	Informatie
Network Code	**Thematic area**
BOND	Short stature (+/- disproportionate)
BOND	
BOND	
BOND	
BOND	Increased bone fragility
BOND	
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BOND	Increased bone density and osteosclerosing dysplasia
BOND	
BOND	
BOND	Abnormal development of skeletal component
BOND	
BOND	
BOND	SE(M)D
BOND	
BOND	
BOND	Disorders of phosphate metabolism
BOND	
BOND	Acromelic dysplasia (ORPHA:93436)
BOND	
BOND	
BOND	
BOND	Ciliopathies with major skeletal involvement (ORPHA:93426)
BOND	
BOND	Dislocations (ORPHA:93441)
BOND	
BOND	
BOND	Overgrowth with skeletal involvement (ORPHA:498448)
BOND	
BOND	Dysostosis (ORPHA:364559)
BOND	
CRANIO	
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CRANIO	Malformations of the cranium
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CRANIO	Facial malformations
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CRANIO	Otorhinolaryngological/ENT malformations
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CRANIO	Rare ordontal or periodontal disorders
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ENDO	Adrenal
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ENDO	Sexual development and maturation
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ENDO	
ENDO	Growth (and genetic obesity syndromes) (ORPHA:90692 en ORPHA:240371)
ENDO	
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ENDO	Pituitary
ENDO	
ENDO	
ENDO	Disorders of calcium and phosphate homeostasis
ENDO	
ENDO	
ENDO	Thyroid (ORPHA:101955 of Rare genetic thyroid disease ORPHA:183631)
ENDO	
ENDO	Genetic disorders of glucose and insulin homeostasis
ENDO	
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ENDO	Genetic endocrine tumour syndromes
ENDO	
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EpiCARE	Genetic epilepsy
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EpiCARE	
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EpiCARE	Structural epilepsies
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EpiCARE	
EpiCARE	Infectious epilepsies
EpiCARE	
EpiCARE	Immune epilepsies
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EpiCARE	Surgically treatable epilepsies
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EpiCARE	Syndromic epilepsies
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EpiCARE	
EpiCARE	Metabolic epilepsies
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EpiCARE	Neonatal seizures (acute)
EpiCARE	
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EpiCARE	Neonatal seizures (metabolic)
EpiCARE	
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EpiCARE	
EpiCARE	
EpiCARE	Neonatal seizures (syndromic)
EpiCARE	
EpiCARE	
EpiCARE	
EpiCARE	Status epiletlcus
EpiCARE	
EpiCARE	
EpiCARE	
ERKNet	Rare glomerulopathies
ERKNet	

ERKNet	Tubulopathies and tubulointerstitial diseases
ERKNet	
ERKNet	Thrombotic microangiopathies
ERKNet	Autosomal dominant renal dysplastic diseases
ERKNet	Severe malformations of kidney & urinary tract
ERKNet	
ERKNet	Ciliopathies
ERKNet	Pediatric chronic kidney disease
ERKNet	
EYE	Retinal Dystrophy
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EYE	Neuro-ophthalmology and rare eye diseases
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EYE	Pediatric Ophthalmology Rare Diseases
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EYE	Anterior Segment Rare Eye Diseases
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ERNICA	Esophageal malformations
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ERNICA	Intestinal malformations/diseases
ERNICA	
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ERNICA	
ERNICA	Inherited and congenital pancreatic diseases
ERNICA	Non-syndromic diaphragmatic or abdominal wall malformations
ERNICA	
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ERNICA	
ERNICA	Syndromic diaphragmatic or abdominal wall malformations
ERNICA	
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LUNG	Rare pulmonary diseases (ILD, CF, nCF BE, PCD, CLAD, AATD)
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LUNG	Pulmonary hypertension (PH)
LUNG	Rare respiratory tumor (MSTO)
RND	Rare neurological diseases
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SKIN	A. Epidermolysis Bullosa (EB)
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SKIN	B. Ichthyosis (I) & Palmoplantar Keratoderma (PPK)
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SKIN	C. Ectodermal Dysplasia (ED) including Skin Fragility Disorders & X-linked cutaneous disorders & unclassified disorders
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SKIN	D. Monogenic Connective Tissue Disorders
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SKIN	E. Cutaneous Mosaic Disorders - Nevi & Nevoid Skin Disorders and Complex Vascular Malformations and vascular Tumours
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SKIN	F. Cutaneous diseases related to DNA Repair

SKIN	Disorders
SKIN	G. Autoimmune bullous diseases and severe cutaneous drug reactions
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SKIN	H. Acquired immunoLogical Low prevalence and Complex Adult diseases of The skin
SKIN	
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SKIN	I. Photosensitivity
SKIN	
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SKIN	J. Non bullous complex auto immune/inflammatory cutaneous diseases
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SKIN	K. Premature Skin Ageing
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SKIN	L. Rare cutaneous proliferations in children and adults
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EURACAN	G1: Rare neoplasm of connective tissue
EURACAN	G2: Rare neoplasm of the female genital organs and placenta
EURACAN	G3: Rare neoplams of the male genital organs, and of the urinary tract
EURACAN	G4: Rare neoplasm of the neuroendocrine system
EURACAN	G5: Rare neoplasm if the digestive tract
EURACAN	G6: Rare neoplasm of endocrine organs
EURACAN	G7: Rare neoplasm of the head and neck
EURACAN	G8: Rare neoplasmof the thorax
EURACAN	G9: Rare neoplasm of the skin and eye melanoma
EURACAN	G10: Rare neoplasm of the brain, spinal cords
EuroBloodNet	Rare red blood cell (RBC) defects
EuroBloodNet	Rare red blood cell Hereditary (RBC) defects erythroenzymopathies

EuroBloodNet	Rare red blood cell (RBC) defects
EuroBloodNet	
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EuroBloodNet	BMF and rare haematopoietic disorders
EuroBloodNet	
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EuroBloodNet	
EuroBloodNet	Rare bleeding - coagulation disorders
EuroBloodNet	
EuroBloodNet	
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EuroBloodNet	
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EuroBloodNet	HH-iron metabolism-Heme synthesis
EuroBloodNet	
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EuroBloodNet	
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EuroBloodNet	Myeloid malignancies
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EuroBloodNet	Lymphoid malignancies
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eUROGEN	1.1 Complex genital reconstructions (DSDs)
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	1.2 Bladder exstrophy/epispadias
eUROGEN	
eUROGEN	
eUROGEN	

eUROGEN	1.3 Rare urological stone and kidney diseases (in 2016 - 'renal disorders')
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	1.4 Non-syndromic urogenital tract malformation
eUROGEN	
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eUROGEN	1.5 Posterior urethral valve
eUROGEN	1.6 Posterior hypospadias
eUROGEN	1.7 Urorectal/anorectal malformations
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	2.1 Complicated & complex pelvic floor disorders
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	
eUROGEN	2.2 Rare diseases & conditions affecting female urethra
eUROGEN	2.3 Urethral reconstruction in rare diseases & conditions
eUROGEN	2.4 Rare retroperitoneal diseases & conditions
eUROGEN	2.5 Interstitial cystitis

eUROGEN	2.6 Surgery (in adulthood) for late effects of congenital and malignant childhood conditions
eUROGEN	
eUROGEN	2.7 Surgery for transgender patients
eUROGEN	3.1 Penile cancer
eUROGEN	3.2 Testis cancer
eUROGEN	3.3 Adrenal tumors
eUROGEN	3.4 Abdomino pelvic sarcoma
eUROGEN	3.5 Rare Renal Tumours requiring highly specialized surgery
EURO-NMD	Muscle Diseases
EURO-NMD	
EURO-NMD	Neuromuscular Junction (NMJ) Defects
EURO-NMD	
EURO-NMD	Peripheral Neuropathies
EURO-NMD	
EURO-NMD	Motor Neuron Disorders
EURO-NMD	
EURO-NMD	Mitochondrial diseases
EURO-NMD	
GENTURIS	Neurofibromatoses
GENTURIS	
GENTURIS	
GENTURIS	Lynch and polyposis
GENTURIS	
GENTURIS	Hereditary breast and ovarian cancer
GENTURIS	
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GENTURIS	
GENTURIS	Other rare PREDOMINANTLY malignant

GENTURIS	genturis
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GUARD-HEART	Familial Electrical Diseases in adults and/or children
GUARD-HEART	
GUARD-HEART	
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GUARD-HEART	
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	Special electrophysiology conditions in children
GUARD-HEART	Familial Cardiomyopathies in adults and/or children
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	Congential Heart Defects in adults (GUCH) or children
GUARD-HEART	Other rare cardiac diseases in adults and children
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	
GUARD-HEART	
ITHACA	Rare Congenital malformations diagnosed and undiagnosed and rare intellectual
ITHACA	
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ITHACA	disability
METABERN	Amino and organic acids- related disorders (AOA)
METABERN	
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METABERN	
METABERN	Disorders of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders, disorders of thiamine transport and metabolism (PM-MD)
METABERN	Carbohydrate, fatty acid oxidation disorders (C-FAO)
METABERN	
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METABERN	Lysosomal disorders (LSD)
METABERN	
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METABERN	Peroxisomal Disorders (PD)
METABERN	
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METABERN	Congenital disorders of Glycosylation (CDG)
METABERN	Disorders of neuromodulators and other small molecules (NOMS)
METABERN	
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METABERN	
PaedCAN	Leukemias, myeloproliferative and myelodysplastic diseases
PaedCAN	Lymphomas and reticuloendothelial neoplasms
PaedCAN	CNS+miscell.intracranial &intraspinal neoplasms
PaedCAN	Neuroblastoma and other peripheral nervous cell tumour
PaedCAN	Retinoblastoma
PaedCAN	Renal tumors
PaedCAN	Hepatic tumors
PaedCAN	Malignant bone tumors
PaedCAN	Soft tissue and other extraosseous sarcomas
PaedCAN	Germ cell tumors, trophoblastic tumors, and neoplasms of gonads
PaedCAN	Other malignant epithelial neoplasms and malignant melanomas
PaedCAN	Other unspecified malignant neoplasmas (Hematopoietic and Lymphoid:LCH)
RARE-LIVER	Autoimmune Liver Disease
RARE-LIVER	
RARE-LIVER	
RARE-LIVER	
RARE-LIVER	Metabolic, Biliary Atresia & Related Diseases
RARE-LIVER	
RARE-LIVER	

RARE-LIVER	
RARE-LIVER	
RARE-LIVER	Structural Liver Disease
RARE-LIVER	
RARE-LIVER	
RARE-LIVER	Rare causes of acute liver failure
RARE-LIVER	Liver disease in pregnancy
RARE-LIVER	
RARE-LIVER	
ReCONNET	Rare autoimmune connective tissue and musculoskeletal diseases
ReCONNET	
ReCONNET	
ReCONNET	Complex autoimmune connective tissue and musculoskeletal diseases
ReCONNET	Hereditary connective tissue diseases
RITA	Primary Immunodeficiencies
RITA	Auto-Inflammatory Diseases
RITA	Autoimmune Diseases
RITA	Autoimmune - Pediatric Rheumatology
TRANSPLANTCHILD	Pre-transplantation
TRANSPLANTCHILD	Transplantation
TRANSPLANTCHILD	
TRANSPLANTCHILD	
TRANSPLANTCHILD	
TRANSPLANTCHILD	

TRANSPLANTCHILD	
TRANSPLANTCHILD	
TRANSPLANTCHILD	Post-transplantation
VASCERN	Rare Multisystemic Vascular Diseases, Hereditary haemorrhagic telangiectasia (HHT)
VASCERN	Rare Multisystemic Vascular Diseases, Heritable Thoracic Aortic Disorders (HTAD)
VASCERN	
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VASCERN	Rare Multisystemic Vascular Diseases, Vascular Anomalies (VASCA)
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VASCERN	Rare Multisystemic Vascular Diseases,

