvement	ORPHA:289825	CMTC-OVM
P 92	U 800	Cerebral arteriovenous malformation	ORPHA:46724	CMTC-OVM
P 92	U 801	Cerebral arteriovenous malformation	ORPHA:46724	CMTC-OVM
P 92	U 802	Primary lymphedema without systemic or visceral involvement	ORPHA:568041	CMTC-OVM
P 92	U 803	Vascular anomaly or angioma	ORPHA:68419	CMTC-OVM
P 92	U 804	Dural sinus malformation	ORPHA:97339	CMTC-OVM
P 92	U 805	Dural sinus malformation	ORPHA:97339	CMTC-OVM
P 147	U 353	Marfan and Marfan-related disorders	ORPHA:284993	Contactgroep Marfan NL
P 147	U 354	Marfan and Marfan-related disorders	ORPHA:284993	Contactgroep Marfan NL
P 147	U 355	Marfan and Marfan-related disorders	ORPHA:284993	Contactgroep Marfan NL
P 147	U 356	Marfan syndrome	ORPHA:558	Contactgroep Marfan NL
P 147	U 357	Marfan syndrome	ORPHA:558	Contactgroep Marfan NL

P 147	U 358	Marfan syndrome	ORPHA:558	Contactgroep Marfan NL
P 147	U 359	Loeys-Dietz syndrome	ORPHA:60030	Contactgroep Marfan NL
P 147	U 360	Loeys-Dietz syndrome	ORPHA:60030	Contactgroep Marfan NL
P 147	U 361	Rare surgical thoracic diseases	ORPHA:97962	Contactgroep Marfan NL
P 147	U 1879	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	Contactgroep Marfan NL
P 147	U 1880	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	Contactgroep Marfan NL
P 242	U 122	Rare tremor disorder	ORPHA:306712	Contactgroep Orthostatische tremor
P 176	U 221	Crigler-Najjar syndrome	ORPHA:205	Crigler-Najjar ST
P 176	U 222	Crigler-Najjar syndrome	ORPHA:205	Crigler-Najjar ST
P 119	U 18	Rare inflammatory bowel disease	ORPHA:104012	Crohn & Colitis NL
P 119	U 19	Rare inflammatory bowel disease	ORPHA:104012	Crohn & Colitis NL
P 119	U 565	Short bowel syndrome	ORPHA:104008	Crohn & Colitis NL
P 119	U 566	Short bowel syndrome	ORPHA:104008	Crohn & Colitis NL
P 119	U 567	Short bowel syndrome	ORPHA:104008	Crohn & Colitis NL
P 119	U 568	Short bowel Syndrome	ORPHA:104008	Crohn & Colitis NL
P 119	U 569	Rare intestinal disease	ORPHA:117569	Crohn & Colitis NL
P 119	U 570	Rare intestinal disease	ORPHA:117569	Crohn & Colitis NL
P 119	U 572	Pouchitis	ORPHA:217067	Crohn & Colitis NL
P 119	U 573	Anal fistula	ORPHA:228113	Crohn & Colitis NL
P 119	U 574	Anal fistula	ORPHA:228113	Crohn & Colitis NL
P 119	U 575	Anal fistula	ORPHA:228113	Crohn & Colitis NL
P 119	U 576	Congenital short bowel syndrome	ORPHA:2301	Crohn & Colitis NL
P 119	U 577	Chronic intestinal failure	ORPHA:294422	Crohn & Colitis NL
P 119	U 578	Chronic intestinal failure	ORPHA:294422	Crohn & Colitis NL
P 119	U 579	Chronic intestinal failure	ORPHA:294422	Crohn & Colitis NL
P 119	U 580	Chronic intestinal failure	ORPHA:294422	Crohn & Colitis NL
P 119	U 581	Primary short bowel syndrome	ORPHA:365563	Crohn & Colitis NL
P 119	U 585	Secondary short bowel syndrome	ORPHA:95427	Crohn & Colitis NL
P 119	U 586	Secondary short bowel syndrome	ORPHA:95427	Crohn & Colitis NL
P 119	U 1941	Idiopathic gastroparesis	ORPHA:558411	Crohn & Colitis NL
P 119	U 1944	Rare disease involving intestinal motility	ORPHA:104009	Crohn & Colitis NL
P 119	U 1945	Rare disease involving intestinal motility	ORPHA:104009	Crohn & Colitis NL
P 92	U 777	Cerebrofacial arteriovenous metamerisic syndrome	ORPHA:141189	CTMC-OVM
P 92	U 778	Facial arteriovenous malformation	ORPHA:156230	CTMC-OVM
P 92	U 779	Rare vascular tumor	ORPHA:211237	CTMC-OVM
P 92	U 780	Rare vascular tumor	ORPHA:211237	CTMC-OVM
P 92	U 781	Simple vascular malformation	ORPHA:211243	CTMC-OVM
P 92	U 782	Rare capillary malformation	ORPHA:211247	CTMC-OVM
P 92	U 783	Rare capillary malformation	ORPHA:211247	CTMC-OVM
P 92	U 784	Rare capillary malformation	ORPHA:211247	CTMC-OVM
P 92	U 785	Rare venous malformation	ORPHA:211252	CTMC-OVM
P 92	U 786	Rare venous malformation	ORPHA:211252	CTMC-OVM
P 92	U 787	Rare venous malformation	ORPHA:211252	CTMC-OVM
P 92	U 792	Complex vascular malformation with associated anomalies	ORPHA:211277	CTMC-OVM
P 92	U 793	Complex vascular malformation with associated anomalies	ORPHA:211277	CTMC-OVM
P 92	U 794	Complex vascular malformation with associated anomalies	ORPHA:211277	CTMC-OVM

P 92	U 795	Rare lymphatic malformation	ORPHA:2415	CTMC-OVM
P 92	U 796	Rare lymphatic malformation	ORPHA:2415	CTMC-OVM
P 92	U 797	Rare lymphatic malformation	ORPHA:2415	CTMC-OVM
P 281	U 72	Hereditary optic neuropathy	ORPHA:98671	Cure ADOA Foundation
P 281	U 73	Autosomal dominant optic atrophy	ORPHA:98672	Cure ADOA Foundation
P 281	U 1949	Mitochondrial disease	ORPHA:68380	Cure ADOA Foundation
P 281	U 1950	Mitochondrial disease	ORPHA:68380	Cure ADOA Foundation
P 132	U 428	Inherited epidermolysis bullosa	ORPHA:79361	Debra NL
P 132	U 429	Auto-immune bullous skin diseases	ORPHA:79669	Debra NL
P 103	U 667	Rare Diabetes Mellitus	ORPHA:101952	Diabetes Vereniging NL
P 70	U 1107	46,XY disorder of sex development of endocrine origin	ORPHA:325351	DSD NL
P 70	U 1108	Gender dysphoria	ORPHA:459690	DSD NL
P 70	U 1109	Disorders of sex development	ORPHA:90771	DSD NL
P 70	U 1110	Disorders of sex development	ORPHA:90771	DSD NL
P 70	U 1111	Posterior hypospadias	ORPHA:95706	DSD NL
P 70	U 1112	Posterior hypospadias	ORPHA:95706	DSD NL
P 70	U 1113	46,XX disorder of sex development induced by androgens excess	ORPHA:98078	DSD NL
P 6	U 1807	Duchenne and Becker muscular dystrophy	ORPHA:262	Duchenne Parent Project
P 6	U 1808	Duchenne and Becker muscular dystrophy	ORPHA:262	Duchenne Parent Project
P 6	U 1809	Duchenne and Becker muscular dystrophy	ORPHA:262	Duchenne Parent Project
P 59	U 1226	Paroxysmal dyskinesia	ORPHA:1431	Dystonie Vereniging
P 59	U 1227	Focal, segmental or multifocal dystonia	ORPHA:1866	Dystonie Vereniging
P 59	U 1228	Hemifacial spasm	ORPHA:221083	Dystonie Vereniging
P 59	U 1229	Rare choreic movement disorder	ORPHA:306715	Dystonie Vereniging
P 59	U 1230	Hyperekplexia	ORPHA:306773	Dystonie Vereniging
P 59	U 1231	Myoclonus-dystonia syndrome	ORPHA:36899	Dystonie Vereniging
P 59	U 1232	Generalized isolated dystonia	ORPHA:376724	Dystonie Vereniging
P 59	U 1233	Rare dystonia	ORPHA:68363	Dystonie Vereniging
P 59	U 1234	Rare dystonia	ORPHA:68363	Dystonie Vereniging
P 59	U 1235	Rare dystonia	ORPHA:68363	Dystonie Vereniging
P 59	U 1236	Psychogenic movement disorders	ORPHA:71519	Dystonie Vereniging
P 59	U 1237	Category Combined dystonia	ORPHA:98203	Dystonie Vereniging
P 309	U 1843	Vascular Ehlers-Danlos syndrome	ORPHA:286	EDS Fonds
P 309	U 1844	Ehlers-Danlos syndrome	ORPHA:98249	EDS Fonds
P 309	U 1845	Ehlers-Danlos syndrome	ORPHA:98249	EDS Fonds
P 285	U 67	Rare non-syndromic intellectual disability	ORPHA:101685	EMB NL
P 285	U 68	Rare non-syndromic intellectual disability	ORPHA:101685	EMB NL
P 285	U 69	Multiple congenital anomalies/dysmorphic syndrome without intellectual disability	ORPHA:102285	EMB NL
P 285	U 70	Rare syndromic intellectual disability	ORPHA:102369	EMB NL
P 285	U 71	Joubert syndrome and related disorders	ORPHA:140874	EMB NL
P 7	U 1791	Rare epilepsy	ORPHA:101998	Epilepsiefonds
P 7	U 1792	Epilepsy syndrome	ORPHA:166463	Epilepsiefonds
P 7	U 1793	Monogenic disease with epilepsy	ORPHA:166472	Epilepsiefonds
P 7	U 1794	Monogenic disease with epilepsy	ORPHA:166472	Epilepsiefonds
P 7	U 1795	Cerebral malformation with epilepsy	ORPHA:166478	Epilepsiefonds
P 7	U 1796	Cerebral malformation with epilepsy	ORPHA:166478	Epilepsiefonds

P 7	U 1797	Inflammatory and autoimmune disease with epilepsy	ORPHA:166484	Epilepsiefonds
P 7	U 1798	Limbic encephalitis with NMDA receptor antibodies	ORPHA:217253	Epilepsiefonds
P 7	U 1799	cerebral cortical dysplasia	ORPHA:268950	Epilepsiefonds
P 7	U 1800	Progressive myoclonic epilepsy type 6	ORPHA:280620	Epilepsiefonds
P 7	U 1801	CLIPPERS	ORPHA:284448	Epilepsiefonds
P 7	U 1802	Epilepsy and/or ataxia with myoclonus as major feature	ORPHA:306756	Epilepsiefonds
P 7	U 1803	Dravet syndrome	ORPHA:33069	Epilepsiefonds
P 7	U 1804	continuous spikes and waves during sleep	ORPHA:725	Epilepsiefonds
P 7	U 1805	Benign adult familial myoclonic epilepsy	ORPHA:86814	Epilepsiefonds
P 7	U 1806	Infantile Epilepsy Syndrome	ORPHA:98258	Epilepsiefonds
P 101	U 668	Genetic photodermatosis	ORPHA:183490	EPP - Erytroepitische Protoporfyrine Vereniging
P 8	U 1790	Non-recovering obstetric brachial plexus lesion	ORPHA:439202	Erbse Parese Vereniging NL
P 209	U 178	Rare deafness	ORPHA:68361	FODOK - Federatie van ouders van dove kinderen
P 209	U 179	Rare deafness	ORPHA:68361	FODOK - Federatie van ouders van dove kinderen
P 209	U 180	Rare deafness	ORPHA:68361	FODOK - Federatie van ouders van dove kinderen
P 162	U 266	Fibrodysplasia Ossificans Progressiva	ORPHA:337	FOP ST NL
P 162	U 267	Primary bone dysplasia with increased bone density	ORPHA:93444	FOP ST NL
P 162	U 268	Primary bone dysplasia with disorganized development of skeletal components	ORPHA:93450	FOP ST NL
P 10	U 1786	Fragile X syndrome	ORPHA:908	Fragiele X Vereniging
P 110	U 626	Absence of uterine body	ORPHA:180142	Freya
P 110	U 627	Rare genetic male infertility	ORPHA:399980	Freya
P 110	U 628	Genetic non-acquired premature ovarian failure	ORPHA:485382	Freya
P 110	U 629	Acquired premature ovarian failure	ORPHA:95709	Freya
P 110	U 630	Acquired premature ovarian failure	ORPHA:95709	Freya
P 110	U 631	Non acquired premature ovarian failure	ORPHA:95710	Freya
P 110	U 632	Rare male infertility	ORPHA:98048	Freya
P 9	U 1787	Frontotemporal dementia with motor neuron disease	ORPHA:275872	FTD Lotgenoten
P 9	U 1788	Frontotemporal dementia	ORPHA:282	FTD Lotgenoten
P 9	U 1789	amyotrophic lateral sclerosis-parkinsonism-dementia complex	ORPHA:90020	FTD Lotgenoten
P 9	U 1908	primary progressive apraxia of speech	ORPHA:314566	FTD Lotgenoten
P 12	U 1779	Disorder of galactose metabolism	ORPHA:308467	Galactosemievereniging
P 12	U 1780	Galactosemia	ORPHA:352	Galactosemievereniging
P 12	U 1781	Galactose mutarotase deficiency	ORPHA:570422	Galactosemievereniging
P 12	U 1782	Galactokinase deficiency	ORPHA:79237	Galactosemievereniging
P 12	U 1783	Galactose epimerase deficiency	ORPHA:79238	Galactosemievereniging
P 12	U 1784	Classic galactosemia	ORPHA:79239	Galactosemievereniging
P 302	U 16	Familial benign chronic pemphigus (Hailey-Hailey disease)	ORPHA:2841	Hailey Hailey
P 302	U 17	Familial benign chronic pemphigus (Hailey-Hailey disease)	ORPHA:2841	Hailey Hailey
P 13	U 1774	Arthrogyposis multiplex congenita	ORPHA:1037	Handvereniging
P 13	U 1775	Non-syndromic limb malformation	ORPHA:109011	Handvereniging
P 13	U 1776	Syndrome with limb reduction defects	ORPHA:294955	Handvereniging
P 13	U 1777	Congenital limb malformation	ORPHA:68378	Handvereniging
P 13	U 1778	Non-syndromic limb reduction defect	ORPHA:93457	Handvereniging
P 58	U 1238	Genetic cardiac rhythm disease	ORPHA:101934	Harteraad
P 58	U 1239	Genetic cardiac rhythm disease	ORPHA:101934	Harteraad
P 58	U 1240	Genetic cardiac rhythm disease	ORPHA:101934	Harteraad

P 58	U 1242	Brugada syndrome	ORPHA:130	Harteraad
P 58	U 1244	Rare cardiomyopathy	ORPHA:167848	Harteraad
P 58	U 1245	Rare cardiomyopathy	ORPHA:167848	Harteraad
P 58	U 1246	Rare cardiomyopathy	ORPHA:167848	Harteraad
P 58	U 1247	Rare Cardiomyopathy	ORPHA:167848	Harteraad
P 58	U 1248	Rare cardiomyopathy	ORPHA:167848	Harteraad
P 58	U 1249	Hypertrophic cardiomyopathy	ORPHA:217569	Harteraad
P 58	U 1250	Dilated cardiomyopathy	ORPHA:217604	Harteraad
P 58	U 1251	Dilated cardiomyopathy	ORPHA:217604	Harteraad
P 58	U 1252	Restrictive cardiomyopathy	ORPHA:217632	Harteraad
P 58	U 1253	Rare cardiac rhythm disease	ORPHA:218436	Harteraad
P 58	U 1254	Rare cardiac rhythm disease	ORPHA:218436	Harteraad
P 58	U 1255	Rare cardiac rhythm disease	ORPHA:218436	Harteraad
P 58	U 1256	Non-genetic cardiac rhythm disease	ORPHA:218439	Harteraad
P 58	U 1257	Idiopathic ventricular fibrillation - not Brugada type	ORPHA:228140	Harteraad
P 58	U 1260	Arrhythmogenic right ventricular cardiomyopathy	ORPHA:247	Harteraad
P 58	U 1261	Arrhythmogenic right ventricular cardiomyopathy	ORPHA:247	Harteraad
P 58	U 1263	Dysbetalipoproteinemia	ORPHA:412	Harteraad
P 58	U 1264	Pulmonary veno-occlusive disease and/or pulmonary capillary haemangiomas	ORPHA:431353	Harteraad
P 58	U 1265	Idiopathic spontaneous coronary artery dissection	ORPHA:458718	Harteraad
P 58	U 1274	Left ventricular noncompaction	ORPHA:54260	Harteraad
P 58	U 1275	Peripartum cardiomyopathy	ORPHA:563	Harteraad
P 58	U 1276	Infective endocarditis	ORPHA:570762	Harteraad
P 58	U 1279	Familial long QT syndrome	ORPHA:768	Harteraad
P 58	U 1280	Hereditary Hemorrhagic Telangiectasia	ORPHA:774	Harteraad
P 58	U 1282	Rare congenital non-syndromic heart malformation	ORPHA:88991	Harteraad
P 58	U 1283	Rare congenital non-syndromic heart malformation	ORPHA:88991	Harteraad
P 58	U 1284	Rare congenital non-syndromic heart malformation	ORPHA:88991	Harteraad
P 58	U 1285	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	Harteraad
P 58	U 1286	Familial thoracic aortic aneurysm and aortic dissection	ORPHA:91387	Harteraad
P 58	U 1288	Cardiogenic Shock	ORPHA:97292	Harteraad
P 58	U 1289	Rare cardiac diseases	ORPHA:97929	Harteraad
P 58	U 1290	Rare surgical cardiac disease	ORPHA:97965	Harteraad
P 58	U 1291	Rare Surgical Cardiac Disease	ORPHA:97965	Harteraad
P 58	U 1292	Rare familial disorder with hypertrophic cardiomyopathy	ORPHA:99739	Harteraad
P 58	U 1857	Rare dyslipidemia	ORPHA:101953	Harteraad
P 58	U 1858	Rare dyslipidemia	ORPHA:101953	Harteraad
P 58	U 1859	Rare dyslipidemia	ORPHA:101953	Harteraad
P 58	U 1888	Arterial thoracic outlet syndrome	ORPHA:357107	Harteraad
P 58	U 1889	Venous thoracic outlet syndrome	ORPHA:357131	Harteraad
P 113	U 620	HELLP syndrome	ORPHA:244242	Hellp ST
P 113	U 621	HELLP syndrome	ORPHA:244242	Hellp ST
P 113	U 622	Preeclampsia	ORPHA:275555	Hellp ST
P 91	U 806	Neonatal hypoxic and ischemic brain injury	ORPHA:137577	Hersenletsel.nl
P 91	U 807	Nasal ganglioglioma	ORPHA:141115	Hersenletsel.nl
P 91	U 808	Glial tumor	ORPHA:182067	Hersenletsel.nl

P 91	U 809	Glial tumor	ORPHA:182067	Hersenletsel.nl
P 91	U 810	Glial Tumor	ORPHA:182067	Hersenletsel.nl
P 91	U 811	Esthesioneuroblastoma	ORPHA:1957	Hersenletsel.nl
P 91	U 812	Esthesioneuroblastoma	ORPHA:1957	Hersenletsel.nl
P 91	U 813	Meningioma	ORPHA:2495	Hersenletsel.nl
P 91	U 814	Meningioma	ORPHA:2495	Hersenletsel.nl
P 91	U 815	Meningioma	ORPHA:2495	Hersenletsel.nl
P 91	U 816	Rare tumor of the neuroepithelial tissue	ORPHA:251558	Hersenletsel.nl
P 91	U 817	Rare tumor of neuroepithelial tissue	ORPHA:251558	Hersenletsel.nl
P 91	U 818	Rare tumors of neuroepithelial tissue	ORPHA:251558	Hersenletsel.nl
P 91	U 819	Medulloblastoma	ORPHA:616	Hersenletsel.nl
P 91	U 820	Rare nervous system tumor	ORPHA:98062	Hersenletsel.nl
P 91	U 1896	Neurovascular malformation	ORPHA:102006	Hersenletsel.nl
P 91	U 1897	Neurovascular malformation	ORPHA:102006	Hersenletsel.nl
P 91	U 1906	Moyamoya disease	ORPHA:2573	Hersenletsel.nl
P 91	U 1909	Pediatric arterial ischemic stroke	ORPHA:439175	Hersenletsel.nl
P 91	U 1910	Moyamoya angiopathy	ORPHA:477768	Hersenletsel.nl
P 91	U 1911	Rare disorder with a moyamoya angiopathy	ORPHA:477771	Hersenletsel.nl
P 91	U 1912	Acute Disseminated Encephalomyelitis	ORPHA:83597	Hersenletsel.nl
P 131	U 430	Cerebrofacial arteriovenous metameris syndrome	ORPHA:141189	HEVAS
P 131	U 431	Facial arteriovenous malformation	ORPHA:156230	HEVAS
P 131	U 432	Rare vascular tumor	ORPHA:211237	HEVAS
P 131	U 433	Rare vascular tumor	ORPHA:211237	HEVAS
P 131	U 434	Simple vascular malformation	ORPHA:211243	HEVAS
P 131	U 435	Rare capillary malformation	ORPHA:211247	HEVAS
P 131	U 436	Rare capillary malformation	ORPHA:211247	HEVAS
P 131	U 437	Rare capillary malformation	ORPHA:211247	HEVAS
P 131	U 438	Rare venous malformation	ORPHA:211252	HEVAS
P 131	U 439	Rare venous malformation	ORPHA:211252	HEVAS
P 131	U 440	Rare venous malformation	ORPHA:211252	HEVAS
P 131	U 441	Rare arteriovenous malformation	ORPHA:211266	HEVAS
P 131	U 442	rare arteriovenous malformation	ORPHA:211266	HEVAS
P 131	U 443	Rare arteriovenous malformation	ORPHA:211266	HEVAS
P 131	U 444	Rare arteriovenous malformation	ORPHA:211266	HEVAS
P 131	U 445	Complex vascular malformation with associated anomalies	ORPHA:211277	HEVAS
P 131	U 446	Complex vascular malformation with associated anomalies	ORPHA:211277	HEVAS
P 131	U 447	Complex vascular malformation with associated anomalies	ORPHA:211277	HEVAS
P 131	U 448	Rare lymphatic malformation	ORPHA:2415	HEVAS
P 131	U 449	Rare lymphatic malformation	ORPHA:2415	HEVAS
P 131	U 450	Rare lymphatic malformation	ORPHA:2415	HEVAS
P 131	U 451	Congenital primary lymphedema without systemic or visceral involvement	ORPHA:2416	HEVAS
P 131	U 452	Late-onset primary lymphedema without systemic or visceral involvement	ORPHA:289825	HEVAS
P 131	U 453	Cerebral arteriovenous malformation	ORPHA:46724	HEVAS
P 131	U 454	Cerebral arteriovenous malformation	ORPHA:46724	HEVAS
P 131	U 455	Primary lymphedema without systemic or visceral involvement	ORPHA:568041	HEVAS
P 131	U 456	Vascular anomaly or angioma	ORPHA:68419	HEVAS