VASCERN	Pediatric and Primary Lymphedema (PPL)
VASCERN	
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VASCERN	Rare Multisystemic Vascular Diseases, Medium Size Arteries (MSA)
VASCERN	
VASCERN	

e verstrekt door de ERNs
Sub-thematic area
Achondroplasia
Hypochondroplasia
Dyschondrosteosis
Acromesomelia
Osteogenesis imperfecta
Hypophosphatasia
Cleido-cranial dysplasia
Osteopetrosis
Camurati engelman
Osteopathia striata cranial sclerosis (OSCS)
Multiple cartilaginous exostosis
Ollier-Maffucci
Fibrodysplasia Ossificans Progressiva
Bone fibrous dysplasia- McC-A
Collagenopathies II
MED
Metatropic and Kozlowski
Hypophosphatemic rickets (XLH)
Tumoral calcinosis and Hyperostosis Hyperphosphatemia
Acrodysostosis
Osteoarthropathy Albright
Myhre
Acromicric dysplasia
Geleophysic dysplasia
Asphyxiating Thoracic Dysplasia
Ellis van Creveld
Larsen
Desbuquois
Diastrophic
Sotos, Weaver
Marshall Smith
Poland, Adams Oliver
Ectrodactyly
Isolated Craniosynostosis
Syndromic craniosynostosis
Frontal encephalocele
Basal encephalocele

Fibrous dysplasia of bone (craniofacial presentation)
Neurofibromatosis type 1 (craniofacial presentation)
Cleidocranial dysostosis
Oculo-auriculo-vertebral spectrum (/Craniofacial microsomia)
Facial dysostosis (Treacher Collins + Nager syndrome)
Miller syndrome
Aplasia cutis congenital scalp
Craniofacial clefts
Frontonasal dysplasia
Isolated hereditary congenital facial paralysis
Moebius Syndrome
Hypoglossia / Aglossia
Macroglossia
Cleft lip with or without cleft palate (syndromic and/or non)
Cleft palate (syndromic and/or non)
Isolated Pierre robin
Genetic syndromic Pierre Robin syndrome
Laryngo-tracheoesophageal cleft
Congenital tracheal stenosis
Congenital subglottic stenosis
Non-syndromic genetic deafness
Nose and cavum anomaly
Congenital anomalies of the neck
Microtia
Anotia
Oligodontia
Dentin dysplasia
Dentinogenesis imperfecta
Amelogenesis imperfecta
Hypodontia
Eruption/resorption/early loss of teeth
Supernumerary teeth
Solitary median maxillary central incisor syndrome
Familial and sporadic pheochromocytoma/ paraganglioma
Adrenocortical carcinoma (ACC, stage I+II)
Cortisol producing adenomas
(Primary) Adrenal insufficiency (AI)
Congenital adrenal hyperplasia (CAH)
Familial Hyperaldosteronism (FH)
DSD
Isolated HH
46,XX Ovarian dysgenesis or Primary Ovarian Insufficiency (POI)

Gender Dysphoria
Prader Willi and Prader Willi-like syndromes
Silver Russell Syndrome
Beckwith Wiedemann syndrome
Noonan Syndrome
Growth hormone resistance
Overgrowth syndrome
Short stature
pituitary adenoma
congenital hypopituitarism
acquired hypopituitarism
subsubthematic network pituitary (guideline pituitary)
Hypercalcemia
Hypocalcemia
Phosphate disturbances
Rare thyroid hormone signalling disorders
Congenital hypothyroidism and hyperthyroidism and paediatric hyperthyroidism
Thyroid carcinoma (RAI sensitive & non-met medullary)
Hyperinsulinism
Insulin-resistance syndrome
Rare diabetes mellitus
Multiple endocrine neoplasias Type I
Multiple endocrine neoplasias Type II
Carney complex
Hereditary pheochromocytoma/ paraganglioma
Von Hippel Lindau syndrome
Dravet (SCN1A)
Angelmans
Ring Chromosome 20
Rett syndrome (MECP2)
CDKL5
PCDH19
KCNq2
SCN8A
lp36 deletion
FOXG1
Tuberous sclerosis
Lissencephaly
Polymicrigyria
Sturge Weber syndrome
Periventricular Nodular Heterotopia

Subcorticcal band heterotopias
Herpes simplex encephaliatis
CMV encephalitis
Autoimmune encephalitis
Rasmussen encephalitis
FIRES
Anti-NMDA receptor limbic encephalitis
Acute Febrile Epileptics Encephalopathy
Focal cortical dysplasia
Hypothalamic Hamartoma
Hemimegalancephaly
Gangliomgioma
DNET
Cerebral cavernous malformations
MTLE with hippocampal sclerosis
benign familial neonatal seizures
West syndrome
Myocloic astatic epilepsy
Lennox Gastaut syndrome
ESES/C5WS
Migrating partial seizures of infancy LKS
Pyridoxine dependent seizures (ALDH7A1)
GAMT
MOCOD
PO LG
Glucose transporter type 1 deficiency
Biotinidase deficiency
Mitochondrial disease
Neonatal hypoxic and ischemic brain injury
Paediatric arterial ischemic stroke
Acute neonatal citrullinemia type 1
Neonatal glycine encephalopathy
Pyridoxal phosphate-responsive seizures
Pyridoxine-dependent epilepsy
Neonatal epilepsy syndrome
Benign familial neonatal epilepsy
Benign familial neonatal-infantile seizures
Benign idiopathic neonatal seizures
new-onset refractory status epilepticus
Acute encephalopathy with inflammation-mediated status epilepticus
csws
FIRES
Hereditary glomerulopathies
Immune-mediated glomerulopathies

Hereditary tubulopathies
Nephropathy sec. to storage or other metabolic disease
Hemolytic uremic syndrome
ADPKD, tuberous sclerosis complex, ADTKD, VHL
Congenital obstructive nephropathies (post. urethral valve, others)
Renal hypo/dysplasia
Nephronophthisis, ARPKD, Bardet Biedl S., others
Chronic kidney disease stage 3-5 and dialysis
Pediatric transplantation
Retinis Pigmentosa
Leber congenital amaurosis
Syndromic retinitis pigmentosa
Metabolic disease associated with ocular features
Metabolic disease with pigmentary retinitis
Cohen syndrome
Joubert syndrome with oculorenal defect
Usher syndrome
Alström syndrome
Bardet-Biedl syndrome
Choroideremia
Genetic macular dystrophy
Progressive cone dystrophy
Best vitelliform macular dystrophy
Patterned dystrophy of the retinal pigment epithelium
Colobomatous and areolar dystrophy
Cone rod dystrophy
Familial flecked retinopathy
Stargardt disease
Color-vision disease
Achromatopsia
Genetic vitreous-retinal disease
Stickler syndrome
X-linked retinoschisis
Familial exudative vitreoretinopathy
Congenital stationary night blindness
Hereditary optic neuropathy
Leber hereditary optic neuropathy
Autosomal dominant optic atrophy
Autosomal recessive isolated optic atrophy
Syndromic optic nerve hypoplasia
Syndromic hereditary optic neuropathy

Metabolic disease associated with ocular features
Mitochondrial disease with eye involvement
Leigh syndrome
Neuro-ophthalmological disease
Craniostenosis associated with a strabismus
Oculomotor palsy
Supranuclear oculomotor palsy
Nuclear oculomotor paralysis
Oculomotor apraxia or related oculomotor disease
Myopathy with eye involvement
Abnormal eye movements
Nervous system anomaly with eye involvement
Rare acquired eye disease
Balhint syndrom
Holmes-Adie syndrome
Progressive supranuclear palsy syndrome
Atypical Progressive supranuclear palsy syndrome
Classic Progressive supranuclear palsy syndrome
Progressive supranuclear palsy-pure akinesia with gait freezing syndrome
Progressive supranuclear palsy- progressive non-fluent aphasia syndrome
Progressive supranuclear palsy- parkinsonism syndrome
Progressive supranuclear palsy- corticobasal syndrome
Ptosis
Carnevale syndrome
Marcus-Gunn syndrome
Septopreoptic holoprosencephaly
Spastic paraplegia-optic atrophy-neuropathy syndrome
Syndromic hereditary optic neuropathy
Syndromic optic nerve hypoplasia
Treft-Sanborn-Carey syndrome
Neuromyelitis optica
Hereditary glaucoma
Iridogoniodysgenesis
Axenfeld-Rieger syndrome
Aniridia
Syndromic aniridia
Isolated aniridia
Peters anomaly
Congenital glaucoma
Juvenile glaucoma
Developmental defect of the eye

Anophthalmia-microphthalmia syndrome
Syndromic microphthalmia
Ocular coloboma
Septo-optic dysplasia spectrum
Rare palpebral, lacrimal system and conjunctival disease
Rare palpebral disease
Congenital Ptosis
Rare lacrimal system disease
Lens and zonula anomaly
Early-onset non- syndromic cataract
Syndromic cataract
Systemic disease with cataract
Genetic vitreous-retinal disease
Norrie disease
Coats plus syndrome
Vitreoretinal degeneration
X-linked retinoschisis
Familial exudative vitreoretinopathy
Genodermatosis with ocular features
Pigmentation disorder with eye involvement
Oculocutaneous albinism
Ocular albinism
Syndromic oculocutaneous albinism
Pigmentation disorder with eye involvement excluding
Phakomatosis with eye involvement
Connective tissue disease with eye involvement
Marfan syndrome
Metabolic disease associated with ocular features
Mitochondrial disease with eye involvement
Hereditary glaucoma
Juvenile glaucoma
Corneal dystrophy
Superficial corneal dystrophy
Stromal corneal dystrophy
Posterior corneal dystrophy
Syndromic corneal dystrophy
Rare conjunctival disease
Rare refraction anomaly
Rare disease with glaucoma as a major feature
Lens and zonula anomaly
Connective tissue disease with eye involvement

Marfan syndrome
Metabolic disease with corneal opacity
Metabolic disease with cataract
Keratoconus
Syndromic keratoconus
Rare acquired eye disease
Esophageal Atresia
Achalasia
Congenital Esophageal Stenosis
Eosinophilic Esophagitis
Idiopathic achalasia
Moyamoya disease with early onset achalasia
Achalasia- alacrimia syndrome
Intestinal disease due to fat malabsorption
Sacrococcygeal teratoma
Rare Inflammatory Bowel Diseases
Familial Adenomatous Polyposis
Intestinal Polyposis Syndrome
Necrotizing Enterocolitis
Duodenal And Small Bowel Atresia
Intestinal Failure
Congenital Enteropathies
Intractable Diarrhea Of Infancy
Congenital chronic diarrhea with protein-losing enteropathy
Rare diseases involving intestinal motility
Microvillus Inclusion Disease
Short Bowel Syndrome
Epithelial Dysplasia
Cipo - Chronic Intestinal Pseudo-Obstruction
Morbus Hirschsprung
Rare Pancreatic Diseases
Congenital Diaphragmatic Hernia
Congenital Hiatus Hernia
Eventration Of Diaphragm
Omphalocele
Gastroschisis
Familial omphalocele syndrome with facial dysmorphism
Lethal omphalocele-cleft palate syndrome
Omphalocele syndrome, Shprintzen-Goldberg type
Omphalocele-diaphragmatic henia cardiovascular anomalies-radial ray syndrome
Pericardial and diaphagmatic defect

Diaphragmatic defect-limb deficiency-skull defect syndrome
Diaphrgmatic hernia-short bowel-asplenia syndrome
Interstitial lung diseases (ILD)
Cystic fibrosis (CF)
Primary Ciliary Dyskinesia (PCD)
non CF bronchiectasis (nCF BE or NCFB)
Alphal antitrypsin deficiency (AATD)
Chronic lung allograft dysfunction (CLAD)
Other rare lung diseases (ORLD)
Sardoidosis
Cystic Lung Diseases
Disorders of Respiratory Drive
Pulmonary hypertension (PH)
Mesothelioma (MSTO)
Cerebellar Ataxias and Spastic Paraplegias
Choreas and Huntington's Disease
Dystonias, paroxysmal disorders (non-epileptical ones) and Neurodegeneration with Brain Iron Accumulation
Frontotemporal Dementia
Leukodystrophies
Atypical parkinsonian syndromes: Genetic PD, Multisystem Atrophy, Progressive Supranuclear Palsy, Corticobasal degeneration
Rare inflammatory and oncological diseases
Rare vascular diseases, malformation and further rare neurological diseases
Simplex
Junctional - generalized severe (Herlitz)
Distrophyc
Other & unspecified
Ichthyosis
Palmoplantar keratoderma
Darier
Hailey-Hailey
Netherton
ED
ED Hypohidrotic/anhidrotic