P 131	U 457	Dural sinus malformation	ORPHA:97339	HEVAS
P 131	U 458	Dural sinus malformation	ORPHA:97339	HEVAS
P 131	U 1956	Progressive hemifacial atrophy	ORPHA:1214	HEVAS
P 131	U 1957	Hemifacial hyperplasia	ORPHA:141145	HEVAS
P 15	U 1763	Ollier disease	ORPHA:296	HME-MO Vereniging NL
P 15	U 1764	Multiple osteochondromas	ORPHA:321	HME-MO Vereniging NL
P 15	U 1765	Multiple osteochondromas	ORPHA:321	HME-MO Vereniging NL
P 15	U 1766	Primary bone dysplasia	ORPHA:364526	HME-MO Vereniging NL
P 15	U 1767	Primary Bone Dysplasia	ORPHA:364526	HME-MO Vereniging NL
P 15	U 1769	Rare bone tumor	ORPHA:68411	HME-MO Vereniging NL
P 15	U 1770	Rare bone tumor	ORPHA:68411	HME-MO Vereniging NL
P 15	U 1771	Solitary bone cyst	ORPHA:83468	HME-MO Vereniging NL
P 15	U 1772	Primary bone dysplasia with disorganized development of skeletal components	ORPHA:93450	HME-MO Vereniging NL
P 32	U 1549	Paroxysmal Hemicrania	ORPHA:157835	Hoofdpijnnet
P 32	U 1550	Trigeminal autonomic cephalgia	ORPHA:157843	Hoofdpijnnet
P 32	U 1551	Rare Genetic Headache	ORPHA:183509	Hoofdpijnnet
P 32	U 1552	Trigeminal neuralgia	ORPHA:221091	Hoofdpijnnet
P 32	U 1553	Trigeminal neuralgia	ORPHA:221091	Hoofdpijnnet
P 32	U 1554	Glossopharyngeal neuralgia	ORPHA:221098	Hoofdpijnnet
P 32	U 1555	Hypnic headache	ORPHA:276429	Hoofdpijnnet
P 32	U 1556	Visual Snow syndrome	ORPHA:420556	Hoofdpijnnet
P 32	U 1557	Hemicrania continua	ORPHA:443070	Hoofdpijnnet
P 32	U 1558	Familial or sporadic hemiplegic migraine	ORPHA:569	Hoofdpijnnet
P 32	U 1559	SUNCT-syndrome	ORPHA:57145	Hoofdpijnnet
P 32	U 1560	Rare headache	ORPHA:98022	Hoofdpijnnet
P 315	U 1890	Neurovascular malformation	ORPHA:102006	HPP - Hersenaneurysma Patiëntenplatform
P 315	U 1891	Neurovascular malformation	ORPHA:102006	HPP - Hersenaneurysma Patiëntenplatform
P 315	U 1892	Familial cerebral saccular aneurysm	ORPHA:231160	HPP - Hersenaneurysma Patiëntenplatform
P 315	U 1893	Acquired aneurysmal subarachnoid hemorrhage	ORPHA:90065	HPP - Hersenaneurysma Patiëntenplatform
P 315	U 1894	Acquired aneurysmal subarachnoid hemorrhage	ORPHA:90065	HPP - Hersenaneurysma Patiëntenplatform
P 315	U 1895	Acquired aneurysmal subarachnoid hemorrhage	ORPHA:90065	HPP - Hersenaneurysma Patiëntenplatform
P 315	U 1904	Familial cerebral saccular aneurysm	ORPHA:231160	HPP - Hersenaneurysma Patiëntenplatform
P 161	U 269	Aspergillosis	ORPHA:1163	Huid NL
P 161	U 270	Allergic bronchopulmonary aspergillosis	ORPHA:1164	Huid NL
P 161	U 271	Chronic mucocutaneous candidiasis	ORPHA:1334	Huid NL
P 161	U 272	Necrobiotic xanthogranuloma	ORPHA:158011	Huid NL
P 161	U 273	Rare mycosis	ORPHA:163591	Huid NL
P 161	U 274	Genetic epidermal appendage anomaly	ORPHA:183447	Huid NL
P 161	U 275	Genetic dermis disorder	ORPHA:183472	Huid NL
P 161	U 276	Congenital lethal erythroderma	ORPHA:1954	Huid NL
P 161	U 277	Darier disease	ORPHA:218	Huid NL
P 161	U 278	Darier disease	ORPHA:218	Huid NL
P 161	U 279	Dermatomyositis	ORPHA:221	Huid NL
P 161	U 280	Familial benign chronic pemphigus (Hailey-Hailey disease)	ORPHA:2841	Huid NL
P 161	U 281	Familial benign chronic pemphigus (Hailey-Hailey disease)	ORPHA:2841	Huid NL
P 161	U 282	Vascular Ehlers-Danlos syndrome	ORPHA:286	Huid NL

P 161	U 283	Systemic disease with skin involvement	ORPHA:290836	Huid NL
P 161	U 284	Complication after organ transplantation	ORPHA:306644	Huid NL
P 161	U 285	Gorlin syndrome	ORPHA:377	Huid NL
P 161	U 286	Histoplasmosis	ORPHA:390	Huid NL
P 161	U 287	Leishmaniasis	ORPHA:507	Huid NL
P 161	U 288	Leprosy	ORPHA:548	Huid NL
P 161	U 289	Rare genetic skin disease	ORPHA:68346	Huid NL
P 161	U 290	Rare genetic skin disease	ORPHA:68346	Huid NL
P 161	U 291	Ichthyosis	ORPHA:79354	Huid NL
P 161	U 292	Ichthyosis	ORPHA:79354	Huid NL
P 161	U 293	Erythrokeratoderma	ORPHA:79355	Huid NL
P 161	U 294	Hereditary palmoplantar keratoderma	ORPHA:79357	Huid NL
P 161	U 295	Hereditary palmoplantar keratoderma	ORPHA:79357	Huid NL
P 161	U 296	Scrub typhus	ORPHA:83317	Huid NL
P 161	U 297	Xeroderma Pigmentosum	ORPHA:910	Huid NL
P 318	U 1864	Rare skin tumor or hamartoma	ORPHA:79386	HUKAS - Huidkanker ST
P 107	U 638	Rare hereditary hemochromatosis	ORPHA:220489	HVN - Hemochromatose Vereniging NL
P 107	U 639	Disorder of iron metabolism and transport	ORPHA:309842	HVN - Hemochromatose Vereniging NL
P 173	U 223	Interstitial cystitis	ORPHA:37202	ICP - Interstitiële Cystitis Patiëntenvereniging
P 79	U 1003	Rare hemorrhagic disorder due to an acquired platelet anomaly	ORPHA:248347	ITP vereniging
P 79	U 1004	Immune thrombocytopenia	ORPHA:3002	ITP vereniging
P 79	U 1005	Immune thrombocytopenia	ORPHA:3002	ITP vereniging
P 79	U 1006	Autoimmune thrombocytopenia	ORPHA:71203	ITP vereniging
P 134	U 416	Rare systemic or rheumatologic diseases of childhood	ORPHA:280342	Jeugdreuma Vereniging NL
P 134	U 417	Rare systemic or rheumatologic diseases of childhood	ORPHA:280342	Jeugdreuma Vereniging NL
P 134	U 418	Polyarticular juvenile idiopathic arthritis	ORPHA:404580	Jeugdreuma Vereniging NL
P 134	U 419	Oligoarticular juvenile idiopathic arthritis	ORPHA:85410	Jeugdreuma Vereniging NL
P 134	U 420	Systemic-onset juvenile idiopathic arthritis	ORPHA:85414	Jeugdreuma Vereniging NL
P 134	U 421	Psoriasis-related juvenile idiopathic arthritis	ORPHA:85436	Jeugdreuma Vereniging NL
P 134	U 422	Juvenile idiopathic arthritis	ORPHA:92	Jeugdreuma Vereniging NL
P 134	U 423	Juvenile idiopathic arthritis	ORPHA:92	Jeugdreuma Vereniging NL
P 134	U 424	Juvenile idiopathic arthritis	ORPHA:92	Jeugdreuma Vereniging NL
P 134	U 425	Juvenile idiopathic arthritis	ORPHA:92	Jeugdreuma Vereniging NL
P 134	U 426	Juvenile idiopathic arthritis	ORPHA:92	Jeugdreuma Vereniging NL
P 134	U 427	Juvenile idiopathic arthritis	ORPHA:92	Jeugdreuma Vereniging NL
P 289	U 50	Pantothenate kinase-associated neurodegeneration	ORPHA:157850	Kans voor PKAN-kinderen
P 289	U 51	Neurodegeneration with brain iron accumulation	ORPHA:385	Kans voor PKAN-kinderen
P 17	U 170	Overgrowth syndrome	ORPHA:93460	LaPosa
P 17	U 1721	Aplasia cutis congenita	ORPHA:1114	LaPosa
P 17	U 1722	Beckwith Wiedemann syndrome	ORPHA:116	LaPosa
P 17	U 1723	Progressive hemifacial atrophy	ORPHA:1214	LaPosa
P 17	U 1724	branchial arch or oral-acral syndrome	ORPHA:139036	LaPosa
P 17	U 1725	Isolated craniosynostosis	ORPHA:139390	LaPosa
P 17	U 1726	Isolated craniosynostosis	ORPHA:139390	LaPosa
P 17	U 1727	Syndromic craniosynostosis	ORPHA:139393	LaPosa
P 17	U 1728	Syndromic craniosynostosis	ORPHA:139393	LaPosa

P 17	U 1729	Nasal glial heterotopia	ORPHA:141112	LaPosa
P 17	U 1730	Oculo-auriculo-vertebral spectrum	ORPHA:141132	LaPosa
P 17	U 1731	Oculo-auriculo-vertebral spectrum	ORPHA:141132	LaPosa
P 17	U 1732	Hemifacial hyperplasia	ORPHA:141145	LaPosa
P 17	U 1733	Facial cleft	ORPHA:141229	LaPosa
P 17	U 1734	Facial cleft	ORPHA:141229	LaPosa
P 17	U 1735	Cleidocranial dysplasia	ORPHA:1452	LaPosa
P 17	U 1736	Cleidocranial dysplasia	ORPHA:1452	LaPosa
P 17	U 1737	Craniosynostosis	ORPHA:1531	LaPosa
P 17	U 1738	Cysts and fistulae of the face and oral cavity	ORPHA:155835	LaPosa
P 17	U 1739	Otomandibular dysplasia	ORPHA:155896	LaPosa
P 17	U 1740	Otomandibular dysplasia	ORPHA:155896	LaPosa
P 17	U 1741	Macroglossia	ORPHA:156207	LaPosa
P 17	U 1742	Hypoglossia/aglossia	ORPHA:156212	LaPosa
P 17	U 1743	Paralytic facial malformation	ORPHA:156224	LaPosa
P 17	U 1744	Paralytic facial malformation	ORPHA:156224	LaPosa
P 17	U 1745	Syndrome or malformation associated with head and neck malformations	ORPHA:156237	LaPosa
P 17	U 1746	Syndrome or malformation associated with head and neck malformations	ORPHA:156237	LaPosa
P 17	U 1747	Rare odontal or periodontal disorder	ORPHA:164001	LaPosa
P 17	U 1748	Rare odontal or periodontal disorder	ORPHA:164001	LaPosa
P 17	U 1749	Temporomandibular joint anomaly	ORPHA:210581	LaPosa
P 17	U 1750	Temporomandibular joint anomaly	ORPHA:210581	LaPosa
P 17	U 1751	Cephalocele	ORPHA:268817	LaPosa
P 17	U 1752	Dysostosis	ORPHA:364559	LaPosa
P 17	U 1753	Primary condylar hyperplasia	ORPHA:477781	LaPosa
P 17	U 1754	Microtia	ORPHA:83463	LaPosa
P 17	U 1755	Microtia	ORPHA:83463	LaPosa
P 17	U 1756	Cleidocranial dysplasia and isolated cranial ossification defect	ORPHA:93451	LaPosa
P 17	U 1757	Dysostosis with predominant craniofacial involvement	ORPHA:93453	LaPosa
P 17	U 1758	Acalvaria	ORPHA:945	LaPosa
P 17	U 1759	Rare otorhinolaryngological malformation	ORPHA:96333	LaPosa
P 17	U 1926	Nasal encephalocele	ORPHA:141118	LaPosa
P 98	U 700	Renal cell carcinoma	ORPHA:217071	Leven met blaas- of nierkanker
P 98	U 1855	Rare urinary tract tumor	ORPHA:98058	Leven met blaas- of nierkanker
P 98	U 1856	Rare urinary tract tumour	ORPHA:98058	Leven met blaas- of nierkanker
P 200	U 196	Interstitial lung disease	ORPHA:182095	LGD Alliance NL
P 200	U 197	Interstitial lung disease	ORPHA:182095	LGD Alliance NL
P 200	U 198	Interstitial lung disease	ORPHA:182095	LGD Alliance NL
P 200	U 199	Interstitial lung disease	ORPHA:182095	LGD Alliance NL
P 200	U 200	Rare vascular tumor	ORPHA:211237	LGD Alliance NL
P 200	U 201	Rare vascular tumor	ORPHA:211237	LGD Alliance NL
P 200	U 202	Rare lymphatic malformation	ORPHA:2415	LGD Alliance NL
P 200	U 203	Rare lymphatic malformation	ORPHA:2415	LGD Alliance NL
P 200	U 204	Rare lymphatic malformation	ORPHA:2415	LGD Alliance NL
P 200	U 205	Genetic interstitial lung disease	ORPHA:264992	LGD Alliance NL
P 200	U 206	Genetic interstitial lung disease	ORPHA:264992	LGD Alliance NL

P 200	U 207	Primary bone dysplasia with disorganized development of skeletal components	ORPHA:93450	LGD Alliance NL
P 260	U 96	Rare carcinoma of pancreas	ORPHA:217074	Living with Hope
P 260	U 97	Neuroendocrine neoplasms of the pancreas	ORPHA:506052	Living with hope
P 121	U 520	Rare pulmonary disease	ORPHA:101944	Longfonds
P 121	U 521	Severe acute respiratory syndrome	ORPHA:140896	Longfonds
P 121	U 523	Respiratory malformation	ORPHA:182111	Longfonds
P 121	U 524	Congenital lobar emphysema	ORPHA:1928	Longfonds
P 121	U 527	Primary pulmonary hypoplasia	ORPHA:2257	Longfonds
P 121	U 528	Bronchogenic cyst	ORPHA:2357	Longfonds
P 121	U 529	Congenital pulmonary airway malformation	ORPHA:2444	Longfonds
P 121	U 530	Congenital pulmonary airway malformation	ORPHA:2444	Longfonds
P 121	U 531	Interstitial lung disease in childhood and adulthood	ORPHA:264757	Longfonds
P 121	U 532	Secondary interstitial lung disease in childhood and adulthood associated with a systemic	ORPHA:264949	Longfonds
P 121	U 533	Exposure-related interstitial lung disease	ORPHA:264984	Longfonds
P 121	U 534	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	Longfonds
P 121	U 535	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	Longfonds
P 121	U 536	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	Longfonds
P 121	U 537	Congenital pulmonary sequestration	ORPHA:3161	Longfonds
P 121	U 538	Tracheal agenesis	ORPHA:3346	Longfonds
P 121	U 541	Pulmonary non-tuberculous mycobacterial infection	ORPHA:411703	Longfonds
P 121	U 542	Pleural empyema	ORPHA:449266	Longfonds
P 121	U 543	Middle East respiratory syndrome	ORPHA:576074	Longfonds
P 121	U 544	Middle East respiratory syndrome	ORPHA:576074	Longfonds
P 121	U 545	Alpha-1-antitrypsin deficiency	ORPHA:60	Longfonds
P 121	U 547	Idiopathic bronchiectasis	ORPHA:60033	Longfonds
P 121	U 548	Idiopathic bronchiectasis	ORPHA:60033	Longfonds
P 121	U 549	Bronchopulmonary dysplasia	ORPHA:70589	Longfonds
P 121	U 550	Bronchopulmonary dysplasia	ORPHA:70589	Longfonds
P 121	U 551	Bronchopulmonary dysplasia	ORPHA:70589	Longfonds
P 121	U 552	Congenital tracheomalacia	ORPHA:95430	Longfonds
P 121	U 553	Congenital tracheomalacia	ORPHA:95430	Longfonds
P 121	U 554	Rare allergic respiratory disease	ORPHA:98052	Longfonds
P 121	U 555	Pulmonary agenesis	ORPHA:984	Longfonds
P 204	U 190	Thymic tumor	ORPHA:100100	Longkankervereniging
P 204	U 191	Thymic tumor	ORPHA:100100	Longkankervereniging
P 204	U 192	Small cell lung cancer	ORPHA:70573	Longkankervereniging
P 204	U 193	Small cell lung cancer	ORPHA:70573	Longkankervereniging
P 204	U 194	Rare respiratory tumor	ORPHA:98060	Longkankervereniging
P 204	U 195	Thymoma	ORPHA:99867	Longkankervereniging
P 105	U 641	Non-syndromic intestinal malformation	ORPHA:108967	Maag Lever Darm ST
P 105	U 642	Atresia of small intestine	ORPHA:1201	Maag Lever Darm ST
P 105	U 643	Microvillus inclusion disease	ORPHA:2290	Maag Lever Darm ST
P 105	U 644	Gastroschisis	ORPHA:2368	Maag Lever Darm ST
P 105	U 645	Gastroschisis	ORPHA:2368	Maag Lever Darm ST
P 105	U 646	Familial visceral myopathy	ORPHA:2604	Maag Lever Darm ST
P 105	U 647	Celiac artery compression syndrome	ORPHA:293208	Maag Lever Darm ST

P 105	U 648	Chronic intestinal failure	ORPHA:294422	Maag Lever Darm ST
P 105	U 649	Caudal regression sequence	ORPHA:3027	Maag Lever Darm ST
P 105	U 650	Inherited digestive cancer-predisposing syndrome	ORPHA:425003	Maag Lever Darm ST
P 105	U 651	Idiopathic gastroparesis	ORPHA:558411	Maag Lever Darm ST
P 105	U 652	Omphalocele	ORPHA:660	Maag Lever Darm ST
P 105	U 653	Radiation proctitis	ORPHA:70475	Maag Lever Darm ST
P 105	U 654	Intractable diarrhea of infancy	ORPHA:73014	Maag Lever Darm ST
P 105	U 655	Intractable diarrhea of infancy	ORPHA:73014	Maag Lever Darm ST
P 105	U 656	Intestinal malformation	ORPHA:97945	Maag Lever Darm ST
P 146	U 362	Marshall-Smith syndrome	ORPHA:561	Marshall Smith Syndroom Research Foundation
P 146	U 363	Overgrowth syndrome	ORPHA:93460	Marshall Smith Syndroom Research Foundation
P 83	U 896	Non-histaminic angioedema	ORPHA:658	Mastocytosevereniging NL
P 83	U 897	Cutaneous mastocytosis	ORPHA:66646	Mastocytosevereniging NL
P 83	U 898	Mast cell sarcoma	ORPHA:66661	Mastocytosevereniging NL
P 83	U 899	Diffuse cutaneous mastocytosis	ORPHA:79456	Mastocytosevereniging NL
P 83	U 900	Mastocytosis	ORPHA:98292	Mastocytosevereniging NL
P 83	U 901	Mastocytosis	ORPHA:98292	Mastocytosevereniging NL
P 83	U 902	Indolent systemic mastocytosis	ORPHA:98848	Mastocytosevereniging NL
P 83	U 903	Systemic mastocytosis with associated hematologic neoplasm	ORPHA:98849	Mastocytosevereniging NL
P 83	U 904	Aggressive systemic mastocytosis	ORPHA:98850	Mastocytosevereniging NL
P 83	U 905	Mast cell leukemia	ORPHA:98851	Mastocytosevereniging NL
P 125	U 490	Rare retinal disorder	ORPHA:519315	MD - Macula Vereniging
P 125	U 491	Rare retinal disorder	ORPHA:519315	MD - Macula Vereniging
P 125	U 492	Inherited retinal disorder	ORPHA:71862	MD - Macula Vereniging
P 125	U 493	Inherited retinal disorder	ORPHA:71862	MD - Macula Vereniging
P 125	U 494	Inherited retinal disorder	ORPHA:71862	MD - Macula Vereniging
P 215	U 169	Postpartum Psychose	ORPHA:443173	Me Mam
P 78	U 1007	Myeloproliferative neoplasm	ORPHA:98274	MPN ST
P 78	U 1008	Myeloproliferative neoplasm	ORPHA:98274	MPN ST
P 21	U 1651	Multiple sclerosis variant	ORPHA:228145	MS Vereniging NL
P 21	U 1652	Pediatric Multiple Sclerosis	ORPHA:477738	MS Vereniging NL
P 21	U 1967	Neuromyelitis optica spectrum disorder	ORPHA:71211	MS Vereniging NL
P 21	U 1968	Neuromyelitis optica spectrum disorder	ORPHA:71211	MS Vereniging NL
P 21	U 1969	Acute Disseminated Encephalomyelitis	ORPHA:83597	MS Vereniging NL
P 21	U 1971	Rare neuroinflammatory or neuroimmunological disease	ORPHA:182064	MS Vereniging NL
P 159	U 304	Rare genetic respiratory disease	ORPHA:156610	NCFS - NLse Cystic Fibrosis ST
P 159	U 307	Cystic fibrosis	ORPHA:586	NCFS - NLse Cystic Fibrosis ST
P 159	U 308	Cystic fibrosis	ORPHA:586	NCFS - NLse Cystic Fibrosis ST
P 159	U 309	Cystic fibrosis	ORPHA:586	NCFS - NLse Cystic Fibrosis ST
P 159	U 310	Cystic Fibrosis	ORPHA:586	NCFS - NLse Cystic Fibrosis ST
P 159	U 311	Cystic Fibrosis	ORPHA:586	NCFS - NLse Cystic Fibrosis ST
P 166	U 265	Rett Syndrome	ORPHA:778	Nederlandse Rett Syndroom Vereniging
P 265	U 93	genetic obesity	ORPHA:77828	Nederlandse ST Over Gewicht
P 151	U 349	Kabuki syndrome	ORPHA:2322	Netwerk Kabuki Syndroom
P 141	U 375	Rare nevus	ORPHA:294057	Nevus Netwerk
P 141	U 376	Large congenital melanocytic nevus	ORPHA:626	Nevus Netwerk

P 18	U 1706	Rare eye tumor	ORPHA:101950	NFK patiëntenplatform zeldzame kankers
P 18	U 1707	primary intraocular lymphoma	ORPHA:279904	NFK patiëntenplatform zeldzame kankers
P 18	U 1708	Rare tumor of gallbladder and extrahepatic biliary tract	ORPHA:306633	NFK patiëntenplatform zeldzame kankers
P 18	U 1709	Rare tumor of gallbladder and extrahepatic biliary tract	ORPHA:306633	NFK patiëntenplatform zeldzame kankers
P 18	U 1710	Rare tumor of liver and intrahepatic biliary tract	ORPHA:306636	NFK patiëntenplatform zeldzame kankers
P 18	U 1711	Uveal melanoma	ORPHA:39044	NFK patiëntenplatform zeldzame kankers
P 18	U 1712	Uveal melanoma	ORPHA:39044	NFK patiëntenplatform zeldzame kankers
P 18	U 1713	Malignant tumor of penis	ORPHA:398043	NFK patiëntenplatform zeldzame kankers
P 18	U 1714	Epithelial tumor of anal canal	ORPHA:424010	NFK patiëntenplatform zeldzame kankers
P 18	U 1715	Carcinoma of the anal canal	ORPHA:424013	NFK patiëntenplatform zeldzame kankers
P 18	U 1716	Primary cutaneous lymphoma	ORPHA:542	NFK patiëntenplatform zeldzame kankers
P 18	U 1717	Cholangiocarcinoma	ORPHA:70567	NFK patiëntenplatform zeldzame kankers
P 18	U 1718	Cholangiocarcinoma	ORPHA:70567	NFK patiëntenplatform zeldzame kankers
P 18	U 1719	Rare urinary tract tumor	ORPHA:98058	NFK patiëntenplatform zeldzame kankers
P 18	U 1720	Rare urinary tract tumour	ORPHA:98058	NFK patiëntenplatform zeldzame kankers
P 30	U 1620	Polymalformative genetic syndrome with increased risk of developing cancer	ORPHA:183422	NFVN - Neurofibromatosevereniging NL
P 30	U 1621	Genetic pigmentation anomaly of the skin	ORPHA:183463	NFVN - Neurofibromatosevereniging NL
P 30	U 1622	Benign peripheral nerve sheath tumor	ORPHA:252131	NFVN - Neurofibromatosevereniging NL
P 30	U 1623	Benign schwannoma	ORPHA:252164	NFVN - Neurofibromatosevereniging NL
P 30	U 1624	Benign schwannoma	ORPHA:252164	NFVN - Neurofibromatosevereniging NL
P 30	U 1625	Vestibular schwannoma	ORPHA:252175	NFVN - Neurofibromatosevereniging NL
P 30	U 1626	Vestibular schwannoma	ORPHA:252175	NFVN - Neurofibromatosevereniging NL
P 30	U 1627	Rasopathies	ORPHA:536391	NFVN - Neurofibromatosevereniging NL
P 30	U 1628	Rasopathies	ORPHA:536391	NFVN - Neurofibromatosevereniging NL
P 30	U 1629	Neurofibromatosis type 1	ORPHA:636	NFVN - Neurofibromatosevereniging NL
P 30	U 1630	Neurofibromatosis type 1	ORPHA:636	NFVN - Neurofibromatosevereniging NL
P 30	U 1631	Neurofibromatosis type 1	ORPHA:636	NFVN - Neurofibromatosevereniging NL
P 30	U 1632	Neurofibromatosis type 2	ORPHA:637	NFVN - Neurofibromatosevereniging NL
P 30	U 1633	Neurofibromatosis type 2	ORPHA:637	NFVN - Neurofibromatosevereniging NL
P 30	U 1634	Rare skin tumor or hamartoma	ORPHA:79386	NFVN - Neurofibromatosevereniging NL
P 30	U 1635	Schwannomatoses	ORPHA:93921	NFVN - Neurofibromatosevereniging NL
P 65	U 1124	Pituitary deficiency	ORPHA:101957	NHS - NLse Hypofyse ST
P 65	U 1125	Pituitary deficiency	ORPHA:101957	NHS - NLse Hypofyse ST
P 65	U 1126	Central diabetes insipidus	ORPHA:178029	NHS - NLse Hypofyse ST
P 65	U 1127	Rare hypothalamic or pituitary disease	ORPHA:181384	NHS - NLse Hypofyse ST
P 65	U 1128	Rare hypothalamic or pituitary disease	ORPHA:181384	NHS - NLse Hypofyse ST
P 65	U 1129	Rare hypothalamic or pituitary disease	ORPHA:181384	NHS - NLse Hypofyse ST
P 65	U 1130	Rare hypothalamic or pituitary disease	ORPHA:181384	NHS - NLse Hypofyse ST
P 65	U 1131	Rare hypothalamic or pituitary disease	ORPHA:181384	NHS - NLse Hypofyse ST
P 65	U 1132	Isolated congenital hypogonadotropic hypogonadism	ORPHA:238666	NHS - NLse Hypofyse ST
P 65	U 1133	Prolactinoma	ORPHA:2965	NHS - NLse Hypofyse ST
P 65	U 1134	Pituitary tumour	ORPHA:304055	NHS - NLse Hypofyse ST
P 65	U 1135	Functioning pituitary adenoma	ORPHA:314753	NHS - NLse Hypofyse ST
P 65	U 1136	Craniopharyngioma	ORPHA:54595	NHS - NLse Hypofyse ST
P 65	U 1137	Rare Endocrine Growth Disease	ORPHA:90692	NHS - NLse Hypofyse ST
P 65	U 1138	Acquired pituitary hormone deficiency	ORPHA:95502	NHS - NLse Hypofyse ST

P 65	U 1139	Rare endocrine disease	ORPHA:97978	NHS - NLse Hypofyse ST
P 65	U 1140	Pituitary adenoma	ORPHA:99408	NHS - NLse Hypofyse ST
P 142	U 368	Congenital primary lymphedema without systemic or visceral involvement	ORPHA:2416	NL Net
P 142	U 369	Late-onset primary lymphedema without systemic or visceral involvement	ORPHA:289825	NL Net
P 142	U 370	Primary lymphedema without systemic or visceral involvement	ORPHA:568041	NL Net
P 142	U 371	Primary lymphedema with systemic or visceral involvement	ORPHA:568044	NL Net
P 142	U 372	Disorder with multisystemic involvement and primary lymphedema	ORPHA:568047	NL Net
P 142	U 373	Primary Lymphedema	ORPHA:77240	NL Net
P 142	U 374	genetic obesity	ORPHA:77828	NL Net
P 27	U 1645	Lyme disease	ORPHA:91546	NI Vereniging Lyme Patiënten
P 117	U 595	Rare genetic male infertility	ORPHA:399980	NLse Klinefelter Vereniging
P 117	U 596	Urogenital tract malformation	ORPHA:83001	NLse Klinefelter Vereniging
P 117	U 597	Urogenital tract malformation	ORPHA:83001	NLse Klinefelter Vereniging
P 117	U 598	Disorders of sex development	ORPHA:90771	NLse Klinefelter Vereniging
P 117	U 599	Disorders of sex development	ORPHA:90771	NLse Klinefelter Vereniging
P 117	U 600	Rare male infertility	ORPHA:98048	NLse Klinefelter Vereniging
P 118	U 587	Meningococcal meningitis	ORPHA:33475	Nlse Meningitis Stichting
P 118	U 588	Arbovirus fever	ORPHA:344	Nlse Meningitis Stichting
P 118	U 589	Arbovirus fever	ORPHA:344	Nlse Meningitis Stichting
P 118	U 590	IgG4-related pachymeningitis	ORPHA:449427	Nlse Meningitis Stichting
P 118	U 591	Tuberculous meningitis	ORPHA:499004	Nlse Meningitis Stichting
P 118	U 592	Leptospirosis	ORPHA:509	Nlse Meningitis Stichting
P 118	U 593	Pneumococcal meningitis	ORPHA:55655	Nlse Meningitis Stichting
P 118	U 594	Infectious encephalitis	ORPHA:98252	Nlse Meningitis Stichting
P 118	U 1281	Susac syndrome	ORPHA:838	Nlse Meningitis Stichting
P 118	U 1898	Limbic encephalitis	ORPHA:163892	Nlse Meningitis Stichting
P 118	U 1903	Limbic encephalitis with NMDA receptor antibodies	ORPHA:217253	Nlse Meningitis Stichting
P 118	U 1913	Postinfectious encephalitis	ORPHA:98253	Nlse Meningitis Stichting
P 118	U 1973	Paraneoplastic neurologic syndrome	ORPHA:36388	Nlse Meningitis Stichting
P 118	U 1974	Autoimmune neurological channelopathy	ORPHA:98750	Nlse Meningitis Stichting
P 118	U 1975	Stiff person spectrum disorder	ORPHA:3198	Nlse Meningitis Stichting
P 118	U 1976	Autoimmune encephalopathy with parasomnia and obstructive sleep apnea	ORPHA:420789	Nlse Meningitis Stichting
P 26	U 1646	Rare pervasive developmental disorder	ORPHA:168778	NLse Vereniging voor Autisme
P 26	U 1647	Autism spectrum disorder due to AUTS2 deficiency	ORPHA:352490	NLse Vereniging voor Autisme
P 19	U 1653	Rare vascular liver disease	ORPHA:101938	NLV - NLse Leverpatiënten Vereniging
P 19	U 1654	Rare hepatic and biliary tract tumor	ORPHA:101943	NLV - NLse Leverpatiënten Vereniging
P 19	U 1655	Rare hepatic and biliary tract tumor	ORPHA:101943	NLV - NLse Leverpatiënten Vereniging
P 19	U 1656	Rare hepatic and biliary tract tumor	ORPHA:101943	NLV - NLse Leverpatiënten Vereniging
P 19	U 1657	Primary Sclerosing Cholangitis	ORPHA:171	NLV - NLse Leverpatiënten Vereniging
P 19	U 1658	Primary sclerosing cholangitis	ORPHA:171	NLV - NLse Leverpatiënten Vereniging
P 19	U 1659	Primary sclerosing cholangitis	ORPHA:171	NLV - NLse Leverpatiënten Vereniging
P 19	U 1660	Progressive Familial Intrahepatic Cholestasis	ORPHA:172	NLV - NLse Leverpatiënten Vereniging
P 19	U 1661	Progressive familial intrahepatic cholestasis	ORPHA:172	NLV - NLse Leverpatiënten Vereniging
P 19	U 1662	Primary Biliary Cholangitis	ORPHA:186	NLV - NLse Leverpatiënten Vereniging
P 19	U 1663	Primary biliary cholangitis	ORPHA:186	NLV - NLse Leverpatiënten Vereniging
P 19	U 1664	Crigler-Najjar syndrome	ORPHA:205	NLV - NLse Leverpatiënten Vereniging

P 19	U 1665	Crigler-Najjar syndrome	ORPHA:205	NLV - NLse Leverpatiënten Vereniging
P 19	U 1666	Adult hepatocellular carcinoma	ORPHA:210159	NLV - NLse Leverpatiënten Vereniging
P 19	U 1667	Autoimmune hepatitis	ORPHA:2137	NLV - NLse Leverpatiënten Vereniging
P 19	U 1668	Autoimmune hepatitis	ORPHA:2137	NLV - NLse Leverpatiënten Vereniging
P 19	U 1669	Acute fatty liver of pregnancy	ORPHA:243367	NLV - NLse Leverpatiënten Vereniging
P 19	U 1670	FTH1-related iron overload	ORPHA:247790	NLV - NLse Leverpatiënten Vereniging
P 19	U 1671	Response to antiviral treatment in hepatitis C	ORPHA:284102	NLV - NLse Leverpatiënten Vereniging
P 19	U 1672	Isolated polycystic liver disease	ORPHA:2924	NLV - NLse Leverpatiënten Vereniging
P 19	U 1673	Isolated biliary atresia	ORPHA:30391	NLV - NLse Leverpatiënten Vereniging
P 19	U 1674	Rare tumor of gallbladder and extrahepatic biliary tract	ORPHA:306633	NLV - NLse Leverpatiënten Vereniging
P 19	U 1675	Rare tumor of gallbladder and extrahepatic biliary tract	ORPHA:306633	NLV - NLse Leverpatiënten Vereniging
P 19	U 1676	Rare tumor of liver and intrahepatic biliary tract	ORPHA:306636	NLV - NLse Leverpatiënten Vereniging
P 19	U 1677	Fulminant viral hepatitis	ORPHA:35063	NLV - NLse Leverpatiënten Vereniging
P 19	U 1678	GSD due to liver glycogen phosphorylase deficiency	ORPHA:369	NLV - NLse Leverpatiënten Vereniging
P 19	U 1679	IgG4-related sclerosing cholangitis	ORPHA:447764	NLV - NLse Leverpatiënten Vereniging
P 19	U 1680	Choledochal cyst	ORPHA:480501	NLV - NLse Leverpatiënten Vereniging
P 19	U 1681	Alagille syndrome	ORPHA:52	NLV - NLse Leverpatiënten Vereniging
P 19	U 1682	Alagille syndrome	ORPHA:52	NLV - NLse Leverpatiënten Vereniging
P 19	U 1683	Growth delay-intellectual disability-hepatopathy syndrome	ORPHA:541423	NLV - NLse Leverpatiënten Vereniging
P 19	U 1684	Hepatocellular adenoma	ORPHA:54272	NLV - NLse Leverpatiënten Vereniging
P 19	U 1685	Lymphoplasmacytic inflammatory pseudotumor of the liver	ORPHA:555437	NLV - NLse Leverpatiënten Vereniging
P 19	U 1686	Carcinoma of gallbladder and extrahepatic biliary tract	ORPHA:56044	NLV - NLse Leverpatiënten Vereniging
P 19	U 1687	Carcinoma of gallbladder and extrahepatic biliary tract	ORPHA:56044	NLV - NLse Leverpatiënten Vereniging
P 19	U 1688	Primary biliary cholangitis/primary sclerosing cholangitis and autoimmune hepatitis overlap	ORPHA:562639	NLV - NLse Leverpatiënten Vereniging
P 19	U 1689	Parental nutrition associated cholestasis	ORPHA:567983	NLV - NLse Leverpatiënten Vereniging
P 19	U 1690	Alpha-1-antitrypsin deficiency	ORPHA:60	NLV - NLse Leverpatiënten Vereniging
P 19	U 1691	Benign Recurrent Intrahepatic Cholestasis	ORPHA:65682	NLV - NLse Leverpatiënten Vereniging
P 19	U 1692	Benign recurrent intrahepatic cholestasis	ORPHA:65682	NLV - NLse Leverpatiënten Vereniging
P 19	U 1693	Intrahepatic cholestasis of pregnancy	ORPHA:69665	NLV - NLse Leverpatiënten Vereniging
P 19	U 1694	Intrahepatic cholestasis of pregnancy	ORPHA:69665	NLV - NLse Leverpatiënten Vereniging
P 19	U 1695	Cholangiocarcinoma	ORPHA:70567	NLV - NLse Leverpatiënten Vereniging
P 19	U 1696	Cholangiocarcinoma	ORPHA:70567	NLV - NLse Leverpatiënten Vereniging
P 19	U 1697	Porphyria	ORPHA:738	NLV - NLse Leverpatiënten Vereniging
P 19	U 1698	Tyrosinemia type 1	ORPHA:882	NLV - NLse Leverpatiënten Vereniging
P 19	U 1699	Hepatocellular carcinoma	ORPHA:88673	NLV - NLse Leverpatiënten Vereniging
P 19	U 1700	Hepatocellular carcinoma	ORPHA:88673	NLV - NLse Leverpatiënten Vereniging
P 19	U 1702	Acute liver failure	ORPHA:90062	NLV - NLse Leverpatiënten Vereniging
P 19	U 1704	Wilson disease	ORPHA:905	NLV - NLse Leverpatiënten Vereniging
P 19	U 1705	Klatskin tumor	ORPHA:99978	NLV - NLse Leverpatiënten Vereniging
P 64	U 1141	Adrenal/paraganglial tumor	ORPHA:100091	NVACP - Bijniervereniging
P 64	U 1142	Adrenal/paraganglial tumor	ORPHA:100091	NVACP - Bijniervereniging
P 64	U 1143	Rare adrenal disease	ORPHA:101954	NVACP - Bijniervereniging
P 64	U 1144	Rare adrenal disease	ORPHA:101954	NVACP - Bijniervereniging
P 64	U 1145	Rare adrenal disease	ORPHA:101954	NVACP - Bijniervereniging
P 64	U 1146	Primary adrenal insufficiency	ORPHA:101958	NVACP - Bijniervereniging
P 64	U 1147	Carney complex	ORPHA:1359	NVACP - Bijniervereniging

P 64	U 1148	Adrenocortical carcinoma	ORPHA:1501	NVACP - Bijniervereniging
P 64	U 1149	Adrenocortical carcinoma	ORPHA:1501	NVACP - Bijniervereniging
P 64	U 1150	Adrenocortical carcinoma	ORPHA:1501	NVACP - Bijniervereniging
P 64	U 1151	Adrenogenital syndrome	ORPHA:181412	NVACP - Bijniervereniging
P 64	U 1152	Rare primary hyperaldosteronism	ORPHA:181415	NVACP - Bijniervereniging
P 64	U 1153	Rare primary hyperaldosteronism	ORPHA:181415	NVACP - Bijniervereniging
P 64	U 1154	Familial hyperaldosteronism	ORPHA:235936	NVACP - Bijniervereniging
P 64	U 1155	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	NVACP - Bijniervereniging
P 64	U 1156	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	NVACP - Bijniervereniging
P 64	U 1157	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	NVACP - Bijniervereniging
P 64	U 1158	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	NVACP - Bijniervereniging
P 64	U 1159	Multiple paragangliomas associated with polycythemia	ORPHA:324299	NVACP - Bijniervereniging
P 64	U 1160	Congenital adrenal hyperplasia	ORPHA:418	NVACP - Bijniervereniging
P 64	U 1161	Congenital adrenal hyperplasia	Orpha:418	NVACP - Bijniervereniging
P 64	U 1162	Cushing syndrome	ORPHA:553	NVACP - Bijniervereniging
P 64	U 1163	Pheochromocytoma-paraganglioma	ORPHA:573163	NVACP - Bijniervereniging
P 64	U 1164	Non-functioning paraganglioma	ORPHA:94080	NVACP - Bijniervereniging
P 64	U 1165	Non-functioning paraganglioma	ORPHA:94080	NVACP - Bijniervereniging
P 64	U 1166	Rare endocrine disease	ORPHA:97978	NVACP - Bijniervereniging
P 194	U 208	Growth hormone insensitivity syndrome	ORPHA:181393	NVGG - NLse Ver. voor Groeihormoondeficiëntie en Groeihormoonbehandeling
P 194	U 209	Non-acquired isolated growth hormone deficiency	ORPHA:631	NVGG - NLse Ver. voor Groeihormoondeficiëntie en Groeihormoonbehandeling
P 194	U 210	Rare Endocrine Growth Disease	ORPHA:90692	NVGG - NLse Ver. voor Groeihormoondeficiëntie en Groeihormoonbehandeling
P 74	U 1034	Rare hemorrhagic disorder	ORPHA:248308	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1035	Rare hemorrhagic disorder	ORPHA:248308	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1036	Rare hemorrhagic disorder	ORPHA:248308	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1037	Rare hemorrhagic disorder due to a coagulation factors defect	ORPHA:248315	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1038	Rare hemorrhagic disorder due to a coagulation factors defect	ORPHA:248315	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1039	Rare hemorrhagic disorder due to a coagulation factors defect	ORPHA:248315	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1040	Rare hemorrhagic disorder due to a coagulation factors defect	ORPHA:248315	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1041	Rare hemorrhagic disorder due to a platelet anomaly	ORPHA:248326	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1042	Rare hemorrhagic disorder due to a platelet anomaly	ORPHA:248326	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1043	Rare hemorrhagic disorder due to a platelet anomaly	ORPHA:248326	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1044	Rare hemorrhagic disorder due to a platelet anomaly	ORPHA:248326	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1045	Congenital amegakaryocytic thrombocytopenia	ORPHA:3319	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1046	Hemophilia	ORPHA:448	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1047	Hemophilia	ORPHA:448	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1048	Hemophilia	ORPHA:448	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1049	Hemophilia	ORPHA:448	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1050	Hemophilia	ORPHA:448	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1051	Hemophilia	ORPHA:448	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1052	Rare hemorrhagic disorder due to a constitutional platelet anomaly	ORPHA:71202	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1053	Von Willebrand disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1054	Von Willebrand disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1055	Von Willebrand Disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1056	Von Willebrand disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1057	Von Willebrand disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten

P 74	U 1058	Von Willebrand disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1059	Von Willebrand Disease	ORPHA:903	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1060	Rare coagulation disorder	ORPHA:98429	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1061	Rare coagulation disorder	ORPHA:98429	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1062	Hemophilia A	ORPHA:98878	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 74	U 1063	Hemophilia B	ORPHA:98879	NVHP- NLse Vereniging van Hemofilie-Patiënten
P 128	U 459	Systemic autoimmune disease	ORPHA:182228	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 460	Systemic autoimmune disease	ORPHA:182228	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 461	Diffuse cutaneous systemic sclerosis	ORPHA:220393	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 462	Limited cutaneous systemic sclerosis	ORPHA:220402	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 463	Overlapping connective tissue disease	ORPHA:251312	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 464	Systemic lupus erythematosus	ORPHA:536	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 465	Systemic lupus erythematosus	ORPHA:536	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 466	Systemic lupus erythematosus	ORPHA:536	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 467	Systemic lupus erythematosus	ORPHA:536	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 468	Undifferentiated connective tissue syndrome	ORPHA:90002	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 469	Localized scleroderma	ORPHA:90289	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 470	Systemic sclerosis	ORPHA:90291	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 471	Systemic sclerosis	ORPHA:90291	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 472	Systemic sclerosis	ORPHA:90291	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 473	Systemic sclerosis	ORPHA:90291	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 474	Glomerular disease	ORPHA:93548	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 475	Glomerular disease	ORPHA:93548	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 476	Pediatric systemic lupus erythematosus	ORPHA:93552	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 477	Pediatric systemic lupus erythematosus	ORPHA:93552	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 478	Pediatric Systemic Lupus Erythematosus	ORPHA:93552	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 479	Pediatric systemic lupus erythematosus	ORPHA:93552	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1958	Secondary interstitial lung disease specific to adulthood associated with a systemic disease	ORPHA:264745	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1959	Interstitial lung disease	ORPHA:182095	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1960	Interstitial lung disease	ORPHA:182095	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1961	Interstitial lung disease	ORPHA:182095	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1962	Interstitial lung disease	ORPHA:182095	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1962	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1963	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 128	U 1964	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	NVLE - Nationale vereniging voor lupus, APS, sclerodermie en MCTD
P 210	U 171	Narcolepsy type 1	ORPHA:2073	NVN - Narcolepsie Vereniging NL
P 210	U 172	Narcolepsy type 1	ORPHA:2073	NVN - Narcolepsie Vereniging NL
P 210	U 173	Idiopathic hypersomnia	ORPHA:33208	NVN - Narcolepsie Vereniging NL
P 210	U 174	Idiopathic Hypersomnia	ORPHA:33208	NVN - Narcolepsie Vereniging NL
P 210	U 175	Rare sleep disorder	ORPHA:68354	NVN - Narcolepsie Vereniging NL
P 210	U 176	Narcolepsy type 2	ORPHA:83465	NVN - Narcolepsie Vereniging NL
P 210	U 177	Narcolepsy Type 2	ORPHA:83465	NVN - Narcolepsie Vereniging NL
P 31	U 1561	Non-syndromic urogenital tract malformation of male and female	ORPHA:182124	NVN - Nierpatiënten Vereniging NL
P 31	U 1562	Diffuse cutaneous systemic sclerosis	ORPHA:220393	NVN - Nierpatiënten Vereniging NL
P 31	U 1563	Limited cutaneous systemic sclerosis	ORPHA:220402	NVN - Nierpatiënten Vereniging NL
P 31	U 1564	Limited systemic sclerosis	ORPHA:220407	NVN - Nierpatiënten Vereniging NL

P 31	U 1565	Exstrophy-epispadias complex	ORPHA:322	NVN - Nierpatiënten Vereniging NL
P 31	U 1566	Takayasu arteritis	ORPHA:3287	NVN - Nierpatiënten Vereniging NL
P 31	U 1567	C3 glomerulonephritis	ORPHA:329931	NVN - Nierpatiënten Vereniging NL
P 31	U 1568	Autosomal dominant tubulointerstitial kidney disease	ORPHA:34149	NVN - Nierpatiënten Vereniging NL
P 31	U 1569	Idiopathic nephrotic syndrome	ORPHA:357502	NVN - Nierpatiënten Vereniging NL
P 31	U 1570	Idiopathic Nephrotic Syndrome	ORPHA:357502	NVN - Nierpatiënten Vereniging NL
P 31	U 1571	Fetal lower urinary tract obstruction	ORPHA:435365	NVN - Nierpatiënten Vereniging NL
P 31	U 1572	Pseudohypoparathyroidism with Albright hereditary osteodystrophy	ORPHA:457059	NVN - Nierpatiënten Vereniging NL
P 31	U 1573	Rare disorder potentially indicated for kidney transplant	ORPHA:506213	NVN - Nierpatiënten Vereniging NL
P 31	U 1574	Rare disorder potentially indicated for kidney transplant	ORPHA:506213	NVN - Nierpatiënten Vereniging NL
P 31	U 1575	Systemic lupus erythematosus	ORPHA:536	NVN - Nierpatiënten Vereniging NL
P 31	U 1576	Systemic lupus erythematosus	ORPHA:536	NVN - Nierpatiënten Vereniging NL
P 31	U 1577	Systemic lupus erythematosus	ORPHA:536	NVN - Nierpatiënten Vereniging NL
P 31	U 1578	Systemic lupus erythematosus	ORPHA:536	NVN - Nierpatiënten Vereniging NL
P 31	U 1579	Hemolytic uremic syndrome	ORPHA:544458	NVN - Nierpatiënten Vereniging NL
P 31	U 1580	Genetic nephrotic syndrome	ORPHA:564127	NVN - Nierpatiënten Vereniging NL
P 31	U 1581	Disorder with multisystemic involvement and glomerulopathy	ORPHA:567562	NVN - Nierpatiënten Vereniging NL
P 31	U 1582	Nephrotic Syndrome without extrarenal manifestations	ORPHA:567564	NVN - Nierpatiënten Vereniging NL
P 31	U 1583	Alport syndrome	ORPHA:63	NVN - Nierpatiënten Vereniging NL
P 31	U 1584	Nephronophthisis	ORPHA:655	NVN - Nierpatiënten Vereniging NL
P 31	U 1585	Microscopic polyangiitis	ORPHA:727	NVN - Nierpatiënten Vereniging NL
P 31	U 1586	Autosomal dominant polycystic kidney disease	ORPHA:730	NVN - Nierpatiënten Vereniging NL
P 31	U 1587	Autosomal dominant polycystic kidney disease	ORPHA:730	NVN - Nierpatiënten Vereniging NL
P 31	U 1588	Autosomal recessive polycystic kidney disease	ORPHA:731	NVN - Nierpatiënten Vereniging NL
P 31	U 1589	Urogenital tract malformation	ORPHA:83001	NVN - Nierpatiënten Vereniging NL
P 31	U 1590	Urogenital tract malformation	ORPHA:83001	NVN - Nierpatiënten Vereniging NL
P 31	U 1591	Granulomatosis with polyangiitis	ORPHA:900	NVN - Nierpatiënten Vereniging NL
P 31	U 1592	Systemic sclerosis	ORPHA:90291	NVN - Nierpatiënten Vereniging NL
P 31	U 1593	Systemic sclerosis	ORPHA:90291	NVN - Nierpatiënten Vereniging NL
P 31	U 1594	Systemic sclerosis	ORPHA:90291	NVN - Nierpatiënten Vereniging NL
P 31	U 1595	Posterior urethral valve	ORPHA:93110	NVN - Nierpatiënten Vereniging NL
P 31	U 1596	Renal or urinary tract malformation	ORPHA:93545	NVN - Nierpatiënten Vereniging NL
P 31	U 1597	Renal or urinary tract malformation	ORPHA:93545	NVN - Nierpatiënten Vereniging NL
P 31	U 1598	Renal or urinary tract malformation	ORPHA:93545	NVN - Nierpatiënten Vereniging NL
P 31	U 1599	Renal or urinary tract malformation	ORPHA:93545	NVN - Nierpatiënten Vereniging NL
P 31	U 1600	Non-syndromic renal or urinary tract malformation	ORPHA:93546	NVN - Nierpatiënten Vereniging NL
P 31	U 1601	Glomerular disease	ORPHA:93548	NVN - Nierpatiënten Vereniging NL
P 31	U 1602	Glomerular disease	ORPHA:93548	NVN - Nierpatiënten Vereniging NL
P 31	U 1603	Pediatric systemic lupus erythematosus	ORPHA:93552	NVN - Nierpatiënten Vereniging NL
P 31	U 1604	Pediatric systemic lupus erythematosus	ORPHA:93552	NVN - Nierpatiënten Vereniging NL
P 31	U 1605	Pediatric Systemic Lupus Erythematosus	ORPHA:93552	NVN - Nierpatiënten Vereniging NL
P 31	U 1606	Pediatric systemic lupus erythematosus	ORPHA:93552	NVN - Nierpatiënten Vereniging NL
P 31	U 1607	Thrombotic microangiopathy	ORPHA:93573	NVN - Nierpatiënten Vereniging NL
P 31	U 1608	Thrombotic microangiopathy	ORPHA:93573	NVN - Nierpatiënten Vereniging NL
P 31	U 1609	Familial cystic renal disease	ORPHA:93587	NVN - Nierpatiënten Vereniging NL
P 31	U 1610	Familial cystic renal disease	ORPHA:93587	NVN - Nierpatiënten Vereniging NL

P 31	U 1611	Familial cystic renal disease	ORPHA:93587	NVN - Nierpatiënten Vereniging NL
P 31	U 1612	familial cystic renal disease	ORPHA:93587	NVN - Nierpatiënten Vereniging NL
P 31	U 1613	Nephropathy secondary to a storage or other metabolic disease	ORPHA:93593	NVN - Nierpatiënten Vereniging NL
P 31	U 1614	Rare renal tubular disease	ORPHA:93603	NVN - Nierpatiënten Vereniging NL
P 31	U 1615	Rare renal tubular disease	ORPHA:93603	NVN - Nierpatiënten Vereniging NL
P 31	U 1616	Rare renal tubular disease	ORPHA:93603	NVN - Nierpatiënten Vereniging NL
P 31	U 1617	rare renal tubular disease	ORPHA:93603	NVN - Nierpatiënten Vereniging NL
P 31	U 1618	Cloacal exstrophy	ORPHA:93929	NVN - Nierpatiënten Vereniging NL
P 31	U 1619	Rare genetic renal disease	ORPHA:98056	NVN - Nierpatiënten Vereniging NL
P 29	U 1636	Sporadic pheochromocytoma/secretory paraganglioma	ORPHA:276621	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1637	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1638	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1639	Hereditary pheochromocytoma-paraganglioma	ORPHA:29072	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1640	Multiple paragangliomas associated with polycythemia	ORPHA:324299	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1642	Pheochromocytoma-paraganglioma	ORPHA:573163	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1643	Non-functioning paraganglioma	ORPHA:94080	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1644	Non-functioning paraganglioma	ORPHA:94080	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1847	Adrenal/paraganglial tumor	ORPHA:100091	NVPG - Nlse Vereniging voor Patiënten met Paragangliomen
P 29	U 1848	Rare adrenal disease	ORPHA:101954	NVPG - Nlse Vereniging voor Patiënten met Paragangliomen
P 29	U 1849	Tumor of endocrine glands	ORPHA:182130	NVPG - NLse Vereniging voor Patiënten met Paragangliomen
P 29	U 1849	Rare endocrine disease	ORPHA:97978	NVPG - Nlse Vereniging voor Patiënten met Paragangliomen
P 22	U 1648	Primary Sjögren syndrome	ORPHA:289390	NVSP - Nationale Vereniging Sjögrenpatiënten
P 22	U 1649	Primary Sjögren Syndrome	ORPHA:289390	NVSP - Nationale Vereniging Sjögrenpatiënten
P 22	U 1650	Sjögren-Larsson syndrome	ORPHA:816	NVSP - Nationale Vereniging Sjögrenpatiënten
P 172	U 224	Primary bone dysplasia	ORPHA:364526	NVVR - Nederlandse Vereniging van Rugpatiënten 'de Wervelkolom'
P 172	U 225	Primary Bone Dysplasia	ORPHA:364526	NVVR - Nederlandse Vereniging van Rugpatiënten 'de Wervelkolom'
P 172	U 226	X-linked osteoporosis with fractures	ORPHA:391330	NVVR - Nederlandse Vereniging van Rugpatiënten 'de Wervelkolom'
P 172	U 227	Primary bone dysplasia with decreased bone density	ORPHA:93446	NVVR - Nederlandse Vereniging van Rugpatiënten 'de Wervelkolom'
P 100	U 669	Hereditary hyperferritinemia-cataract syndrome	ORPHA:163	Oogvereniging
P 100	U 670	Endophthalmitis	ORPHA:199323	Oogvereniging
P 100	U 671	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	ORPHA:247691	Oogvereniging
P 100	U 672	Intermediate uveitis	ORPHA:279914	Oogvereniging
P 100	U 673	anterior uveitis	ORPHA:280886	Oogvereniging
P 100	U 674	posterior uveitis	ORPHA:280892	Oogvereniging
P 100	U 675	panuveitis	ORPHA:280898	Oogvereniging
P 100	U 676	Central serous chorioretinopathy	ORPHA:443079	Oogvereniging
P 100	U 677	Cerebral visual impairment	ORPHA:447788	Oogvereniging
P 100	U 679	IgG4-related ophthalmic disease	ORPHA:449563	Oogvereniging
P 100	U 680	Euthyroid Graves orbitopathy	ORPHA:466682	Oogvereniging
P 100	U 681	Chronic relapsing inflammatory optic neuropathy	ORPHA:499085	Oogvereniging
P 100	U 682	Infective keratitis	ORPHA:519278	Oogvereniging
P 100	U 683	Isolated chorioretinal dystrophy	ORPHA:519300	Oogvereniging
P 100	U 684	Rare choroidal disorder	ORPHA:519309	Oogvereniging
P 100	U 687	Neuromyelitis optica spectrum disorder	ORPHA:71211	Oogvereniging
P 100	U 688	Neuromyelitis optica spectrum disorder	ORPHA:71211	Oogvereniging
P 100	U 689	Inherited retinal disorder	ORPHA:71862	Oogvereniging

P 100	U 690	Inherited retinal disorder	ORPHA:71862	Oogvereniging
P 100	U 691	Inherited retinal disorder	ORPHA:71862	Oogvereniging
P 100	U 692	Retinoblastoma	ORPHA:790	Oogvereniging
P 100	U 693	Usher syndrome	ORPHA:886	Oogvereniging
P 100	U 694	Retinopathy of prematurity	ORPHA:90050	Oogvereniging
P 100	U 695	Rare lens disease	ORPHA:98639	Oogvereniging
P 100	U 696	Color vision disease	ORPHA:98658	Oogvereniging
P 100	U 697	Autosomal recessive isolated optic atrophy	ORPHA:98676	Oogvereniging
P 100	U 698	Oculocutaneous or ocular albinism	ORPHA:98706	Oogvereniging
P 100	U 699	Uveitis	ORPHA:98715	Oogvereniging
P 100	U 1970	Acute Disseminated Encephalomyelitis	ORPHA:83597	Oogvereniging
P 100	U 1972	Rare neuroinflammatory or neuroimmunological disease	ORPHA:182064	Oogvereniging
P 100	U 1977a	Susac syndrome	ORPHA:838	Oogvereniging
P 100	U 685	Rare retinal disorder	ORPHA:519315	Oogvereniging - Patiëntengroep Retina
P 100	U 686	Rare retinal disorder	ORPHA:519315	Oogvereniging - Patiëntengroep Retina
P 73	U 1064	Sickle cell anemia	ORPHA:232	OSCAR Nederland
P 73	U 1065	Alpha-thalassemia and related diseases	ORPHA:275745	OSCAR Nederland
P 73	U 1066	Alpha-thalassemia and related diseases	ORPHA:275745	OSCAR Nederland
P 73	U 1067	Beta-thalassemia and related diseases	ORPHA:275749	OSCAR Nederland
P 73	U 1068	Beta-thalassemia and related diseases	ORPHA:275749	OSCAR Nederland
P 73	U 1069	Sickle cell disease and related diseases	ORPHA:275752	OSCAR Nederland
P 73	U 1070	Sickle cell disease and related diseases	ORPHA:275752	OSCAR Nederland
P 73	U 1071	Hemoglobinopathy	ORPHA:68364	OSCAR Nederland
P 73	U 1072	Hemoglobinopathy	ORPHA:68364	OSCAR Nederland
P 73	U 1073	Hemoglobinopathy	ORPHA:68364	OSCAR Nederland
P 73	U 1074	Hemoglobinopathy	ORPHA:68364	OSCAR Nederland
P 73	U 1846	Hydrops fetalis	ORPHA:1041	OSCAR Nederland
P 35	U 1520	classical progressive supranuclear palsy	ORPHA:240071	Parkinson Vereniging NL
P 35	U 1521	Off-periods in Parkinson disease not responding to oral treatment	ORPHA:391655	Parkinson Vereniging NL
P 35	U 1522	Rare parkinsonian disorders	ORPHA:68402	Parkinson Vereniging NL
P 35	U 1873	corticobasal syndrome	ORPHA:454887	Parkinson Vereniging NL
P 127	U 480	Aortic arch defects	ORPHA:1132	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 481	Brugada syndrome	ORPHA:130	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 482	Rare syndrome with cardiac malformations	ORPHA:156532	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 483	Rare vascular malformation of major vessels	ORPHA:458844	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 484	Rare Vascular Malformation of major vessels	ORPHA:458844	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 485	Genetic cardiac malformation	ORPHA:477805	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 486	Rare congenital non-syndromic heart malformation	ORPHA:88991	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 487	Rare congenital non-syndromic heart malformation	ORPHA:88991	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 488	Rare congenital non-syndromic heart malformation	ORPHA:88991	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 489	Arterial duct anomaly	ORPHA:95485	Patiëntenvereniging Aangeboren Hartafwijkingen
P 127	U 1869	Eisenmenger syndrome	ORPHA:97214	Patiëntenvereniging Aangeboren Hartafwijkingen
P 300	U 20	Porphyria	ORPHA:738	Patiëntenvereniging Acute Porfyrie
P 246	U 120	Fibrous dysplasia of bone	ORPHA:249	Patiëntenvereniging Fibreuze Dysplasie
P 246	U 1914	Primary bone dysplasia with disorganized development of skeletal components	ORPHA:93450	Patiëntenvereniging Fibreuze Dysplasie
P 246	U 1915	Primary bone dysplasia	ORPHA:364526	Patiëntenvereniging Fibreuze Dysplasie

P 246	U 1916	Primary Bone Dysplasia	ORPHA:364526	Patiëntenvereniging Fibreuze Dysplasie
P 169	U 228	Choanal atresia	ORPHA:137914	Patiëntenvereniging Hoofd-Hals
P 169	U 229	Nasolacrimalduct cyst	ORPHA:141083	Patiëntenvereniging Hoofd-Hals
P 169	U 230	Nasal dermoid cyste	ORPHA:141103	Patiëntenvereniging Hoofd-Hals
P 169	U 231	Nasopharyngeal teratoma	ORPHA:141107	Patiëntenvereniging Hoofd-Hals
P 169	U 232	Nasal encephalocele	ORPHA:141118	Patiëntenvereniging Hoofd-Hals
P 169	U 233	Congenital subglottic stenosis	ORPHA:141121	Patiëntenvereniging Hoofd-Hals
P 169	U 234	Nasal dorsum fistulae	ORPHA:141219	Patiëntenvereniging Hoofd-Hals
P 169	U 235	Nasopharyngeal carcinoma	ORPHA:150	Patiëntenvereniging Hoofd-Hals
P 169	U 236	Nasopharyngeal carcinoma	ORPHA:150	Patiëntenvereniging Hoofd-Hals
P 169	U 237	Nose & Cavum anomaly	ORPHA:156246	Patiëntenvereniging Hoofd-Hals
P 169	U 238	Larynx anomaly	ORPHA:156249	Patiëntenvereniging Hoofd-Hals
P 169	U 239	Larynx anomaly	ORPHA:156249	Patiëntenvereniging Hoofd-Hals
P 169	U 240	Larynx anomaly	ORPHA:156249	Patiëntenvereniging Hoofd-Hals
P 169	U 241	Tracheal anomaly	ORPHA:156252	Patiëntenvereniging Hoofd-Hals
P 169	U 242	Tracheal anomaly	ORPHA:156252	Patiëntenvereniging Hoofd-Hals
P 169	U 243	Isolated congenital nasal pyriform aperture stenosis	ORPHA:162516	Patiëntenvereniging Hoofd-Hals
P 169	U 244	Congenital laryngomalacia	ORPHA:2373	Patiëntenvereniging Hoofd-Hals
P 169	U 245	Juvenile nasopharyngeal angiofibroma	ORPHA:289596	Patiëntenvereniging Hoofd-Hals
P 169	U 246	Juvenile nasopharyngeal angiofibroma	ORPHA:289596	Patiëntenvereniging Hoofd-Hals
P 169	U 247	Rare head and neck tumor	ORPHA:290849	Patiëntenvereniging Hoofd-Hals
P 169	U 248	Rare head and neck tumor	ORPHA:290849	Patiëntenvereniging Hoofd-Hals
P 169	U 249	Rare head and neck tumor	ORPHA:290849	Patiëntenvereniging Hoofd-Hals
P 169	U 250	Rare head and neck tumor	ORPHA:290849	Patiëntenvereniging Hoofd-Hals
P 169	U 251	Rare head and neck tumor	ORPHA:290849	Patiëntenvereniging Hoofd-Hals
P 169	U 252	Rare head and neck tumor	ORPHA:290849	Patiëntenvereniging Hoofd-Hals
P 169	U 253	Ameloblastoma	ORPHA:314419	Patiëntenvereniging Hoofd-Hals
P 169	U 254	Squamous cell carcinoma of the hypopharynx	ORPHA:494547	Patiëntenvereniging Hoofd-Hals
P 169	U 255	Squamous cell carcinoma of the larynx	ORPHA:494550	Patiëntenvereniging Hoofd-Hals
P 169	U 256	Squamous cell carcinoma of the larynx	ORPHA:494550	Patiëntenvereniging Hoofd-Hals
P 169	U 257	Squamous cell carcinoma of the nasal cavity and paranasal sinuses	ORPHA:500464	Patiëntenvereniging Hoofd-Hals
P 169	U 258	Squamous cell carcinoma of the oropharynx	ORPHA:500478	Patiëntenvereniging Hoofd-Hals
P 169	U 259	Squamous cell carcinoma of the oropharynx	ORPHA:500478	Patiëntenvereniging Hoofd-Hals
P 169	U 260	Squamous cell carcinoma of oral cavity and lip	ORPHA:502369	Patiëntenvereniging Hoofd-Hals
P 169	U 261	Squamous cell carcinoma of oral cavity and lip	ORPHA:502369	Patiëntenvereniging Hoofd-Hals
P 169	U 1874	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Hoofd-Hals
P 169	U 1875	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Hoofd-Hals
P 169	U 1876	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Hoofd-Hals
P 169	U 1877	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Hoofd-Hals
P 169	U 1878	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Hoofd-Hals
P 169	U 1932	Osteosarcoma	ORPHA:668	Patiëntenvereniging Hoofd-Hals
P 169	U 1933	Osteosarcoma	ORPHA:668	Patiëntenvereniging Hoofd-Hals
P 255	U 98	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Speekselklierkanker
P 255	U 99	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Speekselklierkanker
P 255	U 100	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Speekselklierkanker
P 255	U 101	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Speekselklierkanker