ED with PPK (Clouston, Papillon- Lefevre, Desmosomal)
ED with extracutaneous anomalies (Clefts and EEC, ED Cleft/lip/palate, ADULT, Rapp-Hodgkin)
Incontinentia pigmenti (IP)
Unclassified disorders
Hair diseases
Goltz Syndrome
Ehlers-Danlos syndrome (EDS)
Cutis Laxa (CL)
Pseudoxanthoma elasticum (PXE)
Buschke-Ollendorff syndrome (BOS)
Scleroderma-like syndrome
Congenital melanocytic naevus syndrome and other melanocytic naevus syndromes
Schimmelpenning Syndrome Phakomatosis Pigmentokeratotica, Inflammatory Linear Verrucous Epidermal Naevus
Other epidermal naevi
Hypomelanosis of Ito and other hypopigmentary mosaic disorders
Naevoid and whorled hypermelanosis and other hyperpigmentary mosaic disorders
Extensive Dermal Melanocytosis
Proteus Syndrome
PIK3CA related overgrowth syndromes (incl. CLOVES syndrome)
Klippel-Trenaunay-Weber syndrome
Megalencephaly-capillary malformation polymicrogyria syndrome
Capillary malformation congenital
Cutaneous venous malformations
PHACE syndrome
Sturge Weber syndrome
Phakomatosis Pigmentovascularis
Cutaneous Lymphatic malformations and lymphangiomatosis
Cutaneous arteriovenous malformations
Capillary malformation-arterio venous malformation syndrome
Maffucci syndrome
Glomuvenous malformation, segmental
Cutis marmorata telangiectatica congenita
Undiagnosed mosaic paediatric dermatology conditions
Xeroderma pigmentosum
Trichothiodystrophy

Cockayne syndrome / Rothmund- Thomson syndrome
Pemphigus
Bullous Pemphigoid
Mucous Membrane Pempphigoid
Epidermolysis bullosa acquisita
Linear IgA Disease
Dermatitis Herpetiformis
Stevens-Johnson Syndrome and Toxic epidermal necrolysis Syndrome
Hidradenitis suppurativa/acne
PAPA syndrome
PAPASH syndrome
PASH syndrome
PASS syndrome
SAPHO syndrome
Adamantiades-Behçet's disease
Malignant atrophic papulosis (Degos disease)
Cutaneous Porphyria
Hydroa vaccciniform
Actinic prurigo
Solar urticaria
Chronic actinic dermatitis
Unclassified photosensitivity diseases
Albinism
Alopecia Areata
Vitiligo
Lichen (Cutaneous and mucosal lichen)
Cutaneous scleroderma (Morphea and Linear Scleroderma)
Parry- Romberg syndrome
Cutaneous Lupus
Complex Panniculitis
Cutaneous granulomatous diseases
Pyoderma gangrenosums
Pustular psoriasiform eruption
SWEET syndr
Unclassified cutaneous Vasculitis
Werner syndrome
Progeria
Laminopathies related disorders

Rare cutaneous lymphoma
Lymphomatoid Papulosis
Merkel Tumour
Gorlin Syndrome
Dermatofibrosarcoma protuberans
Sebaceous Gland Carcinoma
Atypical Fibroxanthoma
Fibromatosis
Infantile Myofibromatosis
Mucinosis
Lipomatosis
Xantogranuloma
Cutaneous & indolent mastocytosis
Unclassified primary cutaneous tumours
G1 :Sarcoma of the soft tissue, bone and viscerae
G2: Trophoblastic disease, Rare malignant gynecological cancer
G3: Testis and annexes, extragonadal germ cell tumors
G4: Neuroendocrine tumors
G5: Biliary tract, peritoneal cancer & mesothelioma and anal carcinoma
G6 :Thyroid and adrenal cancers
G7: Salivary gland tumors - nasopharyngeal cancer - nasal and sinonasal cancers
G8: Thymoma, mediastinum and pleura, pleural mesothelioma
G9: Rare skin cancers and eyes melanoma
G10 :glial and non glial tumors
Haemoglobinopathy (SCD, Thalassaemia, [other Hbpathies)
Hereditary (RBC) erythroenzymopathies (chronic conditions)

Hereditary RBC membrane defects
Congenital Erythrocytosis
Phenotyping and genotypmg of haemoglobinopathies
Genetic counselling, prenatal diagnosis for haemoglobinopathies
Phenotyping and genotyping of enzymopathies
BMT for laemoglobinopathies, rejection management
Blood transfusion management. Care of mmunised patient
Iron overload assessment: Iron delation
MRI-T2* for iron monitoring
Acute events management for sickle cell disease
Transcranial Echo-Ooppler for stroke/prevention
Assessment of spleen function and splenectomy
Congenital dyserythropoietic anemia
Blackfan-Diamond anemia
Acquired BMF (Aplastic Anaemia and PNH)
inherited BMF (FA, Dyskeratosis congenital, others)
mmunosuppressive treatments
3MT for bone marrow Failures and rejection management
Diagnosis
Haemophilia A and B (including female carriers)
The rarer congenital deficiencies of other coagulation factors
Von Willebrand
Inherited platelet defects
Care for patients with inhibitors, including surgery
Acute events management for Haemophilia A and B
non-HFE related lereditary lemochromatosis
HFE-related hereditary hemochromatosis
Low iron availability or erythropoiesis: RIDA, ACP
Defects in iron acquisition, transport: IT, DMT1, STEAP3
Defects in heme synthesis or Fe-S cluster biogenesis
Complex molecular diagnosis, genetic counselling
Iron overload management: iron chelation
Biochemical and hematology tests
Bone marrow smears for ring sideroblasts
MRI-T2 for iron monitoring
Phlebotomy unit and erythrocytapheresis, if needed

ilood transfusion management, care of mmunized patients
Bone marrow transplant and rejection management
Liver unit facilities biopsy, access to liver ransplant center)
Myelodysplastic syndrome (MDS)
Acute myeloid eukemia (AML)
Chronic myelomonocytic leukemia (CMML)
Chronic Myeloid Leukemia (CML)
Myeloproliferative neoplasm (MPN)
Primary myelofibrosis
Systemic mastocytosis
Access to MDS diagnostic services
Access to MPN diagnostic services
Stem cell transplantation service
Acute care
Phase I/ll studies open or inclusion on MDS
Phase I/ll studies open for inclusion on MPN in the last 3 years)
Phase lll/IV studies open for inclusion on MPN in the last three
Acute lymphoblastic leukemia (ALL)
Marginal zone lymphomas
Light chain Amyloidosis (AL amyloidosis)
Targeted therapies for marginal zone ymphomas
Expert Diagnosis and classification of amyloidosis
Chemotherapy, autologous SCT
Acute care
Phase l/ll studies open or inclusion for ALL
Phase f/ll studies open or inclusion for imyloidosis
Primary CNS lymphoma*primary vitro retinal lymphoma
Hodgkin Lymphoma
Rare Lymphoma Mantle cell lymphoma
Rare Lymphoma Primary cutaneous lymphomas
Rare Lymphoma Virus associated lymphomas
Hairy cell leukemia
Rare Lymphoma follicuiar lymphoma
46 -XX DSD induced by fetal androgrens excess
46 -XY DSD
Turner syndrome
Classic congenital adrenal hyperplasia
Bladder exstrophy
Exstrophy-epispadias complex
Agenesis and aplasia of uterine body
Cloacal exstrophy

Classic homocystinuria, cystine stones
Familial cystic renal disease; all cystic kidney diseases in children
Bartter's syndrome
Cystinosis
Cystinuria
Dent disease
Spina bifida (ITHACA already having it in 2016)
Isolated partial vaginal agenesis
Septate vagina
Vaginal atresia
Congenital primary megaureter
Congenital urachal anomaly
Duplication of urethra
Congenital agenesis of the scrotum
Congenital bilateral absence of vas deferens
Diphallia
Idiopathic isolated micropenis
Penile agenesis
Penoscrotal transposition
Fetal lower urinary tract obstruction
Neurogenic bladder dysfunction
Congenital posterior urethral valves
Posterior and familial hypospadias
High, intermediate and low anorectal malformation
Hirschsprung disease
Anal fistula
VACTERL/VATER
Caudal regression sequence
Adrenogenital syndrome
Rare primary hyperaldosteronism
Vesicointestinal fistula
Vesical fistula, not elsewhere classified
Stress incontinence
Other specified urinary incontinence
Female genital prolapse
Fistulae involving female genital tract
Urethral stricture, urethral fistula
Urethral stricture, urethral fistula
Obstructive and reflex uropathy, retroperitoneal fibrosis, vesicoureteral reflux

From congenital malformations: 1.1 Complex genital reconstructions (DSDs) 1.2 Bladder exstrophy/epispadias 1.4 Non-syndromic urogenital tract malformation 1.5 Posterior urethral valve 1.6 Posterior hypospadias 1.7 Urorectal/anorectal malformations
Rare renal tumors with highly specialized surgery: Wilms Tumour, Neuroblastoma, Rhabdomyosarcoma
Transgender: Male-to-female and female-to-male surgery and surgery for complications
Aquired skeletal muscle diseases
Genetic skeletal
Myasthenia Gravis Other autoimmune NMJ defects (Lambert-Eaton)
Congenital Myasthenic Syndromes
Inherited Neuropathies
Acquired Neuropathies
Amyotrophic Lateral Sclerosis (ALS) and related diseases
Spinal Muscular Atrophy (SMA) and related diseases
Mitochondrial encephalomyopathies caused by mtDNA
Nuclear gene defects causing mitochondrial encephalomyopathy,
Neurofibromatoses type 1
Neurofibromatoses type 2
Schwannomatoses
Lynch syndrome
Polyposis (FAP, AFAP, MAP, PPAP, NAP, JPS, PJS, mixed, SPS)
hereditary breast and ovarian cancer
PTEN hamartoma tumour syndrome
Li-Fraumeni Syndrome
Birt-Hogg-Dubé Syndrome
Familial Malignant Melanoma
Constitutional Mismatch repair deficiency
Rhabdoid Tumor Predisposition Syndrome 2
CDH1 related Hereditary diffuse gastric cancer
Carney Complex
Hereditary Papillary Renal Cell Carcinoma

Ataxia-Telangiectasia
Bloom syndrome
Nevoid basal cell carcinoma syndrome / Gorlin syndrome
Werner Syndrome
Hereditary Leiomyomatosis and Renal Cell Cancer
Von Hippel –Lindau disease
Fanconi anemia
Paraganglioma, Hereditary pheochromocytoma-paraganglioma
Long QT syndrome
Brugada syndrome
Catecholaminergic Polymorphic Ventricular Tachycardia
Short QT syndrome
Early repolarization syndrome
Progressive cardiac conduction diseases
Idiopathic ventricular fibrillation
Unexplained cardiac arrest
Special electrophysiology conditions in children (ablation in children under 10 kg, ICD implants in young children, stellectomy)
Arrhythmogenic Cardiomyopathy
Hypertrophic Cardiomyopathy
Dilated Cardiomyopathy
Restrictive Cardiomyopathy
Unclassified Cardiomyopathy
Congenital malformations: pulmonary and tricuspid valves aortic and mitral valves cardiac septa cardiac chambers and connections others
Sarcoidosis
Amyloidosis
Myocarditis
Pericarditis
others rare cardiac diseases
Multiple congenital anomalies/dysmorphic syndromes without intellectual disability
Multiple congenital anomalies/dysmorphic syndrome, variable intellectual disability
Multiple congenital anomalies/dysmorphic syndrome intellectual disability
Rare intellectual disability with developmental anomaly
Rare intellectual disability, non syndromic
Rare chromosomal anomaly

Rare but isolated congenital malformation not otherwise covered by an ERN
Phenylketonuria, hyperphenylalaninemia
Disorders of tyrosine metabolism
Maple syrup urine disease
Other disorders of branched-chain amino acids including disorders of cobalamin and biotin metabolism
Disorders of lysine and hydroxylysine metabolism
Cerebral organic acidurias
Disorders of sulfur-containing amino acids
Urea cycle disorders and disorders of ornithine metabolism
Glycogen storage diseases
Disorders of galactose metabolism
Disorders of the pentose phosphate pathway
Disorders of fructose metabolism
Persistent hyperinsulinemic hypoglycemia
Disorders of fatty acid oxidation and ketone body metabolism
Gluconeogenesis
LPIN1
Sphingolipidoses
Oligosaccharidoses
Mucopolysaccharidoses
Mucolipidoses
Neuronal Ceroid Lipofuscinoses (CLN1/CLN2)
Lipid storage diseases
Lysosomal transport defects
Lysosoma glycogen storage diseases
Zellweger spectrum
Rhizomelic chondrodysplasia