P 255	U 102	Rare tumor of salivary glands	ORPHA:276142	Patiëntenvereniging Speekselklierkanker
P 293	U 25	X-linked hypophosphatemia	ORPHA:89936	Patiëntenvereniging voor XLH
P 37	U 1516	Disorder of phenylalanine metabolism	ORPHA:284814	PKU Vereniging NL
P 37	U 1517	Phenylketonuria	ORPHA:716	PKU Vereniging NL
P 37	U 1518	phenylketonuria	ORPHA:716	PKU Vereniging NL
P 37	U 1519	disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167	PKU Vereniging NL
P 122	U 518	Congenital diaphragmatic hernia	ORPHA:2140	Platform Congenitale Hernia Diafragmatica
P 122	U 519	Diaphragmatic or abdominal wall malformation	ORPHA:98043	Platform Congenitale Hernia Diafragmatica
P 57	U 1293	Rare urticaria	ORPHA:79384	PP Urticaria
P 236	U 129	Temple syndrome	ORPHA:254516	Prader-Willi ST
P 236	U 130	Prader-Willi-like syndrome	ORPHA:398073	Prader-Willi ST
P 236	U 131	Prader-Willi syndrome	ORPHA:739	Prader-Willi ST
P 236	U 132	Prader-Willi syndrome	ORPHA:739	Prader-Willi ST
P 236	U 133	Rare Endocrine Growth Disease	ORPHA:90692	Prader-Willi ST
P 307	U 1	Psoriasis-related juvenile idiopathic arthritis	ORPHA:85436	Psoriasispatiënten NL
P 262	U 94	Undifferentiated connective tissue syndrome	ORPHA:90002	Reuma NL
P 262	U 95	Rare systemic or rheumatologic disease	ORPHA:98023	Reuma NL
P 305	U 6	Neurogenic thoracic outlet syndrome	ORPHA:100073	RSI-vereniging
P 305	U 7	Arterial thoracic outlet syndrome	ORPHA:357107	RSI-vereniging
P 305	U 8	Venous thoracic outlet syndrome	ORPHA:357131	RSI-vereniging
P 40	U 1505	Interstitial lung disease	ORPHA:182095	Sarcoïdose NL
P 40	U 1506	Interstitial lung disease	ORPHA:182095	Sarcoïdose NL
P 40	U 1507	Interstitial lung disease	ORPHA:182095	Sarcoïdose NL
P 40	U 1508	Interstitial lung disease	ORPHA:182095	Sarcoïdose NL
P 40	U 1509	Interstitial lung disease specific to adulthood	ORPHA:264735	Sarcoïdose NL
P 40	U 1510	Secondary interstitial lung disease specific to adulthood associated with a systemic disease	ORPHA:264745	Sarcoïdose NL
P 40	U 1511	Sarcoidosis	ORPHA:797	Sarcoïdose NL
P 40	U 1512	Sarcoidosis	ORPHA:797	Sarcoïdose NL
P 40	U 1513	Sarcoidosis	ORPHA:797	Sarcoïdose NL
P 72	U 1075	Combined T and B cell immunodeficiency	ORPHA:101972	SAS - ST voor Afweerstoornissen
P 72	U 1076	Immunodeficiency predominantly affecting antibody production	ORPHA:101977	SAS - ST voor Afweerstoornissen
P 72	U 1077	Quantitative and/or qualitative congenital phagocyte defect	ORPHA:101985	SAS - ST voor Afweerstoornissen
P 72	U 1078	Immunodeficiency due to a complement cascade protein anomaly	ORPHA:101992	SAS - ST voor Afweerstoornissen
P 72	U 1079	Primary immunodeficiency	ORPHA:101997	SAS - ST voor Afweerstoornissen
P 72	U 1080	Primary Immunodeficiency	ORPHA:101997	SAS - ST voor Afweerstoornissen
P 72	U 1081	Primary immunodeficiency	ORPHA:101997	SAS - ST voor Afweerstoornissen
P 72	U 1082	Primary immunodeficiency	ORPHA:101997	SAS - ST voor Afweerstoornissen
P 72	U 1083	Primary immunodeficiency	ORPHA:101997	SAS - ST voor Afweerstoornissen
P 72	U 1084	Primary immunodeficiency	ORPHA:101997	SAS - ST voor Afweerstoornissen
P 72	U 1085	Aspergillosis	ORPHA:1163	SAS - ST voor Afweerstoornissen
P 72	U 1086	Allergic bronchopulmonary aspergillosis	ORPHA:1164	SAS - ST voor Afweerstoornissen
P 72	U 1087	Chronic mucocutaneous candidiasis	ORPHA:1334	SAS - ST voor Afweerstoornissen
P 72	U 1088	Common variable immunodeficiency	ORPHA:1572	SAS - ST voor Afweerstoornissen
P 72	U 1089	Rare mycosis	ORPHA:163591	SAS - ST voor Afweerstoornissen
P 72	U 1090	Primary immunodeficiency due to a defect in adaptive immunity	ORPHA:179006	SAS - ST voor Afweerstoornissen
P 72	U 1091	Primary immunodeficiency due to a defect in adaptive immunity	ORPHA:179006	SAS - ST voor Afweerstoornissen

P 72	U 1092	Severe combined immunodeficiency	ORPHA:183660	SAS - ST voor Afweerstoornissen
P 72	U 1093	ICF syndrome	ORPHA:2268	SAS - ST voor Afweerstoornissen
P 72	U 1094	IgG4-related diseases	ORPHA:284264	SAS - ST voor Afweerstoornissen
P 72	U 1095	IgG4-related diseases	ORPHA:284264	SAS - ST voor Afweerstoornissen
P 72	U 1096	Autosomal dominant Mendelian susceptibility to mycobacterial diseases due to partial	ORPHA:319581	SAS - ST voor Afweerstoornissen
P 72	U 1097	Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial	ORPHA:319589	SAS - ST voor Afweerstoornissen
P 72	U 1098	IgG4-related pachymeningitis	ORPHA:449427	SAS - ST voor Afweerstoornissen
P 72	U 1099	X-linked agammaglobulinemia	ORPHA:47	SAS - ST voor Afweerstoornissen
P 72	U 1100	Pyogenic Arthritis-pyoderma gangrenosum-acne syndrome	ORPHA:69126	SAS - ST voor Afweerstoornissen
P 72	U 1101	Mendelian susceptibility to mycobacterial diseases	ORPHA:748	SAS - ST voor Afweerstoornissen
P 72	U 1102	Complications after hematopoietic stem cell transplantation	ORPHA:90053	SAS - ST voor Afweerstoornissen
P 72	U 1977b	Constitutional neutropenia	ORPHA:101987	SAS - ST voor afweerstoornissen
P 286	U 62	Neural tube closure defect	ORPHA:268357	SBH NL
P 286	U 63	Neural tube closure defect	ORPHA:268357	SBH NL
P 286	U 64	Malformation of the neurenteric canal, spinal cord and column	ORPHA:268843	SBH NL
P 286	U 65	Isolated spina bifida	ORPHA:823	SBH NL
P 286	U 66	Isolated spina bifida	ORPHA:823	SBH NL
P 42	U 1504	Isolated Sternocostoclavicular Hyperostosis	ORPHA:178311	SCCH - NL ver. van patiënten met sternocosto clavulaire hyperostosis
P 287	U 52	Rare odontal or periodontal disorder	ORPHA:164001	Schisis NL
P 287	U 53	Rare odontal or periodontal disorder	ORPHA:164001	Schisis NL
P 287	U 54	Cleft lip with or without cleft palate	ORPHA:1991	Schisis NL
P 287	U 55	Cleft lip with or without cleft palate	ORPHA:1991	Schisis NL
P 287	U 56	Cleft lip with or without cleft palate	ORPHA:1991	Schisis NL
P 287	U 57	Cleft lip with or without cleft palate	ORPHA:1991	Schisis NL
P 287	U 58	Cleft palate	ORPHA:2014	Schisis NL
P 287	U 59	Cleft palate	ORPHA:2014	Schisis NL
P 287	U 60	Cleft palate	ORPHA:2014	Schisis NL
P 287	U 61	Cleft palate	ORPHA:2014	Schisis NL
P 287	U 1920	CHARGE syndrome	ORPHA:138	Schisis NL
P 287	U 1921	CHARGE syndrome	ORPHA:138	Schisis NL
P 287	U 1922	Rare disease with Pierre Robin Syndrome	ORPHA:138044	Schisis NL
P 287	U 1923	Rare disease with Pierre Robin Syndrome	ORPHA:138044	Schisis NL
P 287	U 1924	Oligodontia	ORPHA:99798	Schisis NL
P 287	U 1927	Macroglossia	ORPHA:156207	Schisis NL
P 287	U 1928	Hypoglossia/aglossia	ORPHA:156212	Schisis NL
P 183	U 213	Li-Fraumeni syndrome	ORPHA:524	SDK - ST Diagnose Kanker
P 71	U 1103	Rare chromosomal anomaly	ORPHA:68335	SDS - ST Down Syndroom
P 71	U 1104	Down syndrome	ORPHA:870	SDS - ST Down Syndroom
P 71	U 1105	Autosomal anomaly	ORPHA:98127	SDS - ST Down Syndroom
P 60	U 1213	Thyroid tumor	ORPHA:100087	SON - Schildklier Organisatie NL
P 60	U 1214	Thyroid tumor	ORPHA:100087	SON - Schildklier Organisatie NL
P 60	U 1215	Thyroid Tumor	ORPHA:100087	SON - Schildklier Organisatie NL
P 60	U 1216	Thyroid carcinoma	ORPHA:100088	SON - Schildklier Organisatie NL
P 60	U 1217	Thyroid carcinoma	ORPHA:100088	SON - Schildklier Organisatie NL
P 60	U 1218	Thyroid carcinoma	ORPHA:100088	SON - Schildklier Organisatie NL
P 60	U 1219	Thyroid carcinoma	ORPHA:100088	SON - Schildklier Organisatie NL

P 60	U 1220	Rare thyroid disease	ORPHA:101955	SON - Schildklier Organisatie NL
P 60	U 1221	Rare thyroid disease	ORPHA:101955	SON - Schildklier Organisatie NL
P 60	U 1222	Rare hyperparathyroidism	ORPHA:181408	SON - Schildklier Organisatie NL
P 60	U 1223	Euthyroid Graves orbitopathy	ORPHA:466682	SON - Schildklier Organisatie NL
P 60	U 1224	Rare parathyroid disease and phosphocalcic metabolism anomaly	ORPHA:68415	SON - Schildklier Organisatie NL
P 60	U 1225	Rare endocrine disease	ORPHA:97978	SON - Schildklier Organisatie NL
P 43	U 1418	Neurogenic thoracic outlet syndrome	ORPHA:100073	Spierziekten NL
P 43	U 1419	Arthrogryposis multiplex congenita	ORPHA:1037	Spierziekten NL
P 43	U 1420	Hereditary motor and sensory neuropathy	ORPHA:166	Spierziekten NL
P 43	U 1421	Acquired peripheral neuropathy	ORPHA:182086	Spierziekten NL
P 43	U 1422	Acquired peripheral neuropathy	ORPHA:182086	Spierziekten NL
P 43	U 1423	Genetic Neurodegenerative Diseases	ORPHA:183500	Spierziekten NL
P 43	U 1424	Genetic skeletal muscle disease	ORPHA:206634	Spierziekten NL
P 43	U 1425	Acquired skeletal muscle disease	ORPHA:206638	Spierziekten NL
P 43	U 1426	Progressive muscular dystrophy	ORPHA:206644	Spierziekten NL
P 43	U 1427	Myotonic dystrophy	ORPHA:206647	Spierziekten NL
P 43	U 1428	Myotonic dystrophy	ORPHA:206647	Spierziekten NL
P 43	U 1429	Non-dystrophic myopathy	ORPHA:206656	Spierziekten NL
P 43	U 1430	Bulbospinal muscular atrophy	ORPHA:206701	Spierziekten NL
P 43	U 1431	Chronic polyradiculoneuropathy	ORPHA:208978	Spierziekten NL
P 43	U 1432	Peripheral neuropathy associated with monoclonal gammopathy	ORPHA:209010	Spierziekten NL
P 43	U 1433	Peripheral neuropathy associated with monoclonal gammopathy	ORPHA:209010	Spierziekten NL
P 43	U 1434	Guillain-Barré syndrome	ORPHA:2103	Spierziekten NL
P 43	U 1435	Dermatomyositis	ORPHA:221	Spierziekten NL
P 43	U 1436	Duchenne and Becker muscular dystrophy	ORPHA:262	Spierziekten NL
P 43	U 1437	Duchenne and Becker muscular dystrophy	ORPHA:262	Spierziekten NL
P 43	U 1438	Duchenne and Becker muscular dystrophy	ORPHA:262	Spierziekten NL
P 43	U 1439	Limb Girdle Muscular Dystrophy	ORPHA:263	Spierziekten NL
P 43	U 1440	Multiple acyl-CoA dehydrogenase deficiency	ORPHA:26791	Spierziekten NL
P 43	U 1441	Facioscapulohumeral muscular dystrophy	ORPHA:269	Spierziekten NL
P 43	U 1442	Facioscapulohumeral muscular dystrophy	ORPHA:269	Spierziekten NL
P 43	U 1443	Oculopharyngeal muscular dystrophy	ORPHA:270	Spierziekten NL
P 43	U 1444	Oculopharyngeal muscular dystrophy	ORPHA:270	Spierziekten NL
P 43	U 1445	Neuralgic amyotrophy	ORPHA:2901	Spierziekten NL
P 43	U 1446	Neuralgic amyotrophy	ORPHA:2901	Spierziekten NL
P 43	U 1447	Chronic inflammatory demyelinating polyneuropathy	ORPHA:2932	Spierziekten NL
P 43	U 1448	Chronic inflammatory demyelinating polyneuropathy	ORPHA:2932	Spierziekten NL
P 43	U 1449	Postpoliomyelitis syndrome	ORPHA:2942	Spierziekten NL
P 43	U 1450	Sodium channelopathy-related small fiber neuropathy	ORPHA:306577	Spierziekten NL
P 43	U 1451	Stiff person spectrum disorder	ORPHA:3198	Spierziekten NL
P 43	U 1452	Juvenile idiopathic inflammatory myopathy	ORPHA:329888	Spierziekten NL
P 43	U 1453	Juvenile idiopathic inflammatory myopathy	ORPHA:329888	Spierziekten NL
P 43	U 1454	Paraneoplastic neurologic syndrome	ORPHA:36388	Spierziekten NL
P 43	U 1455	Multiple acyl-CoA dehydrogenase deficiency, severe neonatal type	ORPHA:394529	Spierziekten NL
P 43	U 1456	Multiple acyl-CoA dehydrogenase deficiency, mild type	ORPHA:394532	Spierziekten NL
P 43	U 1457	Rare hyperkinetic movement disorder	ORPHA:494457	Spierziekten NL

P 43	U 1458	Myasthenia gravis	ORPHA:589	Spierziekten NL
P 43	U 1459	Myasthenia gravis	ORPHA:589	Spierziekten NL
P 43	U 1460	Congenital myastenic syndrome	ORPHA:590	Spierziekten NL
P 43	U 1461	Inclusion body myositis	ORPHA:611	Spierziekten NL
P 43	U 1462	Multifocal motor neuropathy	ORPHA:641	Spierziekten NL
P 43	U 1463	Neuromuscular disease	ORPHA:68381	Spierziekten NL
P 43	U 1464	Neuromuscular disease	ORPHA:68381	Spierziekten NL
P 43	U 1465	Neuromuscular disease	ORPHA:68381	Spierziekten NL
P 43	U 1466	Neuromuscular disease	ORPHA:68381	Spierziekten NL
P 43	U 1467	Neuromuscular disease	ORPHA:68381	Spierziekten NL
P 43	U 1468	Neuromuscular disease	ORPHA:68381	Spierziekten NL
P 43	U 1469	Neurometabolic disease	ORPHA:68385	Spierziekten NL
P 43	U 1470	Hereditary spastic paraplegia	ORPHA:685	Spierziekten NL
P 43	U 1471	Hereditary spastic paraplegia	ORPHA:685	Spierziekten NL
P 43	U 1472	Hereditary spastic paraplegia	ORPHA:685	Spierziekten NL
P 43	U 1473	Proximal spinal muscular atrophy	ORPHA:70	Spierziekten NL
P 43	U 1474	Muscular channelopathy	ORPHA:71864	Spierziekten NL
P 43	U 1475	Polymyositis	ORPHA:732	Spierziekten NL
P 43	U 1476	Amyotrophic lateral sclerosis	ORPHA:803	Spierziekten NL
P 43	U 1477	Amyotrophic lateral sclerosis	ORPHA:803	Spierziekten NL
P 43	U 1478	Anti-synthetase syndrome	ORPHA:81	Spierziekten NL
P 43	U 1479	amyotrophic lateral sclerosis-parkinsonism-dementia complex	ORPHA:90020	Spierziekten NL
P 43	U 1480	Juvenile dermatomyositis	ORPHA:93672	Spierziekten NL
P 43	U 1481	Juvenile dermatomyositis	ORPHA:93672	Spierziekten NL
P 43	U 1482	Riboflavin transporter deficiency	ORPHA:97229	Spierziekten NL
P 43	U 1483	Congenital myopathy	ORPHA:97245	Spierziekten NL
P 43	U 1484	Rare cardiac diseases	ORPHA:97929	Spierziekten NL
P 43	U 1485	Central nervous system malformation	ORPHA:98044	Spierziekten NL
P 43	U 1486	Central nervous system malformation	ORPHA:98044	Spierziekten NL
P 43	U 1487	Skeletal muscle disease	ORPHA:98472	Spierziekten NL
P 43	U 1488	Skeletal muscle disease	ORPHA:98472	Spierziekten NL
P 43	U 1489	Skeletal muscle disease	ORPHA:98472	Spierziekten NL
P 43	U 1490	Muscular dystrophy	ORPHA:98473	Spierziekten NL
P 43	U 1491	Idiopathic inflammatory myopathy	ORPHA:98482	Spierziekten NL
P 43	U 1492	Idiopathic inflammatory myopathy	ORPHA:98482	Spierziekten NL
P 43	U 1493	Idiopathic inflammatory myopathy	ORPHA:98482	Spierziekten NL
P 43	U 1494	Neuromuscular junction disease	ORPHA:98491	Spierziekten NL
P 43	U 1495	Neuromuscular junction disease	ORPHA:98491	Spierziekten NL
P 43	U 1496	Acquired neuromuscular junction disease	ORPHA:98494	Spierziekten NL
P 43	U 1497	Genetic neuromuscular junction disease	ORPHA:98495	Spierziekten NL
P 43	U 1498	Rare Peripheral Neuropathy	ORPHA:98496	Spierziekten NL
P 43	U 1499	Rare peripheral neuropathy	ORPHA:98496	Spierziekten NL
P 43	U 1500	Genetic peripheral neuropathy	ORPHA:98497	Spierziekten NL
P 43	U 1501	Motor Neuron Disease	ORPHA:98503	Spierziekten NL
P 43	U 1502	Autoimmune neurological channelopathy	ORPHA:98750	Spierziekten NL
P 43	U 1503	Fetal akinesia deformation sequence	ORPHA:994	Spierziekten NL

P 43	U 1870	glycogen storage disease due to acid maltase deficiency	ORPHA:365	Spierziekten NL
P 43	U 1871	Mitochondrial disease	ORPHA:68380	Spierziekten NL
P 43	U 1872	Mitochondrial disease	ORPHA:68380	Spierziekten NL
P 43	U 1899	Rare neuroinflammatory or neuroimmunological disease	ORPHA:182064	Spierziekten NL
P 63	U 1167	Rare hepatic and biliary tract tumor	ORPHA:101943	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1168	Rare hepatic and biliary tract tumor	ORPHA:101943	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1169	Rare hepatic and biliary tract tumor	ORPHA:101943	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1170	Primary peritoneal tumor	ORPHA:168803	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1171	Rare gastroesophageal tumor	ORPHA:180821	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1172	Rare gastroesophageal tumor	ORPHA:180821	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1173	Rare gastroesophageal tumor	ORPHA:180821	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1174	Rare tumor of pancreas	ORPHA:180824	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1175	Rare tumor of pancreas	ORPHA:180824	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1176	Rare tumor of Pancreas	ORPHA:180824	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1177	High-grade dysplasia in patients with Barrett esophagus	ORPHA:231080	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1178	Hereditary diffuse gastric cancer	ORPHA:26106	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1179	Carcinoma of the ampulla of Vater	ORPHA:300557	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1180	Carcinoma of the ampulla of Vater	ORPHA:300557	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1181	Ampulla of Vater carcinoma	ORPHA:300557	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1182	Gastric adenocarcinoma and proximal polyposis of the stomach	ORPHA:314022	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1183	Gastric linitis plastica	ORPHA:36273	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1184	Hereditary gastric cancer	ORPHA:423776	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1185	Hereditary gastric cancer	ORPHA:423776	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1186	Epithelial tumor of the appendix	ORPHA:423982	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1187	Squamous cell carcinoma of the colon	ORPHA:423994	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1188	Gastrointestinal stromal tumor	ORPHA:44890	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1189	Gastrointestinal stromal tumor	ORPHA:44890	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1190	Gastrointestinal stromal tumor	ORPHA:44890	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1191	Gastrointestinal stromal tumor	ORPHA:44890	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1192	Gastro-intestinale stromale tumor	ORPHA:44890	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1193	Carcinoma of gallbladder and extrahepatic biliary tract	ORPHA:56044	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1194	Carcinoma of gallbladder and extrahepatic biliary tract	ORPHA:56044	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1195	Carcinoma of esophagus	ORPHA:70482	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1196	Hepatocellular carcinoma	ORPHA:88673	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1197	Hepatocellular carcinoma	ORPHA:88673	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 63	U 1198	Rare digestive tract tumor	ORPHA:98059	SPKS - ST voor Patiënten met Kanker aan het Spijsverteringskanaal
P 177	U 215	Non-syndromic central nervous system malformation	ORPHA:108989	SPV - Syringomyelie Patiënten Vereniging
P 177	U 216	Neural tube closure defect	ORPHA:268357	SPV - Syringomyelie Patiënten Vereniging
P 177	U 217	Neural tube closure defect	ORPHA:268357	SPV - Syringomyelie Patiënten Vereniging
P 177	U 218	Neural tube defect	ORPHA:3388	SPV - Syringomyelie Patiënten Vereniging
P 177	U 219	Central nervous system malformation	ORPHA:98044	SPV - Syringomyelie Patiënten Vereniging
P 177	U 220	Central nervous system malformation	ORPHA:98044	SPV - Syringomyelie Patiënten Vereniging
P 76	U 1009	Quantitative and/or qualitative congenital phagocyte defect	ORPHA:101985	SSSH - Shwachman Syndroom Support Holland
P 76	U 1010	Shwachman-Diamond syndrome	ORPHA:811	SSSH - Shwachman Syndroom Support Holland
P 76	U 1011	Shwachman-Diamond syndrome	ORPHA:811	SSSH - Shwachman Syndroom Support Holland
P 104	U 657	Rare acquired aplastic anemia	ORPHA:164823	ST AA & PNH

P 104	U 658	Rare acquired aplastic anemia	ORPHA:164823	ST AA & PNH
P 104	U 659	Aplastic Anemia	ORPHA:182040	ST AA & PNH
P 104	U 660	Paroxysmal nocturnal hemoglobinuria	ORPHA:447	ST AA & PNH
P 104	U 661	Rare constitutional aplastic anemia	ORPHA:68383	ST AA & PNH
P 104	U 662	Rare constitutional aplastic anemia	ORPHA:68383	ST AA & PNH
P 104	U 663	Congenital dyserythropoietic anemia	ORPHA:85	ST AA & PNH
P 104	U 664	Congenital dyserythropoietic anemia	ORPHA:85	ST AA & PNH
P 104	U 665	Ideopathic aplastic anemia	ORPHA:88	ST AA & PNH
P 104	U 666	Primary acquired red cell aplasia	ORPHA:98421	ST AA & PNH
P 295	U 23	Anterior cutaneous nerve entrapment syndrome	ORPHA:51890	ST ACNES
P 295	U 24	Anterior cutaneous nerve entrapment syndrome	ORPHA:51890	ST ACNES
P 138	U 377	ABeta amyloidosis, Dutch type	ORPHA:100006	ST Amyloidose NL
P 138	U 378	Hereditary ATTR amyloidosis	ORPHA:271861	ST Amyloidose NL
P 138	U 379	Primary systemic amyloidosis	ORPHA:314701	ST Amyloidose NL
P 138	U 380	Primary localized amyloidosis	ORPHA:314709	ST Amyloidose NL
P 138	U 381	Wild type ATTR amyloidosis	ORPHA:330001	ST Amyloidose NL
P 138	U 382	Hereditary amyloidosis	ORPHA:444116	ST Amyloidose NL
P 138	U 383	Amyloidosis	ORPHA:69	ST Amyloidose NL
P 138	U 384	AL amyloidosis	ORPHA:85443	ST Amyloidose NL
P 138	U 385	AA amyloidosis	ORPHA:85445	ST Amyloidose NL
P 138	U 386	ATTRV30M amyloidosis	ORPHA:85447	ST Amyloidose NL
P 46	U 1413	Non-Langerhans cell histiocytosis	ORPHA:157987	ST Celhistiose NL
P 46	U 1414	Hemophagocytic syndrome	ORPHA:158032	ST Celhistiose NL
P 46	U 1415	Hemophagocytic syndrome	ORPHA:158032	ST Celhistiose NL
P 46	U 1417	Langerhans cell histiocytosis	ORPHA:389	ST Celhistiose NL
P 306	U 2	Multiple congenital anomalies/dysmorphic syndrome without intellectual disability	ORPHA:102285	ST Complex Care United
P 306	U 3	Rare non-syndromic intellectual disability	ORPHA:101685	ST Complex Care United
P 306	U 4	Rare non-syndromic intellectual disability	ORPHA:101685	ST Complex Care United
P 306	U 5	Rare syndromic intellectual disability	ORPHA:102369	ST Complex Care United
P 304	U 14	Squamous cell carcinoma of the rectum	ORPHA:424002	ST Darmkanker
P 304	U 9	Rare tumor of intestine	ORPHA:104011	ST Darmkanker
P 304	U 10	Rare tumor of the intestine	ORPHA:104011	ST Darmkanker
P 304	U 11	Adenocarcinoma of the small intestine	ORPHA:104075	ST Darmkanker
P 304	U 12	Adenocarcinoma of the small intestine	ORPHA:104075	ST Darmkanker
P 304	U 13	Rare carcinoma of small intestine	ORPHA:423957	ST Darmkanker
P 304	U 15	Radiation proctitis	ORPHA:70475	ST Darmkanker
P 304	U 1881	Radiation proctitis	ORPHA:70475	ST Darmkanker
P 247	U 105	Progressive hemifacial atrophy	ORPHA:1214	ST Eigen Gezicht
P 247	U 106	Nasal glial heterotopia	ORPHA:141112	ST Eigen Gezicht
P 247	U 107	Hemifacial hyperplasia	ORPHA:141145	ST Eigen Gezicht
P 247	U 108	Cysts and fistulae of the face and oral cavity	ORPHA:155835	ST Eigen Gezicht
P 247	U 109	Otomandibular dysplasia	ORPHA:155896	ST Eigen Gezicht
P 247	U 110	Hypoglossia/aglossia	ORPHA:156212	ST Eigen Gezicht
P 247	U 111	Paralytic facial malformation	ORPHA:156224	ST Eigen Gezicht
P 247	U 112	Paralytic facial malformation	ORPHA:156224	ST Eigen Gezicht
P 247	U 113	Syndrome or malformation associated with head and neck malformations	ORPHA:156237	ST Eigen Gezicht

P 247	U 114	Syndrome or malformation associated with head and neck malformations	ORPHA:156237	ST Eigen Gezicht
P 247	U 115	Rare odontal or periodontal disorder	ORPHA:164001	ST Eigen Gezicht
P 247	U 116	Rare odontal or periodontal disorder	ORPHA:164001	ST Eigen Gezicht
P 247	U 117	Cleidocranial dysplasia and isolated cranial ossification defect	ORPHA:93451	ST Eigen Gezicht
P 247	U 118	Dysostosis with predominant craniofacial involvement	ORPHA:93453	ST Eigen Gezicht
P 247	U 119	Oligodontia	ORPHA:99798	ST Eigen Gezicht
P 247	U 1929	Cleidocranial dysplasia	ORPHA:1452	ST Eigen Gezicht
P 247	U 1930	Cleidocranial dysplasia	ORPHA:1452	ST Eigen Gezicht
P 247	U 1934	Microtia	ORPHA:83463	ST Eigen Gezicht
P 247	U 1935	Microtia	ORPHA:83463	ST Eigen Gezicht
P 249	U 103	Dilated cardiomyopathy	ORPHA:217604	ST Genetische Hartspierziekte PLN
P 249	U 104	Dilated cardiomyopathy	ORPHA:217604	ST Genetische Hartspierziekte PLN
P 313	U 1868	Rare intellectual disability	ORPHA:87277	ST Grin syndroom
P 81	U 905	Mast cell leukemia	ORPHA:98851	ST Hematon
P 81	U 956	Constitutional neutropenia	ORPHA:101987	ST Hematon
P 81	U 957	Myeloid hemopathy	ORPHA:171895	ST Hematon
P 81	U 958	Myeloid hemopathy	ORPHA:171895	ST Hematon
P 81	U 959	B-cell non-Hodgkin lymphoma	ORPHA:171915	ST Hematon
P 81	U 960	Dyskeratosis congenita	ORPHA:1775	ST Hematon
P 81	U 961	Dyskeratosis congenita	ORPHA:1775	ST Hematon
P 81	U 962	Lymphoma	ORPHA:223735	ST Hematon
P 81	U 963	POEMS syndrome	ORPHA:2905	ST Hematon
P 81	U 964	Multiple myeloma	ORPHA:29073	ST Hematon
P 81	U 965	Multiple myeloma	ORPHA:29073	ST Hematon
P 81	U 966	Multiple myeloma	ORPHA:29073	ST Hematon
P 81	U 967	Multiple myeloma	ORPHA:29073	ST Hematon
P 81	U 968	Multiple myeloma	ORPHA:29073	ST Hematon
P 81	U 969	Aggressive B-cell non-Hodgkin lymphoma	ORPHA:300846	ST Hematon
P 81	U 970	Waldenström macroglobulinemia	ORPHA:33226	ST Hematon
P 81	U 971	Graft versus host disease	ORPHA:39812	ST Hematon
P 81	U 972	Graft versus host disease	ORPHA:39812	ST Hematon
P 81	U 973	Graft versus Host Disease	ORPHA:39812	ST Hematon
P 81	U 974	Plasma cell leukemia	ORPHA:454714	ST Hematon
P 81	U 975	Primary central nervous system lymphoma	ORPHA:46135	ST Hematon
P 81	U 976	Acute lymphoblastic leukemia	ORPHA:513	ST Hematon
P 81	U 977	Acute lymphoblastic leukemia	ORPHA:513	ST Hematon
P 81	U 978	Acute lymphoblastic leukemia	ORPHA:513	ST Hematon
P 81	U 979	Acute myeloid leukemia	ORPHA:519	ST Hematon
P 81	U 980	Acute myeloid leukemia	ORPHA:519	ST Hematon
P 81	U 981	Acute Myeloid leukemia	ORPHA:519	ST Hematon
P 81	U 982	Acute myeloid leukemia	ORPHA:519	ST Hematon
P 81	U 983	Chronic myeloid leukemia	ORPHA:521	ST Hematon
P 81	U 984	Chronic myeloid leukemia	ORPHA:521	ST Hematon
P 81	U 985	MALT lymphoma	ORPHA:52417	ST Hematon
P 81	U 986	Myelodysplastic syndrome	ORPHA:52688	ST Hematon
P 81	U 987	Myelodysplastic syndrome	ORPHA:52688	ST Hematon

P 81	U 988	CAR T cell therapy-associated cytokine release syndrome	ORPHA:542323	ST Hematon
P 81	U 989	Diffuse large B-cell lymphoma	ORPHA:544	ST Hematon
P 81	U 990	Non Hodgkin Lymphoma	ORPHA:547	ST Hematon
P 81	U 991	B-cell chronic lymphocytic leukemia	ORPHA:67038	ST Hematon
P 81	U 992	B-cell chronic lymphocytic leukemia	ORPHA:67038	ST Hematon
P 81	U 993	Tumor of hematopoietic and lymphoid tissues	ORPHA:68347	ST Hematon
P 81	U 994	Primary myelofibrosis	ORPHA:824	ST Hematon
P 81	U 995	Primary Myelofibrosis	ORPHA:824	ST Hematon
P 81	U 996	Complications after hematopoietic stem cell transplantation	ORPHA:90053	ST Hematon
P 81	U 997	Plasma cell tumor	ORPHA:98282	ST Hematon
P 81	U 998	Hodgkin lymphoma	ORPHA:98293	ST Hematon
P 81	U 999	Chronic myelomonocytic leukemia	ORPHA:98823	ST Hematon
P 81	U 1000	Acute graft versus host disease	ORPHA:99920	ST Hematon
P 81	U 1001	Acute graft versus host disease	ORPHA:99920	ST Hematon
P 81	U 1002	Chronic graft versus host disease	ORPHA:99921	ST Hematon
P 81	U 1964	Primary cutaneous lymphoma	ORPHA:542	ST Hematon
P 81	U 1965	primary intraocular lymphoma	ORPHA:279904	ST Hematon
P 81	U 1966	Myeloproliferative neoplasm	ORPHA:98274	ST Hematon
P 81	U 1967	Myeloproliferative neoplasm	ORPHA:98274	ST Hematon
P 33	U 1524	Pinnae and external auditory canal anomaly	ORPHA:156243	ST Hoormij
P 33	U 1525	Middle ear anomaly	ORPHA:164004	ST Hoormij
P 33	U 1526	Middle ear anomaly	ORPHA:164004	ST Hoormij
P 33	U 1527	Middle ear anomaly	ORPHA:164004	ST Hoormij
P 33	U 1528	Idiopathic Bilateral Vestibulopathy	ORPHA:171684	ST Hoormij
P 33	U 1529	Cockayne Syndrome	ORPHA:191	ST Hoormij
P 33	U 1530	Tumor of cranial and spinal nerves	ORPHA:252057	ST Hoormij
P 33	U 1531	Benign schwannoma	ORPHA:252164	ST Hoormij
P 33	U 1532	Benign schwannoma	ORPHA:252164	ST Hoormij
P 33	U 1533	Vestibular schwannoma	ORPHA:252175	ST Hoormij
P 33	U 1534	Vestibular schwannoma	ORPHA:252175	ST Hoormij
P 33	U 1535	Rare head and neck tumor	ORPHA:290849	ST Hoormij
P 33	U 1536	Rare head and neck tumor	ORPHA:290849	ST Hoormij
P 33	U 1537	Rare head and neck tumor	ORPHA:290849	ST Hoormij
P 33	U 1538	Rare head and neck tumor	ORPHA:290849	ST Hoormij
P 33	U 1539	Rare head and neck tumor	ORPHA:290849	ST Hoormij
P 33	U 1540	Rare head and neck tumor	ORPHA:290849	ST Hoormij
P 33	U 1541	Cochleovestibular dysplasia	ORPHA:502305	ST Hoormij
P 33	U 1542	Cochlear nerve deficiency	ORPHA:502318	ST Hoormij
P 33	U 1543	Rare deafness	ORPHA:68361	ST Hoormij
P 33	U 1544	Rare deafness	ORPHA:68361	ST Hoormij
P 33	U 1545	Rare deafness	ORPHA:68361	ST Hoormij
P 33	U 1546	Rare otorhinolaryngological malformation	ORPHA:96333	ST Hoormij
P 33	U 1547	Rare otorhinolaryngologic tumor	ORPHA:98061	ST Hoormij
P 33	U 1548	Rare otorhinolaryngologic tumor	ORPHA:98061	ST Hoormij
P 311	U 1852	Lymphoma	ORPHA:223735	ST Huidlymfoom
P 311	U 1853	Primary cutaneous lymphoma	ORPHA:542	ST Huidlymfoom

P 189	U 211	Pantothenate kinase-associated neurodegeneration	ORPHA:157850	ST IJzersterk
P 189	U 212	Neurodegeneration with brain iron accumulation	ORPHA:385	ST IJzersterk
P 135	U 387	Periodic fever syndromes	ORPHA:101995	ST KAISZ
P 135	U 388	Behcet disease	ORPHA:117	ST KAISZ
P 135	U 389	Dermatomyositis	ORPHA:221	ST KAISZ
P 135	U 390	Autoinflammatory syndrome with immune deficiency	ORPHA:290839	ST KAISZ
P 135	U 391	Mevalonate kinase deficiency	ORPHA:309025	ST KAISZ
P 135	U 392	Autoinflammatory syndrome of childhood	ORPHA:319719	ST KAISZ
P 135	U 393	Autoinflammatory syndrome of childhood	ORPHA:319719	ST KAISZ
P 135	U 394	Chronic nonbacterial osteomyelitis/Chronic recurrent multifocal osteomyelitis	ORPHA:324964	ST KAISZ
P 135	U 395	Schnitzler syndrome	ORPHA:37748	ST KAISZ
P 135	U 414	Autoinflammatory syndrome	ORPHA:93665	ST KAISZ
P 135	U 415	Autoinflammatory syndrome	ORPHA:93665	ST KAISZ
P 160	U 298	Rare genetic respiratory disease	ORPHA:156610	ST LAM NL
P 160	U 299	Interstitial lung disease	ORPHA:182095	ST LAM NL
P 160	U 300	Interstitial lung disease	ORPHA:182095	ST LAM NL
P 160	U 301	Interstitial lung disease	ORPHA:182095	ST LAM NL
P 160	U 302	Interstitial lung disease	ORPHA:182095	ST LAM NL
P 160	U 303	Interstitial lung disease specific to adulthood	ORPHA:264735	ST LAM NL
P 93	U 748	Intestinal polyposis syndrome	ORPHA:104010	ST Lynch Polyposis
P 93	U 749	Intestinal polyposis syndrome	ORPHA:104010	ST Lynch Polyposis
P 93	U 750	Intestinal polyposis syndrome	ORPHA:104010	ST Lynch Polyposis
P 93	U 751	Intestinal polyposis syndrome	ORPHA:104010	ST Lynch Polyposis
P 93	U 752	Rare intestinal disease	ORPHA:117569	ST Lynch Polyposis
P 93	U 753	Rare intestinal disease	ORPHA:117569	ST Lynch Polyposis
P 93	U 754	Lynch syndrome	ORPHA:144	ST Lynch Polyposis
P 93	U 755	Lynch Syndrome	ORPHA:144	ST Lynch Polyposis
P 93	U 756	Lynch syndrome	ORPHA:144	ST Lynch Polyposis
P 93	U 757	Lynch Syndrome	ORPHA:144	ST Lynch Polyposis
P 93	U 758	Lynch syndroom	ORPHA:144	ST Lynch Polyposis
P 93	U 759	Lynch syndrome	ORPHA:144	ST Lynch Polyposis
P 93	U 760	Hereditary mixed polyposis syndrome	ORPHA:157794	ST Lynch Polyposis
P 93	U 761	Serrated polyposis syndrome	ORPHA:157798	ST Lynch Polyposis
P 93	U 762	Serrated polyposis syndrome	ORPHA:157798	ST Lynch Polyposis
P 93	U 763	Attenuated familial adenomatous polyposis	ORPHA:220460	ST Lynch Polyposis
P 93	U 764	Geattenueerde familiale adenomateuze polyposis	ORPHA:220460	ST Lynch Polyposis
P 93	U 765	Attenuated Familial adenomatous polyposis	ORPHA:220460	ST Lynch Polyposis
P 93	U 766	MUTYH-related attenuated familial adenomatous polyposis	ORPHA:247798	ST Lynch Polyposis
P 93	U 767	MUTYH-related attenuated familial adenomatous polyposis	ORPHA:247798	ST Lynch Polyposis
P 93	U 768	APC-related attenuated familial adenomatous polyposis	ORPHA:247806	ST Lynch Polyposis
P 93	U 769	Peutz-Jeghers syndrome	ORPHA:2869	ST Lynch Polyposis
P 93	U 770	Genetic intestinal polyposis	ORPHA:363314	ST Lynch Polyposis
P 93	U 771	Familial colorectal cancer type X	ORPHA:440437	ST Lynch Polyposis
P 93	U 772	Hereditary nonpolyposis colon cancer	ORPHA:443909	ST Lynch Polyposis
P 93	U 773	Familial adenomatous polyposis	ORPHA:733	ST Lynch Polyposis
P 93	U 774	Familial adenomatous polyposis	ORPHA:733	ST Lynch Polyposis

P 93	U 775	Familial adenomatous polyposis	ORPHA:733	ST Lynch Polyposis
P 93	U 776	Familial adenomatous polyposis	ORPHA:733	ST Lynch Polyposis
P 96	U 701	Malignant melanoma of the mucosa	ORPHA:168999	ST Melanoom
P 96	U 702	Malignant melanoma of the mucosa	ORPHA:168999	ST Melanoom
P 96	U 703	Melanoma and neural system tumor syndrome	ORPHA:252206	ST Melanoom
P 96	U 704	BAP1-related tumor predisposition syndrome	ORPHA:289539	ST Melanoom
P 96	U 705	MITF-related melanoma and renal cell carcinoma predisposition syndrome	ORPHA:293822	ST Melanoom
P 96	U 706	Uveal melanoma	ORPHA:39044	ST Melanoom
P 96	U 707	Uveal melanoma	ORPHA:39044	ST Melanoom
P 96	U 708	Familial atypical multiple mole melanoma syndrome	ORPHA:404560	ST Melanoom
P 96	U 709	Familial melanoma	ORPHA:618	ST Melanoom
P 96	U 1863	Rare skin tumor or hamartoma	ORPHA:79386	ST Melanoom
P 112	U 623	Non-syndromic urogenital tract malformation	ORPHA:165704	ST MRK Vrouwen
P 112	U 624	Urogenital tract malformation	ORPHA:83001	ST MRK Vrouwen
P 112	U 625	Urogenital tract malformation	ORPHA:83001	ST MRK Vrouwen
P 62	U 1199	Laryngeal neuroendocrine tumor	ORPHA:100083	ST NET-Groep
P 62	U 1200	Middle ear neuroendocrine tumor	ORPHA:100084	ST NET-Groep
P 62	U 1201	Gastroenteropancreatic neuroendocrine neoplasm	ORPHA:100092	ST NET-Groep
P 62	U 1202	Tumor of endocrine glands	ORPHA:182130	ST NET-Groep
P 62	U 1203	Cutaneous neuroendocrine carcinoma	ORPHA:79140	ST NET-Groep
P 62	U 1204	Cutaneous neuro-endocrine carcinoma	ORPHA:79140	ST NET-Groep
P 62	U 1205	Cutaneous neuroendocrine carcinoma	ORPHA:79140	ST NET-Groep
P 62	U 1206	Neuroendocrine neoplasm	ORPHA:877	ST NET-Groep
P 62	U 1207	Neuroendocrine neoplasm	ORPHA:877	ST NET-Groep
P 62	U 1208	Neuroendocrine neoplasm	ORPHA:877	ST NET-Groep
P 62	U 1209	Neuroendocrine neoplasm	ORPHA:877	ST NET-Groep
P 62	U 1210	Neuroendocrine neoplasm	ORPHA:877	ST NET-Groep
P 62	U 1211	Neuroendocrine neoplasm	ORPHA:877	ST NET-Groep
P 62	U 1862	Neuroendocrine neoplasms of the pancreas	ORPHA:506052	ST NET-groep
P 47	U 1408	Hypertrophic cardiomyopathy	ORPHA:217569	ST Noonan Syndroom
P 47	U 1409	Rasopathies	ORPHA:536391	ST Noonan Syndroom
P 47	U 1410	Rasopathies	ORPHA:536391	ST Noonan Syndroom
P 47	U 1411	Noonan syndrome	ORPHA:648	ST Noonan Syndroom
P 47	U 1412	Rare cardiac diseases	ORPHA:97929	ST Noonan Syndroom
P 88	U 841	Vulvar intraepithelial neoplasia	ORPHA:137583	ST Olijf
P 88	U 842	Vulvar intraepithelial neoplasia	ORPHA:137583	ST Olijf
P 88	U 843	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 844	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 845	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 846	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 847	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 848	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 849	Hereditary breast and ovarian cancer syndrome	ORPHA:145	ST Olijf
P 88	U 850	Rare uterine adnexal tumor	ORPHA:180220	ST Olijf
P 88	U 851	Malignant tumor of fallopian tubes	ORPHA:180242	ST Olijf
P 88	U 852	Rare vulvovaginal tumor	ORPHA:180312	ST Olijf