X-linked adrenoleukodystrophy
Peroxisomal acyl-CoA oxidase
D-bifunctional protein
Refsum disease (classic)
α- Methylacyl-CoA racemase
Ultra-rare, overarching group is most appropriate
Cerebral creatine dificiency
Disorders of purine and pyrimidine
Disorders of minerals
GLUT1 deficiency
Neurotransmitter and pterin disorders
Vitamin related disorders (folate and vitamin B6)
GABA disorders
ALA dehydratase deficiency porphyria
Hereditary Coproporphyria
Brain Tumours
Neuroblastoma
Retinoblastoma
Nephroblastoma
Hepatoblastoma
Ewing sarcoma
Soft tissue sarcoma
Germ tissue sarcoma
Very Rare tumours
Langerhans cell histiocytosis
Primary Biliary Cholangitis (PBC, formerly Primary Biliary Cirrhosis)
Autoimmune Hepatitis (AIH)
Primary Sclerosing Cholangitis (PSC)
IgG4 Disease
Genetic Cholestatic Disease
Biliary Atresia
Choledochal Cyst

Alpha-1 Antitrypsin Liver Disease
Wilsons Disease
Cystic Liver Disease
Vascular Liver Disease
Rare liver tumors
Rare causes of acute liver failure
Intrahepatic cholestasis of pregnancy (ICP)
HELLP syndrome
Acute fatty liver of pregnancy
Systemic sclerosis
Mixed connective tissue diseases
Polymyositis Dermatomyositis Anti-synthetase syndrome
Anti-phospholipid syndrome
Undifferentiated connective tissue diseases
IgG4 related diseases
Polychondritis
Systemic lupus erythematosus
Sjögren syndrome
Ehlers Danlos syndrome
300+rare PIDs
Defects affecting the inflammasome Non inflammasome-related conditions Type 1 Interferonopathies Monogenic Inflammatory bowel diseases Miscellaneous
Systemic vasculitis Autoimmune neuro-inflammatory disease
Juvenile Idiopathic Arthritis (all subtypes) Juvenile Idiopathic Inflammatory Myopathies Juvenile SLE Juvenile scleroderma and overlap syndromes Acute rheumatic fever
Including both SOT or HSCT: waiting list
Paediatric liver transplantation
Paediatric kidney transplantation
Paediatric lung transplantation
Paediatric heart transplantation
Paediatric intestine transplantation (including small bowel)

Multiorgan or combined transplantation (including: liver, kidney, heart, lung, pancreas or HSCT)
Paediatric Haematopoietic Stem Cell Transplantation (HSCT), including Allogeneic bone marrow transplantation, umbilical cord blood transplantation and autologous and allogeneic peripheral blood transplantation.
Follow up and supportive interventions (as PTLD, rejection, tumor, infectious complications, growth, environment, etc.)
Hereditary Haemorrhagic Telangiectasia (HHT)
Marfan Syndrome type 1 and 2
Loeys Dietz Syndromes 1-6
Aneurysm Osteoarthitis Syndrome
Arterial tortuosity syndrome
Familial thoracic Aortic Aneurysms Dissections (FTAA(D)) with mutations in FBN1, TGFB2, TGFB3, SMAD2, SMAD3, TGFBR1, TGFBR2, ACTA2, MYH11, MYLK, PRKG1
FTAA(D) with mutations in FOXE3, MAT2A, MFAP5
Familial Thoracic Aortic Aneurysm and aortic dissection
Multisystemic Smooth Muscle Cell Dysplasia syndrome
Fibulin 4 related cutis laxa
Venous malformations (VMs)
Lymphatic malformations (LMs)
Capillary malformations (CMs)
Arteriovenous malformations (AVMs)
Complex syndromes
Vascular tumors and miscallenous
Lymphedema
Congenital Lymphedema: Milroy syndrome
Late onset lymphedema:
Meige syndrome
Lymphedema distichiasis syndrome, Emberger syndrome
Lymphedema with systemic involvement:
Hennekam syndrome
PIEZO1 related lymphatic dysplasia

Generalised lymphatic dysplasia
Multi-systemic lymphedema with systemic involvement
Syndromes associated with lymphedema:
Noonan/CFC syndrome (RASOpathies)
Turner syndrome
22q13 microdeletion
Microcephaly with or without Chorioretinopathy, Lymphedema and Mental Retardation (MCLMR)
Various types of complex lymphovascular disorders
Vascular Ehlers Danlos Syndrome (vEDS)
Fibromuscular Dysplasia (FMD)
Spontaneous Coronary Artery Dissection (SCAD)

Corresponderende informatie uit de Orphanet database
Orphanetgegevens (Disease Id - naam groep/aandoening - [Typology] - ORPHAcode)
Achondroplasia [Disorder - Disease] ORPHA:15
Hypochondroplasia [Disorder - Disease] ORPHA:429
Léri-Weill dyschondrosteosis [Disorder - Malformation syndrome] ORPHA:240
Acromesomelic dysplasia [Group of disorders - Clinical group] ORPHA:93437
Osteogenesis imperfecta [Disorder - Disease] ORPHA:666
Hypophosphatasia [Disorder - Disease] ORPHA:436
Cleidocranial dysplasia [Disorder - Malformation syndrome] ORPHA:1452
Osteopetrosis and related disorders [Group of disorders - Clinical group] ORPHA:2781
Camurati-Engelmann disease [Disorder - Malformation syndrome] ORPHA:1328
Osteopathia striata-cranial sclerosis syndrome [Disorder - Malformation syndrome] ORPHA:2780
Multiple osteochondromas [Disorder - Disease] ORPHA:321
Ollier disease [Disorder - Disease] ORPHA:296 Maffucci syndrome [Disorder - Disease] ORPHA:163634
Fibrodysplasia ossificans progressiva [Disorder - Disease] ORPHA:337
Fibrous dysplasia/McCune-Albright syndrome [Group of disorders - Clinical group] ORPHA:595216
Type 2 collagen-related bone disorder [Group of disorders - Category] ORPHA:93421
Multiple epiphyseal dysplasia [Group of disorders - Clinical group] ORPHA:251
Metatropic dysplasia [Disorder - Disease] ORPHA:2635 Spondylometaphyseal dysplasia, Kozlowski type [Disorder - Disease] ORPHA:93314
Hypophosphatemic rickets [Group of disorders - Clinical group] ORPHA:437 X-linked hypophosphatemia [Disorder - Disease] ORPHA:89936
Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome [Subtype of disorder - Clinical subtype] ORPHA:306661
Acrodysostosis [Disorder - Malformation syndrome] ORPHA:950
Pseudohypoparathyroidism with Albright hereditary osteodystrophy [Group of disorders - Clinical group] ORPHA:457059
Myhre syndrome [Disorder - Malformation syndrome] ORPHA:2588
Acromicric dysplasia [Disorder - Malformation syndrome] ORPHA:969
Geleophysic dysplasia [Disorder - Malformation syndrome] ORPHA:2623
Jeune syndrome [Disorder - Malformation syndrome] ORPHA:474
Ellis Van Creveld syndrome [Disorder - Malformation syndrome] ORPHA:289
Larsen syndrome [Disorder - Malformation syndrome] ORPHA:503
Desbuquois syndrome [Disorder - Malformation syndrome] ORPHA:1425
Diastrophic dysplasia [Disorder - Disease] ORPHA:628
Sotos syndrome [Disorder - Disease] ORPHA:821 Weaver syndrome [Disorder - Malformation syndrome] ORPHA:3447
Marshall-Smith syndrome [Disorder - Malformation syndrome] ORPHA:561
Poland syndrome [Disorder - Malformation syndrome] ORPHA:2911 Adams-Oliver syndrome [Disorder - Malformation syndrome] ORPHA:974
Ectrodactyly with and without other manifestations [Group of disorders - Category] ORPHA:498477
Isolated craniosynostosis [Group of disorders - Clinical group] ORPHA:139390
Syndromic craniosynostosis [Group of disorders - Category] ORPHA:139393
Frontal encephalocele [Subtype of disorder - Clinical subtype] ORPHA:1931
Basal encephalocele [Subtype of disorder - Clinical subtype] ORPHA:268829

nvt
nvt
Cleidocranial dysplasia [Disorder - Malformation syndrome] ORPHA:1452
Oculo-auriculo-vertebral spectrum [Disorder - Malformation syndrome] ORPHA:141132
Treacher-Collins syndrome [Disorder - Malformation syndrome] ORPHA:861 Nager syndrome [Disorder - Malformation syndrome] ORPHA:245
Postaxial acrofacial dysostosis [Disorder - Malformation syndrome] ORPHA:246
Aplasia cutis congenita [Disorder - Malformation syndrome] ORPHA:1114
Facial cleft [Group of disorders - Category] ORPHA:141229
Frontonasal dysplasia [Group of disorders - Clinical group] ORPHA:250
Isolated hereditary congenital facial paralysis [Disorder - Morphological anomaly] ORPHA:306527
Moebius syndrome [Disorder - Disease] ORPHA:570
Hypoglossia/aglossia [Group of disorders - Category] ORPHA:156212
Macroglossia [Group of disorders - Category] ORPHA:156207
Cleft lip with or without cleft palate [Group of disorders - Clinical group] ORPHA:1991
Cleft palate [Group of disorders - Clinical group] ORPHA:2014
Isolated Pierre Robin syndrome [Disorder - Malformation syndrome] ORPHA:718
Rare disease with Pierre Robin syndrome [Group of disorders - Category] ORPHA:138044
Laryngotracheoesophageal cleft [Disorder - Morphological anomaly] ORPHA:2004
Congenital tracheal stenosis [Disorder - Morphological anomaly] ORPHA:141127
Congenital subglottic stenosis [Disorder - Malformation syndrome] ORPHA:141121
Non-syndromic genetic deafness [Disorder - Disease] ORPHA:87884
Nose and cavum anomaly [Group of disorders - Category] ORPHA:156246
Groep niet in de Orphanet database
Microtia [Disorder - Morphological anomaly] ORPHA:83463
Anotia [Disorder - Morphological anomaly] ORPHA:93976
Oligodontia [Disorder - Morphological anomaly] ORPHA:99798
Dentin dysplasia [Disorder - Disease] ORPHA:1653
Dentinogenesis imperfecta [Disorder - Disease] ORPHA:49042
Amelogenesis imperfecta [Disorder - Disease] ORPHA:88661
Niet in de Orphanet database
Primary failure of tooth eruption [Disorder - Disease] ORPHA:412206
Niet in de Orphanet database
Niet in de Orphanet database
Hereditary pheochromocytoma-paraganglioma [Disorder - Disease] ORPHA:29072 Sporadic pheochromocytoma/secreting paraganglioma [Disorder - Disease] ORPHA:276621
Adrenocortical carcinoma [Disorder - Disease] ORPHA:1501
ACTH-independent Cushing syndrome due to rare cortisol-producing adrenal tumor [Group of disorders - Category] ORPHA:443287
Primary adrenal insufficiency [Group of disorders - Category] ORPHA:101958
Congenital adrenal hyperplasia [Group of disorders - Clinical group] ORPHA:418
Familial hyperaldosteronism [Group of disorders - Clinical group] ORPHA:235936
Disorder of sex development [Group of disorders - Category] ORPHA:90771
Isolated congenital hypogonadotropic hypogonadism [Disorder - Disease] ORPHA:238666
46,XX gonadal dysgenesis [Disorder - Malformation syndrome] ORPHA:243