P 88	U 853	Rare vulvovaginal tumor	ORPHA:180312	ST Olijf
P 88	U 854	Borderline epithelial tumor of ovary	ORPHA:206473	ST Olijf
P 88	U 855	Ovarian Cancer	ORPHA:213500	ST Olijf
P 88	U 856	Ovarian cancer	ORPHA:213500	ST Olijf
P 88	U 857	Ovarian cancer	ORPHA:213500	ST Olijf
P 88	U 858	Familial ovarian cancer	ORPHA:213517	ST Olijf
P 88	U 859	Hereditary site-specific ovarian cancer syndrome	ORPHA:213524	ST Olijf
P 88	U 860	Hereditary site-specific ovarian cancer syndrome	ORPHA:213524	ST Olijf
P 88	U 861	Rare uterine cancer	ORPHA:213564	ST Olijf
P 88	U 862	Rare cancer of the Corpus Uteri	ORPHA:213569	ST Olijf
P 88	U 863	Rare cancer of corpus uteri	ORPHA:213569	ST Olijf
P 88	U 864	Endometrial stromal sarcoma	ORPHA:213711	ST Olijf
P 88	U 865	Undifferentiated carcinoma of the corpus uteri	ORPHA:213721	ST Olijf
P 88	U 866	Papillary carcinoma of the corpus uteri	ORPHA:213726	ST Olijf
P 88	U 867	Rare cancer of cervix uteri	ORPHA:213761	ST Olijf
P 88	U 868	Rare cancer of cervix uteri	ORPHA:213761	ST Olijf
P 88	U 869	Rare cancer of the cervix uteri	ORPHA:213761	ST Olijf
P 88	U 870	Rare cancer of cervix uteri	ORPHA:213761	ST Olijf
P 88	U 871	Squamous cell carcinoma of the cervix uteri	ORPHA:213767	ST Olijf
P 88	U 872	Adenocarcinoma of the cervix uteri	ORPHA:213772	ST Olijf
P 88	U 873	Gestational trophoblastic disease	ORPHA:254685	ST Olijf
P 88	U 874	Gestational trophoblastic disease	ORPHA:254685	ST Olijf
P 88	U 875	Malignant epithelial tumor of ovary	ORPHA:398934	ST Olijf
P 88	U 876	Malignant epithelial tumor of ovary	ORPHA:398934	ST Olijf
P 88	U 877	Malignant non-epithelial tumor of ovary	ORPHA:398940	ST Olijf
P 88	U 878	Vulvar carcinoma	ORPHA:494418	ST Olijf
P 88	U 879	Vulvar carcinoma	ORPHA:494418	ST Olijf
P 88	U 880	Vulvar squamous cell carcinoma	ORPHA:494448	ST Olijf
P 88	U 881	Vulvar squamous cell carcinoma	ORPHA:494448	ST Olijf
P 88	U 882	Rare gynecological tumor	ORPHA:98063	ST Olijf
P 88	U 883	Rare gynecological tumor	ORPHA:98063	ST Olijf
P 88	U 884	Rare gynecological tumor	ORPHA:98063	ST Olijf
P 88	U 885	Malignant Granulosaceltumor of ovary	ORPHA:99915	ST Olijf
P 220	U 138	Rare disorder due to toxic effects	ORPHA:108999	ST OPS
P 220	U 139	Rare parkinsonian syndrome due to intoxication	ORPHA:306679	ST OPS
P 220	U 140	Manganese poisoning	ORPHA:306682	ST OPS
P 220	U 141	Lead poisoning	ORPHA:330015	ST OPS
P 220	U 142	Mercury poisoning	ORPHA:330021	ST OPS
P 94	U 713	Chordoma	ORPHA:178	ST Patientenplatform Sarcomen
P 94	U 714	Chordoma	ORPHA:178	ST Patientenplatform Sarcomen
P 94	U 715	Chordoma	ORPHA:178	ST Patientenplatform Sarcomen
P 94	U 716	Bone sarcoma	ORPHA:223727	ST Patientenplatform Sarcomen
P 94	U 717	Bone sarcoma	ORPHA:223727	ST Patientenplatform Sarcomen
P 94	U 718	Bone sarcoma	ORPHA:223727	ST Patientenplatform Sarcomen
P 94	U 719	Bone Sarcoma	ORPHA:223727	ST Patientenplatform Sarcomen
P 94	U 721	Malignant peripheral nerve sheath tumor	ORPHA:3148	ST Patientenplatform Sarcomen

P 94	U 722	Skeletal Ewing sarcoma	ORPHA:319	ST Patiëntenplatform Sarcomen
P 94	U 723	Soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
P 94	U 724	soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
P 94	U 725	Soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
P 94	U 726	Soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
P 94	U 727	Soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
P 94	U 728	Soft tissue sarcoma	ORPHA:3394	ST Patiëntenplatform Sarcomen
P 94	U 729	Gastrointestinal stromal tumor	ORPHA:44890	ST Patiëntenplatform Sarcomen
P 94	U 730	Gastrointestinal stromal tumor	ORPHA:44890	ST Patiëntenplatform Sarcomen
P 94	U 731	Gastrointestinal stromal tumor	ORPHA:44890	ST Patiëntenplatform Sarcomen
P 94	U 732	Gastrointestinal stromal tumor	ORPHA:44890	ST Patiëntenplatform Sarcomen
P 94	U 733	Gastro-intestinale stromale tumor	ORPHA:44890	ST Patiëntenplatform Sarcomen
P 94	U 735	Chondrosarcoma	ORPHA:55880	ST Patiëntenplatform Sarcomen
P 94	U 736	Osteosarcoma	ORPHA:668	ST Patiëntenplatform Sarcomen
P 94	U 737	Osteosarcoma	ORPHA:668	ST Patiëntenplatform Sarcomen
P 94	U 738	Rare bone tumor	ORPHA:68411	ST Patiëntenplatform Sarcomen
P 94	U 739	Rare bone tumor	ORPHA:68411	ST Patiëntenplatform Sarcomen
P 94	U 740	Rare soft tissue tumor	ORPHA:71209	ST Patiëntenplatform Sarcomen
P 94	U 741	Rare soft tissue tumor	ORPHA:71209	ST Patiëntenplatform Sarcomen
P 94	U 742	Rare soft tissue tumor	ORPHA:71209	ST Patiëntenplatform Sarcomen
P 94	U 743	Rare soft tissue tumor	ORPHA:71209	ST Patiëntenplatform Sarcomen
P 94	U 745	Desmoid tumor	ORPHA:873	ST Patiëntenplatform Sarcomen
P 94	U 746	Desmoid tumor	ORPHA:873	ST Patiëntenplatform Sarcomen
P 94	U 747	Rare digestive tract tumor	ORPHA:98059	ST Patiëntenplatform Sarcomen
P 94	U 1936	Adamantinoma	ORPHA:55881	ST Patiëntenplatform Sarcomen
P 94	U 1937	Bone sarcoma	ORPHA:223727	ST Patiëntenplatform Sarcomen
P 94	U 1938	Bone sarcoma	ORPHA:223727	ST Patiëntenplatform Sarcomen
P 94	U 1939	Bone sarcoma	ORPHA:223727	ST Patiëntenplatform Sarcomen
P 94	U 1940	Bone Sarcoma	ORPHA:223727	ST Patiëntenplatform Sarcomen
P 143	U 366	Primary ciliary dyskinesia	ORPHA:244	ST PCD Belangengroep
P 143	U 367	Primary ciliary dyskinesia	ORPHA:244	ST PCD Belangengroep
P 279	U 77	Rare disease with Pierre Robin Syndrome	ORPHA:138044	ST Pierre Robin Europe
P 279	U 78	Rare disease with Pierre Robin Syndrome	ORPHA:138044	ST Pierre Robin Europe
P 279	U 79	Pierre Robin syndrome associated with a chromosomal anomaly	ORPHA:138047	ST Pierre Robin Europe
P 279	U 80	Isolated Pierre Robin syndrome	ORPHA:718	ST Pierre Robin Europe
P 279	U 81	Isolated Pierre Robin Syndrome	ORPHA:718	ST Pierre Robin Europe
P 279	U 82	Isolated Pierre Robin syndrome	ORPHA:718	ST Pierre Robin Europe
P 145	U 364	Pitt-Hopkins syndrome	ORPHA:2896	ST Pitt Hopkins Syndroom
P 312	U 1865	Genetic non-acquired premature ovarian failure	ORPHA:485382	ST POI-POF
P 312	U 1866	Acquired premature ovarian failure	ORPHA:95709	ST POI-POF
P 312	U 1867	Non acquired premature ovarian failure	ORPHA:95710	ST POI-POF
P 95	U 710	PTEN hamartoma tumor syndrome	ORPHA:306498	ST PTEN België-NL
P 95	U 711	Rare skin tumor or hamartoma	ORPHA:79386	ST PTEN België-NL
P 95	U 712	Malformation syndrome with hamartosis	ORPHA:98196	ST PTEN België-NL
P 158	U 315	Pulmonary Arterial Hypertension	ORPHA:182090	ST Pulmonale Hypertensie
P 158	U 316	Idiopathic pulmonary arterial hypertension	ORPHA:275766	ST Pulmonale Hypertensie

P 158	U 317	Idiopathic pulmonary arterial hypertension	ORPHA:275766	ST Pulmonale Hypertensie
P 158	U 318	Pulmonary arterial hypertension associated with another disease	ORPHA:275791	ST Pulmonale Hypertensie
P 158	U 319	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	ST Pulmonale Hypertensie
P 158	U 320	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	ST Pulmonale Hypertensie
P 158	U 321	Pulmonary arterial hypertension associated with connective tissue disease	ORPHA:275798	ST Pulmonale Hypertensie
P 158	U 322	Pulmonary arterial hypertension associated with congenital heart disease	ORPHA:275803	ST Pulmonale Hypertensie
P 158	U 323	Pulmonary arterial hypertension associated with congenital heart disease	ORPHA:275803	ST Pulmonale Hypertensie
P 158	U 324	Pulmonary arterial hypertension associated with portal hypertension	ORPHA:275813	ST Pulmonale Hypertensie
P 158	U 325	Pulmonary arterial hypertension associated with chronic hemolytic anemia	ORPHA:275828	ST Pulmonale Hypertensie
P 158	U 326	Pulmonary hypertension owing to lung disease and/or hypoxia	ORPHA:275837	ST Pulmonale Hypertensie
P 158	U 327	Pulmonary hypertension owing to lung disease and/or hypoxia	ORPHA:275837	ST Pulmonale Hypertensie
P 158	U 328	Pulmonary hypertension with unclear multifactorial mechanism	ORPHA:275844	ST Pulmonale Hypertensie
P 158	U 329	Rare Pulmonary Hypertension	ORPHA:71198	ST Pulmonale Hypertensie
P 158	U 330	Rare pulmonary hypertension	ORPHA:71198	ST Pulmonale Hypertensie
P 158	U 331	Rare pulmonary hypertension	ORPHA:71198	ST Pulmonale Hypertensie
P 158	U 332	Rare pulmonary hypertension	ORPHA:71198	ST Pulmonale Hypertensie
P 158	U 333	Rare pulmonary hypertension	ORPHA:71198	ST Pulmonale Hypertensie
P 158	U 1850	Chronic thromboembolic pulmonary hypertension	ORPHA:70591	ST Pulmonale Hypertensie
P 158	U 1851	Chronic thromboembolic pulmonary hypertension	ORPHA:70591	ST Pulmonale Hypertensie
P 280	U 76	IgG4-related retroperitoneal fibrosis	ORPHA:49041	ST RPF NL
P 48	U 1407	Rubinstein-Taybi syndrome	ORPHA:783	ST RTS - ST Rubinstein-Taybi Syndroom
P 271	U 88	22q11.2 deletion syndrome	ORPHA:567	ST Steun 22q11
P 271	U 89	22q11.2 deletion syndrome	ORPHA:567	ST Steun 22q11
P 75	U 1012	Sideroblastic anemia	ORPHA:1047	ST Zeldzame Bloedziekten
P 75	U 1013	Sideroblastic anemia	ORPHA:1047	ST Zeldzame Bloedziekten
P 75	U 1016	Hemophagocytic syndrome	ORPHA:158032	ST Zeldzame Bloedziekten
P 75	U 1017	Hemophagocytic syndrome	ORPHA:158032	ST Zeldzame Bloedziekten
P 75	U 1018	Hereditary elliptocytosis	ORPHA:288	ST Zeldzame Bloedziekten
P 75	U 1025	Rare constitutional hemolytic anemia due to a red cell membrane anomaly	ORPHA:98364	ST Zeldzame Bloedziekten
P 75	U 1026	Rare constitutional hemolytic anemia due to a red cell membrane anomaly	ORPHA:98364	ST Zeldzame Bloedziekten
P 75	U 1027	Hereditary stomatocytosis	ORPHA:98365	ST Zeldzame Bloedziekten
P 75	U 1032	Autoimmune hemolytic anemia	ORPHA:98375	ST Zeldzame Bloedziekten - AIHA Contactgroep
P 75	U 1033	Autoimmune hemolytic anemia	ORPHA:98375	ST Zeldzame Bloedziekten - AIHA Contactgroep
P 75	U 1014	Blackfan-Diamond anemia	ORPHA:124	ST Zeldzame Bloedziekten - BDA Contactgroep
P 75	U 1015	Blackfan-Diamond anemia	ORPHA:124	ST Zeldzame Bloedziekten - BDA Contactgroep
P 75	U 1022	Congenital dyserythropoietic anemia	ORPHA:85	ST Zeldzame Bloedziekten - CDA contactgroep
P 75	U 1023	Congenital dyserythropoietic anemia	ORPHA:85	ST Zeldzame Bloedziekten - CDA contactgroep
P 75	U 1020	Hemolytic anemia due to red cell pyruvate kinase deficiency	ORPHA:766	ST Zeldzame Bloedziekten - PKD Contactgroep
P 75	U 1028	Rare constitutional hemolytic anemia due to an enzyme disorder	ORPHA:98369	ST Zeldzame Bloedziekten - PKD Contactgroep
P 75	U 1021	Hereditary spherocytosis	ORPHA:822	ST Zeldzame Bloedziekten - Sferocytose Contactgroep
P 75	U 1029	Rare thrombotic disease of hematologic origin	ORPHA:182054	ST Zeldzame Bloedziekten - TTP Contactgroep
P 75	U 1030	Thrombotic microangiopathy	ORPHA:93573	ST Zeldzame Bloedziekten - TTP Contactgroep
P 75	U 1031	Thrombotic microangiopathy	ORPHA:93573	ST Zeldzame Bloedziekten - TTP Contactgroep
P 33	U 1905	Tumor of cranial and spinal nerves	ORPHA:252057	Stichting Hoormij
P 199	U 1951	Mitochondrial disease	ORPHA:68380	Stichting Nemo
P 199	U 1952	Mitochondrial disease	ORPHA:68380	Stichting Nemo

P 240	U 123	Usher syndrome	ORPHA:886	Stichting Ushersyndroom
P 226	U 135	Tumor of testis and paratestis	ORPHA:363472	Stichting Zaadbalkanker
P 226	U 136	Tumor of testis and paratestis	ORPHA:363472	Stichting Zaadbalkanker
P 226	U 1854	Testicular seminomatous germ cell tumor	ORPHA:842	Stichting Zaadbalkanker
P 226	U 1882	Germ cell tumor	ORPHA:3399	Stichting Zaadbalkanker
P 226	U 1883	Non-seminomatous germ cell tumor of testis	ORPHA:363494	Stichting Zaadbalkanker
P 226	U 1884	Germ cell tumor of testis	ORPHA:363504	Stichting Zaadbalkanker
P 226	U 1885	Germ cell tumor of testis	ORPHA:363504	Stichting Zaadbalkanker
P 226	U 1886	Extragenadal germ cell tumor	ORPHA:363579	Stichting Zaadbalkanker
P 226	U 1887	Testicular seminomatous germ cell tumor	ORPHA:842	Stichting Zaadbalkanker
P 206	U 181	Genetic cardiac rhythm disease	ORPHA:101934	STIN - ST ICD dragers NL
P 206	U 182	Genetic cardiac rhythm disease	ORPHA:101934	STIN - ST ICD dragers NL
P 206	U 183	Genetic cardiac rhythm disease	ORPHA:101934	STIN - ST ICD dragers NL
P 206	U 184	Rare cardiac rhythm disease	ORPHA:218436	STIN - ST ICD dragers NL
P 206	U 185	Rare cardiac rhythm disease	ORPHA:218436	STIN - ST ICD dragers NL
P 206	U 186	Rare cardiac rhythm disease	ORPHA:218436	STIN - ST ICD dragers NL
P 206	U 187	Non-genetic cardiac rhythm disease	ORPHA:218439	STIN - ST ICD dragers NL
P 206	U 188	Idiopathic ventricular fibrillation - not Brugada type	ORPHA:228140	STIN - ST ICD dragers NL
P 49	U 1404	Rare skin tumor or hamartoma	ORPHA:79386	STSN
P 49	U 1405	Tuberous sclerosis complex	ORPHA:805	STSN
P 49	U 1406	Tuberous sclerosis complex	ORPHA:805	STSN
P 317	U 1946	Twin to twin transfusion syndrome	ORPHA:95431	Taps support
P 317	U 1953	Selective IUGR	Pending	Taps Support
P 317	U 1954	Twin anemia polycythemia sequence	Pending	Taps Support
P 317	U 1955	Twin reversed arterial perfusion sequence	Pending	Taps Support
P 50	U 1403	Turner syndrome	ORPHA:881	Turner Contact NL
P 50	U 1860	Disorders of sex development	ORPHA:90771	Turner Contact NL
P 51	U 1392	Takayasu arteritis	ORPHA:3287	Vasculitis ST
P 51	U 1402	Rare systemic or rheumatologic disease	ORPHA:98023	Vasculitis ST
P 51	U 1380	Behcet disease	ORPHA:117	Vasculitits ST
P 51	U 1381	Predominantly large-vessel vasculitis	ORPHA:156140	Vasculitits ST
P 51	U 1382	Predominantly medium-vessel vasculitis	ORPHA:156143	Vasculitits ST
P 51	U 1383	Predominantly small-vessel vasculitis	ORPHA:156146	Vasculitits ST
P 51	U 1384	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152	Vasculitits ST
P 51	U 1385	Systemic autoimmune disease	ORPHA:182228	Vasculitits ST
P 51	U 1386	Systemic autoimmune disease	ORPHA:182228	Vasculitits ST
P 51	U 1387	Eosinophilic granulomatosis and polyangiitis	ORPHA:183	Vasculitits ST
P 51	U 1388	Kawasaki disease	ORPHA:2331	Vasculitits ST
P 51	U 1389	Kawasaki disease	ORPHA:2331	Vasculitits ST
P 51	U 1390	Rare pediatric vasculitis	ORPHA:280369	Vasculitits ST
P 51	U 1391	Rare Pediatric Vasculitis	ORPHA:280369	Vasculitits ST
P 51	U 1393	Anti-glomerular basement membrane disease	ORPHA:375	Vasculitits ST
P 51	U 1394	Secondary vasculitis	ORPHA:445197	Vasculitits ST
P 51	U 1395	Vasculitis	ORPHA:52759	Vasculitits ST
P 51	U 1396	Vasculitis	ORPHA:52759	Vasculitits ST
P 51	U 1397	Vasculitis	ORPHA:52759	Vasculitits ST