Niet zeldzaam
Prader-Willi syndrome [Disorder - Disease] ORPHA:739 Prader-Willi-like syndrome [Group of disorders - Clinical group] ORPHA:398073
Silver-Russell syndrome [Disorder - Disease] ORPHA:813
Beckwith-Wiedemann syndrome [Disorder - Malformation syndrome] ORPHA:116
Noonan syndrome [Disorder - Malformation syndrome] ORPHA:648
Growth hormone insensitivity syndrome [Group of disorders - Category] ORPHA:181393
Overgrowth syndrome [Group of disorders - Category] ORPHA:93460
Malformation syndrome with short stature [Group of disorders - Category] ORPHA:139021
Pituitary adenoma [Group of disorders - Clinical group] ORPHA:99408
Non-acquired combined pituitary hormone deficiency [Group of disorders - Category] ORPHA:467
Acquired pituitary hormone deficiency [Group of disorders - Category] ORPHA:95502
nvt
Familial hypocalciuric hypercalcemia [Disorder - Disease] ORPHA:405 Autosomal recessive infantile hypercalcemia [Disorder - Disease] ORPHA:300547
Autosomal dominant hypocalcemia [Subtype of disorder - Clinical subtype] ORPHA:428 Hypocalcemic rickets [Group of disorders - Clinical group] ORPHA:289103
Hypophosphatemic rickets [Group of disorders - Clinical group] ORPHA:437
Rare hypothyroidism [Group of disorders - Category] ORPHA:181396 Rare hyperthyroidism [Group of disorders - Category] ORPHA:181399
Congenital hypothyroidism [Group of disorders - Category] ORPHA:442 Familial hyperthyroidism due to mutations in TSH receptor [Disorder - Disease] ORPHA:424 Pediatric-onset Graves disease [Disorder - Disease] ORPHA:525731
Thyroid carcinoma [Group of disorders - Category] ORPHA:100088
Hyperinsulinemic hypoglycaemia [Group of disorders - Category] ORPHA:443095
Rare insulin-resistance syndrome [Group of disorders - Category] ORPHA:181368
Rare diabetes mellitus [Group of disorders - Category] ORPHA:101952
Multiple endocrine neoplasia type 1 [Disorder - Disease] ORPHA:652
Multiple endocrine neoplasia type 2 [Disorder - Disease] ORPHA:653
Carney complex [Disorder - Disease] ORPHA:1359
Hereditary pheochromocytoma-paraganglioma [Disorder - Disease] ORPHA:29072
Von Hippel-Lindau disease [Disorder - Disease] ORPHA:892
Dravet syndrome [Disorder - Disease] ORPHA:33069
Angelman syndrome [Disorder - Malformation syndrome] ORPHA:72
Ring chromosome 20 syndrome [Disorder - Malformation syndrome] ORPHA:1444
Rett syndrome [Disorder - Disease] ORPHA:778
Early infantile epileptic encephalopathy [Disorder - Clinical syndrome] ORPHA:1934
Female restricted epilepsy with intellectual disability [Disorder - Disease] ORPHA:101039
KCNQ2-related epileptic encephalopathy [Disorder - Disease] ORPHA:439218
Non-specific early-onset epileptic encephalopathy [Disorder - Disease] ORPHA:442835
1p36 deletion syndrome [Disorder - Malformation syndrome] ORPHA:1606
FOXG1 syndrome [Disorder - Disease] ORPHA:561854
Tuberous sclerosis complex [Disorder - Disease] ORPHA:805
Lissencephaly [Group of disorders - Category] ORPHA:48471
Polymicrogyria [Group of disorders - Clinical group] ORPHA:35981
Sturge-Weber syndrome [Disorder - Malformation syndrome] ORPHA:3205
Periventricular nodular heterotopia [Subtype of disorder - Clinical subtype] ORPHA:98892

Subcortical band heterotopia [Disorder - Morphological anomaly] ORPHA:99796
Herpes simplex virus encephalitis [Disorder - Disease] ORPHA:1930
Niet in de Orphanet database
Autoimmune encephalitis [Group of disorders - Clinical group] ORPHA:622014
Rasmussen subacute encephalitis [Disorder - Disease] ORPHA:1929
Febrile infection-related epilepsy syndrome [Disorder - Disease] ORPHA:163703
Limbic encephalitis with NMDA receptor antibodies [Disorder - Disease] ORPHA:217253
Febrile infection-related epilepsy syndrome [Disorder - Disease] ORPHA:163703
Isolated focal cortical dysplasia [Disorder - Disease] ORPHA:65683
Hypothalamic hamartomas with gelastic seizures [Disorder - Disease] ORPHA:86906
Hemimegalencephaly [Disorder - Malformation syndrome] ORPHA:99802
Ganglioglioma [Disorder - Disease] ORPHA:251949
Dysembryoplastic neuroepithelial tumor [Disorder - Disease] ORPHA:251946
Familial cerebral cavernous malformation [Disorder - Malformation syndrome] ORPHA:221061
Mesial temporal lobe epilepsy with hippocampal sclerosis [Disorder - Disease] ORPHA:99701
Benign familial neonatal epilepsy [Disorder - Disease] ORPHA:1949
West syndrome [Disorder - Clinical syndrome] ORPHA:3451
Myoclonic-astastic epilepsy [Disorder - Disease] ORPHA:1942
Lennox-Gastaut syndrome [Disorder - Disease] ORPHA:2382
Continuous spikes and waves during sleep [Disorder - Disease] ORPHA:725
Malignant migrating focal seizures of infancy [Disorder - Disease] ORPHA:293181
Pyridoxine-dependent epilepsy [Disorder - Disease] ORPHA:3006
Guanidinoacetate methyltransferase deficiency [Disorder - Disease] ORPHA:382
Sulfite oxidase deficiency due to molybdenum cofactor deficiency [Subtype of disorder - Clinical subtype] ORPHA:99732
Alpers-Huttenlocher syndrome [Disorder - Disease] ORPHA:726
Classic glucose transporter type 1 deficiency syndrome [Disorder - Disease] ORPHA:71277
Biotinidase deficiency [Disorder - Disease] ORPHA:79241
Mitochondrial disease with epilepsy [Group of disorders - Category] ORPHA:225700
Neonatal hypoxic and ischemic brain injury [Disorder - Particular clinical situation in a disease or syndrome] ORPHA:137577
Pediatric arterial ischemic stroke [Disorder - Clinical syndrome] ORPHA:439175
Acute neonatal citrullinemia type I [Subtype of disorder - Clinical subtype] ORPHA:247546
Neonatal glycine encephalopathy [Subtype of disorder - Clinical subtype] ORPHA:289857
Pyridoxal phosphate-responsive seizures [Disorder - Disease] ORPHA:79096
Pyridoxine-dependent epilepsy [Disorder - Disease] ORPHA:3006
Neonatal epilepsy syndrome [Group of disorders - Category] ORPHA:98257
Benign familial neonatal epilepsy [Disorder - Disease] ORPHA:1949
Benign familial neonatal-infantile seizures [Disorder - Disease] ORPHA:140927
Benign idiopathic neonatal seizures [Disorder - Disease] ORPHA:64545
New-onset refractory status epilepticus [Disorder - Disease] ORPHA:363558
Acute encephalopathy with inflammation-mediated status epilepticus [Group of disorders - Clinical group] ORPHA:363567
Continuous spikes and waves during sleep [Disorder - Disease] ORPHA:725
Febrile infection-related epilepsy syndrome [Disorder - Disease] ORPHA:163703
Genetic glomerular disease [Group of disorders - Category] ORPHA:183586
C3 glomerulopathy [Subtype of disorder - Clinical subtype] ORPHA:329918

Genetic renal tubular disease [Group of disorders - Category] ORPHA:183592
Nephropathy secondary to a storage or other metabolic disease [Group of disorders - Category] ORPHA:93593
Hemolytic uremic syndrome [Group of disorders - Clinical group] ORPHA:544458
Autosomal dominant polycystic kidney disease [Disorder - Disease] ORPHA:730 Tuberous sclerosis complex [Disorder - Disease] ORPHA:805 Autosomal dominant tubulointerstitial kidney disease [Disorder - Disease] ORPHA:34149 Von Hippel-Lindau disease [Disorder - Disease] ORPHA:892
Fetal lower urinary tract obstruction [Group of disorders - Clinical group] ORPHA:435365
Renal hypoplasia [Disorder - Morphological anomaly] ORPHA:93101 Renal dysplasia [Disorder - Morphological anomaly] ORPHA:93108
Nephropathy-associated ciliopathy [Group of disorders - Category] ORPHA:156162 Autosomal recessive polycystic kidney disease [Disorder - Disease] ORPHA:731
nvt
nvt
Retinitis pigmentosa [Disorder - Disease] ORPHA:791
Leber congenital amaurosis [Disorder - Disease] ORPHA:65
Syndromic rod-cone dystrophy [Group of disorders - Category] ORPHA:98661
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Cohen syndrome [Disorder - Malformation syndrome] ORPHA:193
Joubert syndrome with oculorenal defect [Disorder - Malformation syndrome] ORPHA:2318
Usher syndrome [Disorder - Disease] ORPHA:886
Alström syndrome [Disorder - Disease] ORPHA:64
Bardet-Biedl syndrome [Disorder - Disease] ORPHA:110
Choroideremia [Disorder - Disease] ORPHA:180
Isolated macular dystrophy [Group of disorders - Category] ORPHA:519302 Syndromic macular dystrophy [Group of disorders - Category] ORPHA:519323
Progressive cone dystrophy [Disorder - Disease] ORPHA:1871
Best vitelliform macular dystrophy [Disorder - Disease] ORPHA:1243
Pattern dystrophy [Group of disorders - Category] ORPHA:63454
Niet meer in de Orphanet database
Cone rod dystrophy [Disorder - Disease] ORPHA:1872
Familial benign flecked retina [Disorder - Disease] ORPHA:363989
Stargardt disease [Disorder - Disease] ORPHA:827
Color-vision disease [Group of disorders - Category] ORPHA:98658
Achromatopsia [Disorder - Disease] ORPHA:49382
Niet meer in de Orphanet database
Stickler syndrome [Disorder - Disease] ORPHA:828
X-linked retinoschisis [Disorder - Malformation syndrome] ORPHA:792
Familial exudative vitreoretinopathy [Disorder - Disease] ORPHA:891
Congenital stationary night blindness [Disorder - Disease] ORPHA:215
Hereditary optic neuropathy [Group of disorders - Category] ORPHA:98671
Leber hereditary optic neuropathy [Disorder - Disease] ORPHA:104
Autosomal dominant optic atrophy [Group of disorders - Clinical group] ORPHA:98672
Autosomal recessive isolated optic atrophy [Disorder - Disease] ORPHA:98676
Syndromic optic nerve hypoplasia [Group of disorders - Category] ORPHA:137905
Syndromic hereditary optic neuropathy [Group of disorders - Category] ORPHA:441434

Niet meer in de Orphanet database
Niet meer in de Orphanet database
Leigh syndrome [Group of disorders - Clinical group] ORPHA:506
Neuro-ophthalmological disease [Group of disorders - Category] ORPHA:140653
Craniostenosis with strabismus [Group of disorders - Category] ORPHA:98684
Rare oculomotor nerve disorder [Group of disorders - Category] ORPHA:98685
Supranuclear eye movement disorder [Group of disorders - Category] ORPHA:98687
Niet meer in de Orphanet database
Oculomotor apraxia [Group of disorders - Category] ORPHA:98688
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Balint syndrome [Disorder - Disease] ORPHA:363746
Holmes-Adie syndrome [Disorder - Disease] ORPHA:454718
Progressive supranuclear palsy [Disorder - Disease] ORPHA:683
Atypical progressive supranuclear palsy syndrome [Subtype of disorder - Clinical subtype] ORPHA:99750
Classic progressive supranuclear palsy syndrome [Subtype of disorder - Clinical subtype] ORPHA:240071
Progressive supranuclear palsy-pure akinesia with gait freezing syndrome [Subtype of disorder - Clinical subtype] ORPHA:240094
Progressive supranuclear palsy-progressive non-fluent aphasia syndrome [Subtype of disorder - Clinical subtype] ORPHA:240112
Progressive supranuclear palsy-parkinsonism syndrome [Subtype of disorder - Clinical subtype] ORPHA:240085
Progressive supranuclear palsy-corticobasal syndrome [Subtype of disorder - Clinical subtype] ORPHA:240103
Rare disorder with ptosis [Group of disorders - Category] ORPHA:98578
3MC syndrome [Disorder - Malformation syndrome] ORPHA:293843
Marcus-Gunn syndrome [Disorder - Disease] ORPHA:91412
Septopreoptic holoprosencephaly [Subtype of disorder - Clinical subtype] ORPHA:280195
Spastic paraplegia-optic atrophy-neuropathy syndrome [Disorder - Disease] ORPHA:320406
Syndromic hereditary optic neuropathy [Group of disorders - Category] ORPHA:441434
Syndromic optic nerve hypoplasia [Group of disorders - Category] ORPHA:137905
Autosomal dominant optic atrophy plus syndrome [Disorder - Disease] ORPHA:1215
Neuromyelitis optica [Disorder - Disease] ORPHA:71211
Pediatric-onset glaucoma of genetic origin [Group of disorders - Category] ORPHA:359
Anterior segment developmental anomaly without extraocular manifestations [Group of disorders - Category] ORPHA:98634
Axenfeld-Rieger syndrome [Disorder - Malformation syndrome] ORPHA:782
Niet meer in de Orphanet database
Syndromic aniridia [Group of disorders - Category] ORPHA:98557
Isolated aniridia [Disorder - Morphological anomaly] ORPHA:250923
Peters anomaly [Disorder - Morphological anomaly] ORPHA:708
Congenital glaucoma [Disorder - Disease] ORPHA:98976
Juvenile glaucoma [Disorder - Disease] ORPHA:98977
Developmental defect of the eye [Group of disorders - Category] ORPHA:98553