P 51	U 1398	Vasculitis	ORPHA:52759	Vasculitits ST
P 51	U 1399	Microscopic polyangiitis	ORPHA:727	Vasculitits ST
P 51	U 1400	Granulomatosis with polyangiitis	ORPHA:900	Vasculitits ST
P 51	U 1401	Cryoglobulinemic vasculitis	ORPHA:91138	Vasculitits ST
P 148	U 350	Vascular Ehlers-Danlos syndrome	ORPHA:286	VED - Vereniging van Ehlers Danlos Patiënten
P 148	U 351	Ehlers-Danlos syndrome	ORPHA:98249	VED - Vereniging van Ehlers Danlos Patiënten
P 148	U 352	Ehlers-Danlos syndrome	ORPHA:98249	VED - Vereniging van Ehlers Danlos Patiënten
P 3	U 1820	Angelman syndrome	ORPHA:72	Vereniging Angelman Syndroom NL
P 120	U 556	Anal fistula	ORPHA:228113	Vereniging Anusatesie
P 120	U 557	Anal fistula	ORPHA:228113	Vereniging Anusatesie
P 120	U 558	Anal fistula	ORPHA:228113	Vereniging Anusatesie
P 120	U 559	Ileal pouch anal anastomosis related faecal incontinence	ORPHA:238621	Vereniging Anusatesie
P 120	U 560	Isolated anorectal malformation	ORPHA:557	Vereniging Anusatesie
P 120	U 561	VACTERL/VATER association	ORPHA:887	Vereniging Anusatesie
P 120	U 562	Anorectal Malformations	ORPHA:96346	Vereniging Anusatesie
P 120	U 563	Anorectal Malformations	ORPHA:96346	Vereniging Anusatesie
P 120	U 564	Anorectal Malformations	ORPHA:96346	Vereniging Anusatesie
P 120	U 1917	Solitary rectal ulcer syndrome	ORPHA:209964	Vereniging Anusatesie
P 144	U 365	Cornelia de Lange syndrome	ORPHA:199	Vereniging Cornelia de Lange Syndroom
P 14	U 1773	Genetic cerebral small vessel disease	ORPHA:477754	Vereniging HCHWA-D
P 82	U 906	Blackfan-Diamond anemia	ORPHA:124	Vereniging Kinderkanker NL
P 82	U 907	Blackfan-Diamond anemia	ORPHA:124	Vereniging Kinderkanker NL
P 82	U 908	Epignathus	ORPHA:141077	Vereniging Kinderkanker NL
P 82	U 909	Nasopharyngeal teratoma	ORPHA:141107	Vereniging Kinderkanker NL
P 82	U 910	Glial tumor	ORPHA:182067	Vereniging Kinderkanker NL
P 82	U 911	Glial tumor	ORPHA:182067	Vereniging Kinderkanker NL
P 82	U 912	Glial Tumor	ORPHA:182067	Vereniging Kinderkanker NL
P 82	U 913	Polymalformative genetic syndrome with increased risk of developing cancer	ORPHA:183422	Vereniging Kinderkanker NL
P 82	U 914	Bone sarcoma	ORPHA:223727	Vereniging Kinderkanker NL
P 82	U 915	Bone sarcoma	ORPHA:223727	Vereniging Kinderkanker NL
P 82	U 916	Bone sarcoma	ORPHA:223727	Vereniging Kinderkanker NL
P 82	U 917	Bone Sarcoma	ORPHA:223727	Vereniging Kinderkanker NL
P 82	U 918	Rare tumor of the neuroepithelial tissue	ORPHA:251558	Vereniging Kinderkanker NL
P 82	U 919	Rare tumor of neuroepithelial tissue	ORPHA:251558	Vereniging Kinderkanker NL
P 82	U 920	Rare tumors of neuroepithelial tissue	ORPHA:251558	Vereniging Kinderkanker NL
P 82	U 921	Ollier disease	ORPHA:296	Vereniging Kinderkanker NL
P 82	U 922	Skeletal Ewing sarcoma	ORPHA:319	Vereniging Kinderkanker NL
P 82	U 923	Multiple osteochondromas	ORPHA:321	Vereniging Kinderkanker NL
P 82	U 924	Multiple osteochondromas	ORPHA:321	Vereniging Kinderkanker NL
P 82	U 925	Soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL
P 82	U 926	soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL
P 82	U 927	Soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL
P 82	U 928	Soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL
P 82	U 929	Soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL
P 82	U 930	Soft tissue sarcoma	ORPHA:3394	Vereniging Kinderkanker NL
P 82	U 931	Germ cell tumor	ORPHA:3399	Vereniging Kinderkanker NL

P 82	U 932	Non-seminomatous germ cell tumor of testis	ORPHA:363494	Vereniging Kinderkanker NL
P 82	U 933	Germ cell tumor of testis	ORPHA:363504	Vereniging Kinderkanker NL
P 82	U 934	Germ cell tumor of testis	ORPHA:363504	Vereniging Kinderkanker NL
P 82	U 935	Extragenitaal germ cell tumor	ORPHA:363579	Vereniging Kinderkanker NL
P 82	U 936	Dysostosis	ORPHA:364559	Vereniging Kinderkanker NL
P 82	U 937	Aneurysmal bone cyst	ORPHA:480553	Vereniging Kinderkanker NL
P 82	U 938	Sacrococcygeal teratoma	ORPHA:494421	Vereniging Kinderkanker NL
P 82	U 939	Sacrococcygeal teratoma	ORPHA:494421	Vereniging Kinderkanker NL
P 82	U 940	Sacrococcygeal teratoma	ORPHA:494421	Vereniging Kinderkanker NL
P 82	U 941	MALT lymphoma	ORPHA:52417	Vereniging Kinderkanker NL
P 82	U 942	Craniopharyngioma	ORPHA:54595	Vereniging Kinderkanker NL
P 82	U 943	Chondrosarcoma	ORPHA:55880	Vereniging Kinderkanker NL
P 82	U 944	Adamantinoma	ORPHA:55881	Vereniging Kinderkanker NL
P 82	U 945	Osteosarcoma	ORPHA:668	Vereniging Kinderkanker NL
P 82	U 946	Osteosarcoma	ORPHA:668	Vereniging Kinderkanker NL
P 82	U 947	Rare bone tumor	ORPHA:68411	Vereniging Kinderkanker NL
P 82	U 948	Rare bone tumor	ORPHA:68411	Vereniging Kinderkanker NL
P 82	U 949	Rare soft tissue tumor	ORPHA:71209	Vereniging Kinderkanker NL
P 82	U 950	Rare soft tissue tumor	ORPHA:71209	Vereniging Kinderkanker NL
P 82	U 951	Rare soft tissue tumor	ORPHA:71209	Vereniging Kinderkanker NL
P 82	U 952	Rare soft tissue tumor	ORPHA:71209	Vereniging Kinderkanker NL
P 82	U 953	Solitary bone cyst	ORPHA:83468	Vereniging Kinderkanker NL
P 82	U 954	Testicular seminomatous germ cell tumor	ORPHA:842	Vereniging Kinderkanker NL
P 82	U 955	Rare nervous system tumor	ORPHA:98062	Vereniging Kinderkanker NL
P 154	U 334	Primary bone dysplasia	ORPHA:364526	Vereniging MED-SED
P 154	U 335	Primary Bone Dysplasia	ORPHA:364526	Vereniging MED-SED
P 16	U 1760	Huntington disease	ORPHA:399	Vereniging van Huntington
P 16	U 1761	Huntington disease	ORPHA:399	Vereniging van Huntington
P 16	U 1762	Huntington disease	ORPHA:399	Vereniging van Huntington
P 231	U 134	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	ORPHA:95455	Vereniging van mensen met brandwonden
P 84	U 894	Non-histaminic angioedema	ORPHA:658	Vereniging voor angio oedeem
P 84	U 895	Hereditary angioedema	ORPHA:91378	Vereniging voor angio oedeem
P 237	U 124	Inherited ichthyosis	ORPHA:183435	Vereniging voor Ichthyosisnetwerken
P 237	U 125	Darier disease	ORPHA:218	Vereniging voor Ichthyosisnetwerken
P 237	U 126	Darier disease	ORPHA:218	Vereniging voor Ichthyosisnetwerken
P 237	U 127	Ichthyosis	ORPHA:79354	Vereniging voor Ichthyosisnetwerken
P 237	U 128	Ichthyosis	ORPHA:79354	Vereniging voor Ichthyosisnetwerken
P 123	U 503	Rare disease involving intestinal motility	ORPHA:104009	Vereniging Ziekte van Hirschsprung
P 123	U 504	Rare disease involving intestinal motility	ORPHA:104009	Vereniging Ziekte van Hirschsprung
P 123	U 505	Myopathic intestinal pseudoobstruction	ORPHA:104077	Vereniging Ziekte van Hirschsprung
P 123	U 506	Unclassified intestinal pseudoobstruction	ORPHA:104078	Vereniging Ziekte van Hirschsprung
P 123	U 507	Rare intestinal disease	ORPHA:117569	Vereniging Ziekte van Hirschsprung
P 123	U 508	Rare intestinal disease	ORPHA:117569	Vereniging Ziekte van Hirschsprung
P 123	U 509	Chronic intestinal pseudoobstruction	ORPHA:2978	Vereniging Ziekte van Hirschsprung
P 123	U 510	Chronic intestinal pseudoobstruction	ORPHA:2978	Vereniging Ziekte van Hirschsprung
P 123	U 511	Chronic intestinal pseudoobstruction	ORPHA:2978	Vereniging Ziekte van Hirschsprung

P 123	U 512	Chronic intestinal pseudoobstruction	ORPHA:2978	Vereniging Ziekte van Hirschsprung
P 123	U 513	Hirschsprung disease	ORPHA:388	Vereniging Ziekte van Hirschsprung
P 123	U 514	Hirschsprung disease	ORPHA:388	Vereniging Ziekte van Hirschsprung
P 123	U 515	Hirschsprung disease	ORPHA:388	Vereniging Ziekte van Hirschsprung
P 123	U 516	Hirschsprung disease	ORPHA:388	Vereniging Ziekte van Hirschsprung
P 123	U 517	Neuronal intestinal pseudoobstruction	ORPHA:99811	Vereniging Ziekte van Hirschsprung
P 56	U 1294	Rare dyslipidemia	ORPHA:101953	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1295	Rare dyslipidemia	ORPHA:101953	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1296	Rare dyslipidemia	ORPHA:101953	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1297	Non-syndromic central nervous system malformation	ORPHA:108989	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1298	Congenital disorder of glycosylation	ORPHA:137	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1299	Pantothenate kinase-associated neurodegeneration	ORPHA:157850	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1300	Rare hyperlipidemia	ORPHA:181422	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1301	Cockayne Syndrome	ORPHA:191	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1302	Essential fructosuria	ORPHA:2056	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1303	Neuronal ceroid lipofuscinosis	ORPHA:216	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1304	Neuronal ceroid lipofuscinosis	ORPHA:216	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1305	CLN3-ziekte	ORPHA:228346	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1306	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	ORPHA:247691	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1307	Glutaryl-CoA dehydrogenase deficiency	ORPHA:25	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1308	Glycogen storage disease due to liver phosphorylase kinase deficiency	ORPHA:264580	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1309	Phosphoserine aminotransferase deficiency, infantile/juvenile form	ORPHA:284417	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1310	Disorder of phenylalanine metabolism	ORPHA:284814	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1311	Disorder of folate metabolism and transport	ORPHA:285657	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1312	organic aciduria	ORPHA:289899	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1313	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A	ORPHA:308386	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1314	Disorder of fructose metabolism	ORPHA:308463	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1315	Disorder of carnitine cycle and carnitine transport	ORPHA:309130	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1316	Disorder of carnitine cycle and carnitine transport	ORPHA:309130	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1317	glycoproteinosis	ORPHA:309279	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1318	Trichothiodystrophy	ORPHA:33364	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1319	glycogen storage disease due LAMP2 deficiency	ORPHA:34587	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1320	Fructose-1,6-biphosphatasedeficiency	ORPHA:348	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1321	glycogen storage disease due to acid maltase deficiency	ORPHA:365	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1322	Glycogen storage disease due to glycogen debranching enzyme deficiency	ORPHA:366	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1323	Glycogen storage disease due to glycogen branching enzyme deficiency	ORPHA:367	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1324	Neurodegeneration with brain iron accumulation	ORPHA:385	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1325	Classic homocystinuria	ORPHA:394	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1326	Gyrate atrophy of choroid and retina	ORPHA:414	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1327	Gyrate atrophy of choroid and retina	ORPHA:414	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1328	Bilirubin encephalopathy	ORPHA:415286	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1329	Primary Hyperoxaluria	ORPHA:416	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1330	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1331	Hypophosphatemic Rickets	ORPHA:437	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1332	Hereditary fructose intolerance	ORPHA:469	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1333	Leukodystrophies	ORPHA:68356	VKS - Volwassenen, Kinderen en Stofwisselingsziekten

P 56	U 1334	Lysosomal disease	ORPHA:68366	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1335	Lysosomal disease	ORPHA:68366	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1336	Lysosomal disease	ORPHA:68366	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1337	Rare inborn errors of metabolism	ORPHA:68367	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1338	Rare inborn errors of metabolism	ORPHA:68367	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1339	rare inborn errors of metabolism	ORPHA:68367	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1340	Peroxisomal disease	ORPHA:68373	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1341	Mitochondrial disease	ORPHA:68380	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1342	Mitochondrial disease	ORPHA:68380	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1343	Neurometabolic disease	ORPHA:68385	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1344	Classic glucose transporter type 1 deficiency syndrome	ORPHA:71277	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1345	disorder of amino acid and other organic acid metabolism	ORPHA:79062	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1346	disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1347	Disorders of Neurotransmitter metabolism and transport	ORPHA:79169	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1348	Creatine deficiency syndrome	ORPHA:79172	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1349	Disorder of fatty acid oxidation and ketone body metabolism	ORPHA:79174	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1350	Disorder of fatty acid oxidation and ketone body metabolism	ORPHA:79174	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1351	Disorder of fatty acid oxidation and ketone body metabolism	ORPHA:79174	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1352	Disorders of pyridoxine metabolism	ORPHA:79192	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1353	Disorders of pyridoxine metabolism	ORPHA:79192	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1354	disorder of branched-chain amino acid metabolism	ORPHA:79197	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1355	Disorder of energy metabolism	ORPHA:79200	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1356	Glycogen storage disease	ORPHA:79201	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1357	Glycogen storage disease	ORPHA:79201	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1358	disorder of lysosomal amino acid transport	ORPHA:79207	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1359	mucopolysaccharidosis	ORPHA:79213	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1360	Disorder of purine or pyrimidine metabolism	ORPHA:79224	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1361	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ia	ORPHA:79258	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1362	Glycogen storage disease due to glucose-6-phosphatase deficiency type Ib	ORPHA:79259	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1363	Juvenile neuronal ceroid lipofuscinosis	ORPHA:79264	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1364	3-phosphoserine phosphatase deficiency, infantile/juvenile form	ORPHA:79350	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1365	3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form	ORPHA:79351	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1366	Wilson disease	ORPHA:905	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1367	Primary lipodystrophy	ORPHA:90970	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1368	Primary bone dysplasia with defective bone mineralization	ORPHA:93447	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1369	Rare renal tubular disease	ORPHA:93603	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1370	Rare renal tubular disease	ORPHA:93603	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1371	Rare renal tubular disease	ORPHA:93603	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1372	rare renal tubular disease	ORPHA:93603	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1373	Central nervous system malformation	ORPHA:98044	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 56	U 1374	Central nervous system malformation	ORPHA:98044	VKS - Volwassenen, Kinderen en Stofwisselingsziekten
P 53	U 1377	Osteogenesis imperfecta	ORPHA:666	VOI - Vereniging Osteogenesis Imperfecta
P 53	U 1378	Osteogenesis imperfecta	ORPHA:666	VOI - Vereniging Osteogenesis Imperfecta
P 53	U 1379	Primary bone dysplasia with decreased bone density	ORPHA:93446	VOI - Vereniging Osteogenesis Imperfecta
P 53	U 1947	Primary bone dysplasia	ORPHA:364526	VOI - Vereniging Osteogenesis Imperfecta
P 53	U 1948	Primary Bone Dysplasia	ORPHA:364526	VOI - Vereniging Osteogenesis Imperfecta

P 124	U 495	Tubular duplication of the esophagus	ORPHA:100048	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 496	Esophageal Atresia	ORPHA:1199	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 497	Esophageal atresia	ORPHA:1199	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 498	Esophageal atresia	ORPHA:1199	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 499	Tracheal anomaly	ORPHA:156252	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 500	Isolated tracheoesophageal fistula	ORPHA:454750	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 501	VACTERL/VATER association	ORPHA:887	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 124	U 502	Idiopathic achalasia	ORPHA:930	VOKS - Vereniging voor Ouderen en Kinderen met Slokdarmafsluiting
P 55	U 1375	Ectodermal dysplasia syndrome	ORPHA:79373	VVED- Vereniging voor Ectodermale Dysplasie
P 55	U 1376	Ectodermal dysplasia syndrome	ORPHA:79373	VVED- Vereniging voor Ectodermale Dysplasie
P 55	U 1925	Oligodontia	ORPHA:99798	VVED- Vereniging voor Ectodermale Dysplasie
P 55	U 1931	Gorlin syndrome	ORPHA:377	VVED- Vereniging voor Ectodermale Dysplasie
P 221	U 137	Prepubertal anorexia nervosa	ORPHA:525738	Weet: ST rond eetstoornissen
P 178	U 214	Rare chromosomal anomaly	ORPHA:68335	XXX-Contactgroep Triple-X Syndroom
P 290	U 26	Multiple congenital anomalies/dysmorphic syndrome-intellectual disability	ORPHA:102283	Zeldsamen
P 290	U 27	Multiple congenital anomalies/dysmorphic syndrome-variable intellectual disability syndrome	ORPHA:102284	Zeldsamen
P 290	U 28	Rare syndromic intellectual disability	ORPHA:102369	Zeldsamen
P 290	U 29	Rare syndromic intellectual disability	ORPHA:102369	Zeldsamen
P 290	U 30	CHARGE syndrome	ORPHA:138	Zeldsamen
P 290	U 31	CHARGE syndrome	ORPHA:138	Zeldsamen
P 290	U 32	KBG syndrome	ORPHA:2332	Zeldsamen
P 290	U 33	15q11q13 microduplication syndrome	ORPHA:238446	Zeldsamen
P 290	U 34	Proximal 16p11.2 microdeletion syndrome	ORPHA:261197	Zeldsamen
P 290	U 35	16p11.2p12.2 microduplication syndrome	ORPHA:261204	Zeldsamen
P 290	U 36	Distal 16p11.2 microdeletion syndrome	ORPHA:261222	Zeldsamen
P 290	U 37	Kleefstra syndrome	ORPHA:261494	Zeldsamen
P 290	U 38	Anomaly of Chromosome 6	ORPHA:261712	Zeldsamen
P 290	U 39	Malan overgrowth syndrome	ORPHA:420179	Zeldsamen
P 290	U 40	DYRK1A-related intellectual disability syndrome	ORPHA:464306	Zeldsamen
P 290	U 41	Monosomy 22q13.3 (Phelan-McDermid syndrome)	ORPHA:48652	Zeldsamen
P 290	U 42	SIN3A-related intellectual disability syndrome/Witteveen-Kolk syndrome	ORPHA:500163	Zeldsamen
P 290	U 43	Menke-Hennekam syndrome	ORPHA:592574	Zeldsamen
P 290	U 44	Rare chromosomal anomaly	ORPHA:68335	Zeldsamen
P 290	U 45	Rare chromosomal anomaly	ORPHA:68335	Zeldsamen
P 290	U 46	Multiple congenital anomalies/dysmorphic syndrome	ORPHA:68341	Zeldsamen
P 290	U 47	Rare intellectual disability	ORPHA:87277	Zeldsamen
P 290	U 48	Koolen-de Vries syndrome	ORPHA:96169	Zeldsamen
P 290	U 49	Autosomal anomaly	ORPHA:98127	Zeldsamen
P nvt	U 146	Periventricular leukomalacia	ORPHA:171676	Zonder patiëntenorganisatie
P nvt	U 157	Kleine-Levin syndrome	ORPHA:33543	Zonder patiëntenorganisatie
P nvt	U 159	Rare autonomic nervous system disorder	ORPHA:423662	Zonder patiëntenorganisatie
P nvt	U 161	Pure autonomic failure	ORPHA:441	Zonder patiëntenorganisatie
P nvt	U 525	Congenital alveolar capillary dysplasia	ORPHA:210122	Zonder patiëntenorganisatie
P nvt	U 526	Congenital alveolar capillary dysplasia	ORPHA:210122	Zonder patiëntenorganisatie
P nvt	U 539	Tuberculosis	ORPHA:3389	Zonder patiëntenorganisatie
P nvt	U 540	Tuberculosis	ORPHA:3389	Zonder patiëntenorganisatie

P nvt	U 546	Recurrent Respiratory Papillomatosis	ORPHA:60032	Zonder patiëntenorganisatie
P nvt	U 1024	Constitutional anemia due to iron metabolism disorder	ORPHA:98360	Zonder patiëntenorganisatie
P nvt	U 1106	Rare hemorrhagic disorder due to an acquired coagulation factor defect	ORPHA:166775	Zonder patiëntenorganisatie
P nvt	U 1641	Glomus tumor	ORPHA:391651	Zonder patiëntenorganisatie
P nvt	U 1785	sphingolipidosis	ORPHA:79225	Zonder patiëntenorganisatie
P nvt	U nvt	Rickettsial disease	ORPHA:102021	Zonder patiëntenorganisatie
P nvt	U nvt	Birt-Hogg-Dubé syndrome	ORPHA:122	Zonder patiëntenorganisatie
P nvt	U nvt	Birt-Hogg-Dubé syndrome	ORPHA:122	Zonder patiëntenorganisatie
P nvt	U nvt	Schistosomiasis	ORPHA:1247	Zonder patiëntenorganisatie
P nvt	U nvt	Schistosomiasis	ORPHA:1247	Zonder patiëntenorganisatie
P nvt	U nvt	Camurati-Engelmann disease	ORPHA:1328	Zonder patiëntenorganisatie
P nvt	U nvt	Rare parasitic disease	ORPHA:163588	Zonder patiëntenorganisatie
P nvt	U nvt	Rare parasitic disease	ORPHA:163588	Zonder patiëntenorganisatie
P nvt	U nvt	Mycetoma	ORPHA:2583	Zonder patiëntenorganisatie
P nvt	U nvt	Osteochondritis dissecans	ORPHA:2764	Zonder patiëntenorganisatie
P nvt	U nvt	Viral hemorrhagic fever	ORPHA:341	Zonder patiëntenorganisatie
P nvt	U nvt	Viral hemorrhagic fever	ORPHA:341	Zonder patiëntenorganisatie
P nvt	U nvt	Semicircular canal dehiscence syndrome	ORPHA:420402	Zonder patiëntenorganisatie
P nvt	U nvt	Semicircular canal dehiscence syndrome	ORPHA:420402	Zonder patiëntenorganisatie
P nvt	U nvt	Pudendal neuralgia	ORPHA:60039	Zonder patiëntenorganisatie
P nvt	U nvt	Malaria	ORPHA:673	Zonder patiëntenorganisatie
P nvt	U nvt	Malaria	ORPHA:673	Zonder patiëntenorganisatie
P nvt	U nvt	Strongyloidiasis	ORPHA:76	Zonder patiëntenorganisatie
P nvt	U nvt	Rabies	ORPHA:770	Zonder patiëntenorganisatie
P nvt	U nvt	Rabies	ORPHA:770	Zonder patiëntenorganisatie
P nvt	U nvt	Rare form of salmonellosis	ORPHA:795	Zonder patiëntenorganisatie
P nvt	U nvt	Hinman syndrome	ORPHA:84085	Zonder patiëntenorganisatie
P nvt	U nvt	Hinman syndrome	ORPHA:84085	Zonder patiëntenorganisatie
P nvt	U nvt	Thoracic outlet syndrome	ORPHA:97330	Zonder patiëntenorganisatie
P nvt	U nvt	Rare surgical thoracic diseases	ORPHA:97962	Zonder patiëntenorganisatie
P nvt	U nvt	Yellow fever	ORPHA:99829	Zonder patiëntenorganisatie
P nvt	U nvt	Implant-Related Infections	Pending	Zonder patiëntenorganisatie