Microphthalmia-anophthalmia-coloboma [Group of disorders - Category] ORPHA:98555
Syndromic microphthalmia-anophthalmia-coloboma [Group of disorders - Category] ORPHA:202948
Coloboma of choroid and retina [Disorder - Morphological anomaly] ORPHA:98942 Coloboma of eye lens [Disorder - Morphological anomaly] ORPHA:98943 Coloboma of iris [Disorder - Morphological anomaly] ORPHA:98944 Coloboma of macula [Disorder - Morphological anomaly] ORPHA:98945 Coloboma of eyelid [Disorder - Morphological anomaly] ORPHA:98946 Coloboma of optic disc [Disorder - Morphological anomaly] ORPHA:98947
Septo-optic dysplasia spectrum [Disorder - Malformation syndrome] ORPHA:3157
Rare disorder of the ocular adnexa [Group of disorders - Category] ORPHA:519266 Rare disorder with conjunctival involvement as a major feature [Group of disorders - Category] ORPHA:98610
Rare palpebral disorder [Group of disorders - Category] ORPHA:98560
Congenital ptosis [Disorder - Disease] ORPHA:91411
Rare disorder of the lacrimal apparatus [Group of disorders - Category] ORPHA:98602
Rare lens disease [Group of disorders - Category] ORPHA:98639
Early-onset non-syndromic cataract [Disorder - Disease] ORPHA:91492
Syndromic cataract [Group of disorders - Category] ORPHA:98641
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Norrie disease [Disorder - Malformation syndrome] ORPHA:649
Coats plus syndrome [Disorder - Disease] ORPHA:313838
Vitreoretinopathy [Group of disorders - Category] ORPHA:98668
X-linked retinoschisis [Disorder - Malformation syndrome] ORPHA:792
Familial exudative vitreoretinopathy [Disorder - Disease] ORPHA:891
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Oculocutaneous albinism [Group of disorders - Clinical group] ORPHA:55
Ocular albinism [Group of disorders - Clinical group] ORPHA:284804
Syndromic oculocutaneous albinism [Group of disorders - Category] ORPHA:284811
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Marfan syndrome [Disorder - Disease] ORPHA:558
Niet meer in de Orphanet database
Niet meer in de Orphanet database
Pediatric-onset glaucoma of genetic origin [Group of disorders - Category] ORPHA:359
Juvenile glaucoma [Disorder - Disease] ORPHA:98977
Corneal dystrophy [Group of disorders - Category] ORPHA:34533
Superficial corneal dystrophy [Group of disorders - Category] ORPHA:98625
Stromal corneal dystrophy [Group of disorders - Category] ORPHA:98626
Posterior corneal dystrophy [Group of disorders - Category] ORPHA:98627
Syndromic corneal dystrophy [Group of disorders - Category] ORPHA:98628
Rare disorder with conjunctival involvement as a major feature [Group of disorders - Category] ORPHA:98610
Rare refraction anomaly [Group of disorders - Category] ORPHA:98618
Rare disease with glaucoma as a major feature [Group of disorders - Category] ORPHA:98638
Rare lens disease [Group of disorders - Category] ORPHA:98639
Niet meer in de Orphanet database

Marfan syndrome [Disorder - Disease] ORPHA:558
Niet meer in de Orphanet database
Door wijzigingen in de classificaties is erkenning voor deze groep niet meer aan te vragen
Niet meer in de Orphanet database
Syndromic keratoconus [Group of disorders - Category] ORPHA:98623
Niet meer in de Orphanet database
Esophageal atresia [Disorder - Morphological anomaly] ORPHA:1199
Idiopathic achalasia [Disorder - Disease] ORPHA:930
Niet in de Orphanet database
Niet meer zeldzaam
Idiopathic achalasia [Disorder - Disease] ORPHA:930
Moyamoya disease with early-onset achalasia [Disorder - Disease] ORPHA:401945
Triple A syndrome [Disorder - Disease] ORPHA:869
Intestinal disease due to fat malabsorption [Group of disorders - Category] ORPHA:104005
Sacrococcygeal teratoma [Subtype of disorder - Clinical subtype] ORPHA:494421
Rare inflammatory bowel disease [Group of disorders - Category] ORPHA:104012
Familial adenomatous polyposis [Disorder - Disease] ORPHA:733
Intestinal polyposis syndrome [Group of disorders - Clinical group] ORPHA:104010
Necrotizing enterocolitis [Disorder - Disease] ORPHA:391673
Duodenal atresia [Disorder - Morphological anomaly] ORPHA:1203 Atresia of small intestine [Disorder - Morphological anomaly] ORPHA:1201
Chronic intestinal failure [Disorder - Clinical syndrome] ORPHA:294422
Congenital intestinal transport defect [Group of disorders - Category] ORPHA:104003 Congenital intestinal disease due to an enzymatic defect [Group of disorders - Category] ORPHA:104006 Congenital enteropathy involving intestinal mucosa development [Group of disorders - Category] ORPHA:104007
Intractable diarrhea of infancy [Group of disorders - Category] ORPHA:73014
Congenital chronic diarrhea with protein-losing enteropathy [Disorder - Disease] ORPHA:329242
Rare disease involving intestinal motility [Group of disorders - Category] ORPHA:104009
Microvillus inclusion disease [Disorder - Disease] ORPHA:2290
Short bowel syndrome [Group of disorders - Clinical group] ORPHA:104008
Congenital tufting enteropathy [Disorder - Disease] ORPHA:92050
Chronic intestinal pseudoobstruction [Disorder - Clinical syndrome] ORPHA:2978
Hirschsprung disease [Disorder - Disease] ORPHA:388
Rare pancreatic disease [Group of disorders - Category] ORPHA:101937
Congenital diaphragmatic hernia [Disorder - Morphological anomaly] ORPHA:2140
Niet in de Orphanet database
Niet in de Orphanet database
Omphalocele [Disorder - Morphological anomaly] ORPHA:660
Gastroschisis [Disorder - Morphological anomaly] ORPHA:2368
Familial omphalocele syndrome with facial dysmorphism [Disorder - Malformation syndrome] ORPHA:280403
Lethal omphalocele-cleft palate syndrome [Disorder - Malformation syndrome] ORPHA:2736
Omphalocele syndrome, Shprintzen-Goldberg type [Disorder - Malformation syndrome] ORPHA:3164
Omphalocele-diaphragmatic hernia-cardiovascular anomalies-radial ray defect syndrome [Disorder - Malformation syndrome] ORPHA:496693
Pericardial and diaphragmatic defect [Disorder - Malformation syndrome] ORPHA:2847

Diaphragmatic defect-limb deficiency-skull defect syndrome [Disorder - Malformation syndrome] ORPHA:2141
Diaphragmatic hernia-short bowel-asplenia syndrome [Disorder - Malformation syndrome] ORPHA:527468
Interstitial lung disease [Group of disorders - Category] ORPHA:182095
Cystic fibrosis [Disorder - Disease] ORPHA:586
Primary ciliary dyskinesia [Disorder - Disease] ORPHA:244
Idiopathic bronchiectasis [Disorder - Disease] ORPHA:60033 Allergic bronchopulmonary aspergillosis [Disorder - Disease] ORPHA:1164 Hyper-IgE syndrome [Group of disorders - Clinical group] ORPHA:331223
Alpha-1-antitrypsin deficiency [Disorder - Disease] ORPHA:60
Bronchiolitis obliterans with obstructive pulmonary disease [Disorder - Disease] ORPHA:1303
Congenital alveolar capillary dysplasia [Disorder - Disease] ORPHA:210122 Ondine syndrome [Disorder - Disease] ORPHA:661
Sarcoidosis [Disorder - Disease] ORPHA:797
Lymphangioleiomyomatosis [Disorder - Disease] ORPHA:538
Ondine syndrome [Disorder - Disease] ORPHA:661
Rare pulmonary hypertension [Group of disorders - Category] ORPHA:71198
Pleural mesothelioma [Disorder - Disease] ORPHA:50251
Rare ataxia [Group of disorders - Category] ORPHA:102002 Hereditary spastic paraplegia [Group of disorders - Clinical group] ORPHA:685
Rare choreic movement disorder [Group of disorders - Category] ORPHA:306715
Rare dystonia [Group of disorders - Category] ORPHA:68363 Rare paroxysmal movement disorder [Group of disorders - Category] ORPHA:306768 Neurodegeneration with brain iron accumulation [Group of disorders - Clinical group] ORPHA:385
Frontotemporal dementia [Group of disorders - Clinical group] ORPHA:282
Leukodystrophy [Group of disorders - Category] ORPHA:68356
Rare genetic parkinsonian disorder [Group of disorders - Category] ORPHA:307052 Multiple system atrophy [Disorder - Disease] ORPHA:102 Progressive supranuclear palsy [Disorder - Disease] ORPHA:683 Corticobasal syndrome [Disorder - Disease] ORPHA:454887
Rare neuroinflammatory or neuroimmunological disease [Group of disorders - Category] ORPHA:182064 Rare nervous system tumor [Group of disorders - Category] ORPHA:98062
Rare central nervous system and retinal vascular disease [Group of disorders - Category] ORPHA:71281 Central nervous system malformation [Group of disorders - Category] ORPHA:98044
Epidermolysis bullosa simplex [Group of disorders - Clinical group] ORPHA:304
Severe generalized junctional epidermolysis bullosa [Disorder - Disease] ORPHA:79404
Dystrophic epidermolysis bullosa [Group of disorders - Clinical group] ORPHA:303
Inherited epidermolysis bullosa [Group of disorders - Category] ORPHA:79361
Ichthyosis [Group of disorders - Category] ORPHA:79354
Hereditary palmoplantar keratoderma [Group of disorders - Category] ORPHA:79357
Darier disease [Disorder - Disease] ORPHA:218
Familial benign chronic pemphigus [Disorder - Disease] ORPHA:2841
Netherton syndrome [Disorder - Disease] ORPHA:634
Ectodermal dysplasia syndrome [Group of disorders - Category] ORPHA:79373
Hypohidrotic ectodermal dysplasia [Disorder - Disease] ORPHA:238468 Schöpf-Schulz-Passarge syndrome [Disorder - Disease] ORPHA:50944

Hidrotic ectodermal dysplasia [Disorder - Disease] ORPHA:189 Papillon-Lefèvre syndrome [Disorder - Disease] ORPHA:678 Naxos disease [Disorder - Disease] ORPHA:34217
Cleft lip/palate-ectodermal dysplasia syndrome [Disorder - Malformation syndrome] ORPHA:3253 ADULT syndrome [Disorder - Malformation syndrome] ORPHA:978 EEC syndrome [Disorder - Malformation syndrome] ORPHA:1896 Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome [Disorder - Malformation syndrome] ORPHA:1071
Incontinentia pigmenti [Disorder - Malformation syndrome] ORPHA:464
Unclassified genetic skin disorder [Group of disorders - Category] ORPHA:79385
Hair anomaly [Group of disorders - Category] ORPHA:79363
Focal dermal hypoplasia [Disorder - Malformation syndrome] ORPHA:2092
Ehlers-Danlos syndrome [Group of disorders - Clinical group] ORPHA:98249
Cutis laxa [Group of disorders - Clinical group] ORPHA:209
Pseudoxanthoma elasticum [Disorder - Disease] ORPHA:758
Buschke-Ollendorff syndrome [Disorder - Malformation syndrome] ORPHA:1306
Niet in de Orphanet database
Large congenital melanocytic nevus [Disorder - Disease] ORPHA:626
Linear nevus sebaceus syndrome [Disorder - Disease] ORPHA:2612 Phakomatosis pigmentokeratotica [Disorder - Malformation syndrome] ORPHA:2874 Inflammatory linear verrucous epidermal nevus [Subtype of disorder - Clinical subtype] ORPHA:79466
Woolly hair nevus [Disorder - Disease] ORPHA:79414
Hypopigmentation of the skin [Group of disorders - Category] ORPHA:79376
Linear and whorled nevoid hypermelanosis [Disorder - Disease] ORPHA:79150 Hyperpigmentation of the skin [Group of disorders - Category] ORPHA:79375
Niet in de Orphanet database
Proteus syndrome [Disorder - Malformation syndrome] ORPHA:744
PIK3CA-related overgrowth syndrome [Group of disorders - Clinical group] ORPHA:530313
Angioosteohypertrophic syndrome [Disorder - Disease] ORPHA:2346
Megalencephaly-capillary malformation-polymicrogyria syndrome [Disorder - Malformation syndrome] ORPHA:60040
Rare capillary malformation [Group of disorders - Category] ORPHA:211247
Rare venous malformation [Group of disorders - Category] ORPHA:211252
PHACE syndrome [Disorder - Malformation syndrome] ORPHA:42775
Sturge-Weber syndrome [Disorder - Malformation syndrome] ORPHA:3205
Phakomatosis pigmentovascularis [Disorder - Disease] ORPHA:2875
Rare lymphatic malformation [Group of disorders - Category] ORPHA:2415
Rare arteriovenous malformation [Group of disorders - Category] ORPHA:211266
Capillary malformation-arteriovenous malformation [Disorder - Malformation syndrome] ORPHA:137667
Maffucci syndrome [Disorder - Disease] ORPHA:163634
Glomuvenous malformation [Disorder - Malformation syndrome] ORPHA:83454
Cutis marmorata telangiectatica congenita [Disorder - Malformation syndrome] ORPHA:1556
nvt
Xeroderma pigmentosum [Disorder - Disease] ORPHA:910
Trichothiodystrophy [Disorder - Disease] ORPHA:33364

Cockayne syndrome [Disorder - Disease] ORPHA:191 Rothmund-Thomson syndrome [Disorder - Disease] ORPHA:2909
Pemphigus vulgaris [Disorder - Disease] ORPHA:704
Bullous pemphigoid [Disorder - Disease] ORPHA:703
Mucous membrane pemphigoid [Disorder - Disease] ORPHA:46486
Epidermolysis bullosa acquisita [Disorder - Disease] ORPHA:46487
Linear IgA dermatosis [Disorder - Disease] ORPHA:46488
Dermatitis herpetiformis [Disorder - Disease] ORPHA:1656
Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum [Disorder - Disease] ORPHA:95455
Niet zeldzaam
Pyogenic arthritis-pyoderma gangrenosum-acne syndrome [Disorder - Disease] ORPHA:69126
Niet in de Orphanet database
Pyoderma gangrenosum-acne-suppurative hidradenitis syndrome [Disorder - Disease] ORPHA:289478
Niet in de Orphanet database
SAPHO syndrome [Disorder - Disease] ORPHA:793
Behçet disease [Disorder - Disease] ORPHA:117
Malignant atrophic papulosis [Disorder - Disease] ORPHA:679
nvt
Hydroa vacciniforme [Disorder - Disease] ORPHA:330058
Actinic prurigo [Disorder - Disease] ORPHA:330061
Solar urticaria [Disorder - Disease] ORPHA:97230
Chronic actinic dermatitis [Disorder - Disease] ORPHA:330064
nvt
Hypopigmentation of the skin [Group of disorders - Category] ORPHA:79376
Alopecia [Group of disorders - Category] ORPHA:79364
Niet in de Orphanet database
Rare lichen planus [Group of disorders - Category] ORPHA:254367
Localized scleroderma [Disorder - Disease] ORPHA:90289
Progressive hemifacial atrophy [Disorder - Disease] ORPHA:1214
Rare cutaneous lupus erythematosus [Group of disorders - Clinical group] ORPHA:535
Cytophagic histiocytic panniculitis [Disorder - Disease] ORPHA:94087 IgG4-related mesenteritis [Disorder - Disease] ORPHA:238593 Infantile onset panniculitis with uveitis and systemic granulomatosis [Disorder - Disease] ORPHA:251304 Infantile-onset periodic fever-panniculitis-dermatosis syndrome [Disorder - Disease] ORPHA:500062 Lupus erythematosus panniculitis [Disorder - Disease] ORPHA:90285 Nodular non-suppurative panniculitis [Disorder - Disease] ORPHA:33577 Panniculitis-induced localized lipodystrophy [Disorder - Disease] ORPHA:90159 Subcutaneous panniculitis-like T-cell lymphoma [Disorder - Disease] ORPHA:86884
Groep niet in de Orphanet database
Pyoderma gangrenosum [Disorder - Disease] ORPHA:48104
Generalized pustular psoriasis [Disorder - Disease] ORPHA:247353
Sweet syndrome [Disorder - Disease] ORPHA:3243
nvt
Werner syndrome [Disorder - Disease] ORPHA:902
Hutchinson-Gilford progeria syndrome [Disorder - Disease] ORPHA:740
Laminopathy with premature aging [Group of disorders - Category] ORPHA:300766

Primary cutaneous lymphoma [Group of disorders - Category] ORPHA:542
Lymphomatoid papulosis [Disorder - Disease] ORPHA:98842
Cutaneous neuroendocrine carcinoma [Disorder - Disease] ORPHA:79140
Gorlin syndrome [Disorder - Malformation syndrome] ORPHA:377
Dermatofibrosarcoma protuberans [Disorder - Disease] ORPHA:31112
Niet in de Orphanet database
Niet in de Orphanet database
Superficial fibromatosis [Group of disorders - Clinical group] ORPHA:199257
Infantile myofibromatosis [Disorder - Disease] ORPHA:2591
Localized lichen myxedematosus [Group of disorders - Clinical group] ORPHA:86795
Multiple symmetric lipomatosis [Disorder - Disease] ORPHA:2398 Encephalocraniocutaneous lipomatosis [Disorder - Disease] ORPHA:2396 Roch-Leri mesosomatous lipomatosis [Disorder - Disease] ORPHA:529 Adiposis dolorosa [Disorder - Disease] ORPHA:36397 Familial multiple lipomatosis [Disorder - Disease] ORPHA:199276 Familial angiolipomatosis [Disorder - Disease] ORPHA:199279 Hemihyperplasia-multiple lipomatosis syndrome [Disorder - Malformation syndrome] ORPHA:276280
Juvenile xanthogranuloma [Disorder - Disease] ORPHA:158000 Necrobiotic xanthogranuloma [Disorder - Disease] ORPHA:158011
Cutaneous mastocytosis [Group of disorders - Clinical group] ORPHA:66646 Indolent systemic mastocytosis [Disorder - Disease] ORPHA:98848
nvt
Soft tissue sarcoma [Group of disorders - Clinical group] ORPHA:3394 Bone sarcoma [Group of disorders - Clinical group] ORPHA:223727
Gestational trophoblastic disease [Group of disorders - Category] ORPHA:254685 Rare gynecological tumor [Group of disorders - Category] ORPHA:98063
Tumor of testis and paratestis [Group of disorders - Category] ORPHA:363472 Extragonadal germ cell tumor [Group of disorders - Category] ORPHA:363579
Neuroendocrine neoplasm [Group of disorders - Category] ORPHA:877
Rare tumor of gallbladder and extrahepatic biliary tract [Group of disorders - Category] ORPHA:306633 Primary peritoneal tumor [Group of disorders - Category] ORPHA:168803 Carcinoma of the anal canal [Group of disorders - Clinical group] ORPHA:424013
Thyroid carcinoma [Group of disorders - Category] ORPHA:100088 Adrenal/paraganglial tumor [Group of disorders - Category] ORPHA:100091
Rare tumor of salivary glands [Group of disorders - Category] ORPHA:276142 Nasopharyngeal carcinoma [Disorder - Disease] ORPHA:150 Nasopharyngeal teratoma [Subtype of disorder - Clinical subtype] ORPHA:141107 Nasal glial heterotopia [Disorder - Disease] ORPHA:141112 Nasal ganglioglioma [Subtype of disorder - Clinical subtype] ORPHA:141115 Juvenile nasopharyngeal angiofibroma [Disorder - Disease] ORPHA:289596 Squamous cell carcinoma of the nasal cavity and paranasal sinuses [Disorder - Disease] ORPHA:500464
Thymoma [Disorder - Disease] ORPHA:99867 Pleural mesothelioma [Disorder - Disease] ORPHA:50251
Rare skin tumor or hamartoma [Group of disorders - Category] ORPHA:79386 Uveal melanoma [Disorder - Disease] ORPHA:39044
Glial tumor [Group of disorders - Clinical group] ORPHA:182067 Rare tumor of neuroepithelial tissue [Group of disorders - Category] ORPHA:251558
Hemoglobinopathy [Group of disorders - Category] ORPHA:68364
Rare constitutional hemolytic anemia due to an enzyme disorder [Group of disorders - Category] ORPHA:98369

Rare constitutional hemolytic anemia due to a red cell membrane anomaly [Group of disorders - Category] ORPHA:98364
Primary familial polycythemia [Disorder - Disease] ORPHA:90042 Congenital secondary polycythemia [Group of disorders - Category] ORPHA:238536
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nvt
nvt
nvt
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nvt
Congenital dyserythropoietic anemia [Group of disorders - Clinical group] ORPHA:85
Blackfan-Diamond anemia [Disorder - Disease] ORPHA:124
Rare acquired aplastic anemia [Group of disorders - Category] ORPHA:164823
Rare constitutional aplastic anemia [Group of disorders - Category] ORPHA:68383
nvt
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nvt
Hemophilia [Group of disorders - Clinical group] ORPHA:448
Rare hemorrhagic disorder due to a constitutional coagulation factors defect [Group of disorders - Category] ORPHA:68334
Von Willebrand disease [Disorder - Disease] ORPHA:903
Rare hemorrhagic disorder due to a constitutional platelet anomaly [Group of disorders - Category] ORPHA:71202
nvt
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Hemochromatosis type 2 [Disorder - Disease] ORPHA:79230 Hemochromatosis type 4 [Disorder - Disease] ORPHA:139491 Hemochromatosis type 3 [Disorder - Disease] ORPHA:225123
Symptomatic form of hemochromatosis type 1 [Disorder - Disease] ORPHA:465508
IRIDA syndrome [Disorder - Disease] ORPHA:209981 Aceruloplasminemia [Disorder - Disease] ORPHA:48818
Congenital atransferrinemia [Disorder - Disease] ORPHA:1195 Microcytic anemia with liver iron overload [Disorder - Disease] ORPHA:83642 Severe congenital hypochromic anemia with ringed sideroblasts [Disorder - Disease] ORPHA:300298
Constitutional sideroblastic anemia [Group of disorders - Category] ORPHA:98362
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Myelodysplastic syndrome [Group of disorders - Clinical group] ORPHA:52688
Acute myeloid leukemia [Group of disorders - Clinical group] ORPHA:519
Chronic myelomonocytic leukemia [Disorder - Disease] ORPHA:98823
Chronic myeloid leukemia [Disorder - Disease] ORPHA:521
Myeloproliferative neoplasm [Group of disorders - Clinical group] ORPHA:98274
Primary myelofibrosis [Disorder - Disease] ORPHA:824
Systemic mastocytosis [Group of disorders - Clinical group] ORPHA:2467
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Acute lymphoblastic leukemia [Group of disorders - Clinical group] ORPHA:513
Marginal zone lymphoma [Group of disorders - Clinical group] ORPHA:300912
AL amyloidosis [Disorder - Disease] ORPHA:85443
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Primary central nervous system lymphoma [Disorder - Disease] ORPHA:46135
Hodgkin lymphoma [Group of disorders - Clinical group] ORPHA:98293
Mantle cell lymphoma [Disorder - Disease] ORPHA:52416
Primary cutaneous lymphoma [Group of disorders - Category] ORPHA:542
Epstein-Barr virus-associated malignant lymphoproliferative disorder [Group of disorders - Category] ORPHA:289644 Adult T-cell leukemia/lymphoma [Disorder - Disease] ORPHA:86875 Primary effusion lymphoma [Disorder - Disease] ORPHA:48686
Classic hairy cell leukemia [Disorder - Disease] ORPHA:58017
Follicular lymphoma [Disorder - Disease] ORPHA:545
46,XX disorder of sex development induced by fetal androgens excess [Group of disorders - Category] ORPHA:90776
46,XY disorder of sex development [Group of disorders - Category] ORPHA:98085
44 Turner syndrome [Disorder - Malformation syndrome] ORPHA:881
Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency [Disorder - Disease] ORPHA:90794
Bladder exstrophy [Subtype of disorder - Clinical subtype] ORPHA:93930
Exstrophy-epispadias complex [Disorder - Malformation syndrome] ORPHA:322
Absence of uterine body [Disorder - Morphological anomaly] ORPHA:180142
Cloacal exstrophy [Subtype of disorder - Clinical subtype] ORPHA:93929

Classic homocystinuria [Disorder - Disease] ORPHA:394
Familial cystic renal disease [Group of disorders - Category] ORPHA:93587
Bartter syndrome [Disorder - Disease] ORPHA:112
Cystinosis [Disorder - Disease] ORPHA:213
Cystinuria [Disorder - Disease] ORPHA:214
Dent disease [Disorder - Disease] ORPHA:1652
Isolated spina bifida [Group of disorders - Clinical group] ORPHA:823
Isolated partial vaginal agenesis [Disorder - Morphological anomaly] ORPHA:96269
Septate vagina [Disorder - Morphological anomaly] ORPHA:180154
Vaginal atresia [Disorder - Morphological anomaly] ORPHA:65681
Congenital primary megaureter [Disorder - Morphological anomaly] ORPHA:617
Congenital urachal anomaly [Group of disorders - Category] ORPHA:435743
Duplication of urethra [Disorder - Morphological anomaly] ORPHA:237
Congenital agenesis of the scrotum [Disorder - Morphological anomaly] ORPHA:495879
Congenital bilateral absence of vas deferens [Disorder - Morphological anomaly] ORPHA:48
Diphallia [Disorder - Morphological anomaly] ORPHA:227
Idiopathic isolated micropenis [Disorder - Morphological anomaly] ORPHA:95707
Penile agenesis [Disorder - Morphological anomaly] ORPHA:49
Penoscrotal transposition [Disorder - Morphological anomaly] ORPHA:2842
Fetal lower urinary tract obstruction [Group of disorders - Clinical group] ORPHA:435365
Hinman syndrome [Disorder - Disease] ORPHA:84085 Ochoa syndrome [Disorder - Malformation syndrome] ORPHA:2704
Posterior urethral valve [Disorder - Morphological anomaly] ORPHA:93110
Non-syndromic posterior hypospadias [Disorder - Morphological anomaly] ORPHA:95706
Isolated anorectal malformation [Group of disorders - Clinical group] ORPHA:557
Hirschsprung disease [Disorder - Disease] ORPHA:388
Anal fistula [Disorder - Particular clinical situation in a disease or syndrome] ORPHA:228113
VACTERL/VATER association [Disorder - Malformation syndrome] ORPHA:887
Caudal regression sequence [Disorder - Malformation syndrome] ORPHA:3027
Adrenogenital syndrome [Group of disorders - Category] ORPHA:181412
Rare primary hyperaldosteronism [Group of disorders - Category] ORPHA:181415
Niet in de Orphanet database
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IgG4-related retroperitoneal fibrosis [Subtype of disorder - Clinical subtype] ORPHA:49041 Familial vesicoureteral reflux [Disorder - Malformation syndrome] ORPHA:289365
Interstitial cystitis [Disorder - Disease] ORPHA:37202

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Malignant tumor of penis [Group of disorders - Category] ORPHA:398043
Tumor of testis and paratestis [Group of disorders - Category] ORPHA:363472
Adrenal/paraganglial tumor [Group of disorders - Category] ORPHA:100091
Niet in de Orphanet database
Rare renal tumor [Group of disorders - Category] ORPHA:93619
Acquired skeletal muscle disease [Group of disorders - Category] ORPHA:206638
Genetic skeletal muscle disease [Group of disorders - Category] ORPHA:206634
Immune-mediated acquired neuromuscular junction disease [Group of disorders - Clinical group] ORPHA:464764
Congenital myasthenic syndrome [Disorder - Disease] ORPHA:590
Genetic peripheral neuropathy [Group of disorders - Category] ORPHA:98497
Acquired peripheral neuropathy [Group of disorders - Category] ORPHA:182086
Amyotrophic lateral sclerosis [Disorder - Disease] ORPHA:803
Proximal spinal muscular atrophy [Disorder - Disease] ORPHA:70 Distal hereditary motor neuropathy [Group of disorders - Clinical group] ORPHA:53739 Bulbospinal muscular atrophy [Group of disorders - Category] ORPHA:206701
Mitochondrial disease [Group of disorders - Category] ORPHA:68380
Neurofibromatosis type 1 [Disorder - Disease] ORPHA:636
Neurofibromatosis type 2 [Disorder - Disease] ORPHA:637
Schwannomatosis [Disorder - Disease] ORPHA:93921
Lynch syndrome [Disorder - Disease] ORPHA:144
Intestinal polyposis syndrome [Group of disorders - Clinical group] ORPHA:104010
Hereditary breast and ovarian cancer syndrome [Disorder - Disease] ORPHA:145
PTEN hamartoma tumor syndrome [Group of disorders - Clinical group] ORPHA:306498
Li-Fraumeni syndrome [Disorder - Disease] ORPHA:524
Birt-Hogg-Dubé syndrome [Disorder - Malformation syndrome] ORPHA:122
Familial melanoma [Disorder - Disease] ORPHA:618 Familial atypical multiple mole melanoma syndrome [Disorder - Disease] ORPHA:404560
Constitutional mismatch repair deficiency syndrome [Disorder - Disease] ORPHA:252202
Familial rhabdoid tumor [Subtype of disorder - Clinical subtype] ORPHA:231108
Hereditary diffuse gastric cancer [Disorder - Disease] ORPHA:26106
Carney complex [Disorder - Disease] ORPHA:1359
Hereditary papillary renal cell carcinoma [Disorder - Disease] ORPHA:47044

Ataxia-telangiectasia [Disorder - Disease] ORPHA:100
Bloom syndrome [Disorder - Disease] ORPHA:125
Gorlin syndrome [Disorder - Malformation syndrome] ORPHA:377
Werner syndrome [Disorder - Disease] ORPHA:902
Hereditary leiomyomatosis and renal cell cancer [Disorder - Disease] ORPHA:523
Von Hippel-Lindau disease [Disorder - Disease] ORPHA:892
Fanconi anemia [Disorder - Malformation syndrome] ORPHA:84
Hereditary pheochromocytoma-paraganglioma [Disorder - Disease] ORPHA:29072
Familial long QT syndrome [Group of disorders - Clinical group] ORPHA:768
Brugada syndrome [Disorder - Disease] ORPHA:130
Catecholaminergic polymorphic ventricular tachycardia [Disorder - Disease] ORPHA:3286
Familial short QT syndrome [Disorder - Disease] ORPHA:51083
Niet in de Orphanet database
Familial progressive cardiac conduction defect [Disorder - Disease] ORPHA:871
Idiopathic ventricular fibrillation, non Brugada type [Disorder - Disease] ORPHA:228140
Niet in de Orphanet database
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Arrhythmogenic right ventricular cardiomyopathy [Group of disorders - Clinical group] ORPHA:247
Rare hypertrophic cardiomyopathy [Group of disorders - Category] ORPHA:217569
Dilated cardiomyopathy [Group of disorders - Category] ORPHA:217604
Familial restrictive cardiomyopathy [Group of disorders - Category] ORPHA:217635
Unclassified cardiomyopathy [Group of disorders - Category] ORPHA:217678
Rare congenital non-syndromic heart malformation [Group of disorders - Category] ORPHA:88991
Sarcoidosis [Disorder - Disease] ORPHA:797
Amyloidosis [Group of disorders - Category] ORPHA:69
Idiopathic giant cell myocarditis [Disorder - Disease] ORPHA:329874
Idiopathic recurrent pericarditis [Disorder - Disease] ORPHA:251307
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Multiple congenital anomalies/dysmorphic syndrome without intellectual disability [Group of disorders - Category] ORPHA:102285
Groep niet meer in de Orphanet database
Multiple congenital anomalies/dysmorphic syndrome-intellectual disability [Group of disorders - Category] ORPHA:102283
Rare syndromic intellectual disability [Group of disorders - Category] ORPHA:102369
Rare non-syndromic intellectual disability [Disorder - Disease] ORPHA:101685
Rare chromosomal anomaly [Group of disorders - Category] ORPHA:68335

Non-syndromic limb malformation [Group of disorders - Category] ORPHA:109011 Non-syndromic urogenital tract malformation [Group of disorders - Category] ORPHA:165704 Cranial malformation [Group of disorders - Category] ORPHA:98038 Non-syndromic central nervous system malformation [Group of disorders - Category] ORPHA:108989 Developmental defect of the eye [Group of disorders - Category] ORPHA:98553 Rare bone development disorder [Group of disorders - Category] ORPHA:139012 Rare head and neck malformation [Group of disorders - Category] ORPHA:155832 Non-syndromic respiratory or mediastinal malformation [Group of disorders - Category] ORPHA:108993 Non-syndromic visceral malformation [Group of disorders - Category] ORPHA:108971
Phenylketonuria [Disorder - Disease] ORPHA:716 Maternal phenylketonuria [Disorder - Malformation syndrome] ORPHA:2209 Hyperphenylalaninemia due to tetrahydrobiopterin deficiency [Disorder - Disease] ORPHA:238583 Hyperphenylalaninemia due to DNAJC12 deficiency [Disorder - Disease] ORPHA:508523
Disorder of tyrosine metabolism [Group of disorders - Category] ORPHA:284818
Maple syrup urine disease [Disorder - Disease] ORPHA:511
Disorder of branched-chain amino acid metabolism [Group of disorders - Category] ORPHA:79197 Disorder of cobalamin metabolism and transport [Group of disorders - Category] ORPHA:79171
Disorder of lysine and hydroxylysine metabolism [Group of disorders - Category] ORPHA:289832
Cerebral organic aciduria [Group of disorders - Category] ORPHA:79158
Disorder of methionine cycle and sulfur amino acid metabolism [Group of disorders - Category] ORPHA:79173
Disorder of urea cycle metabolism and ammonia detoxification [Group of disorders - Category] ORPHA:79167 Disorder of ornithine metabolism [Group of disorders - Category] ORPHA:289869
Pyruvate metabolism disorder [Group of disorders - Category] ORPHA:254746 Tricarboxylic acid cycle disorder [Group of disorders - Category] ORPHA:254749 Mitochondrial oxidative phosphorylation disorder [Group of disorders - Category] ORPHA:223713 Disorder of thiamine metabolism and transport [Group of disorders - Category] ORPHA:298644
Glycogen storage disease [Group of disorders - Category] ORPHA:79201
Disorder of galactose metabolism [Group of disorders - Category] ORPHA:308467
Disorder of pentose phosphate metabolism [Group of disorders - Category] ORPHA:79186
Disorder of fructose metabolism [Group of disorders - Category] ORPHA:308463
Hyperinsulinemic hypoglycaemia [Group of disorders - Category] ORPHA:443095
Disorder of fatty acid oxidation and ketone body metabolism [Group of disorders - Category] ORPHA:79174
Gluconeogenesis disorder [Group of disorders - Category] ORPHA:79177
Genetic recurrent myoglobinuria [Disorder - Disease] ORPHA:99845
Sphingolipidosis [Group of disorders - Category] ORPHA:79225
Oligosaccharidosis [Group of disorders - Category] ORPHA:79215
Mucopolysaccharidosis [Group of disorders - Category] ORPHA:79213
Mucolipidosis [Group of disorders - Category] ORPHA:79212
Neuronal ceroid lipofuscinosis [Group of disorders - Clinical group] ORPHA:216
Lipid storage disease [Group of disorders - Category] ORPHA:79204
Disorder of lysosomal amino acid transport [Group of disorders - Category] ORPHA:79207
Lysosomal glycogen storage disease [Group of disorders - Category] ORPHA:309337
Peroxisome biogenesis disorder [Group of disorders - Clinical group] ORPHA:79189
Rhizomelic chondrodysplasia punctata [Disorder - Disease] ORPHA:177

X-linked adrenoleukodystrophy [Disorder - Disease] ORPHA:43
Peroxisomal acyl-CoA oxidase deficiency [Disorder - Disease] ORPHA:2971
Bifunctional enzyme deficiency [Disorder - Disease] ORPHA:300
Refsum disease [Disorder - Disease] ORPHA:773
Congenital bile acid synthesis defect type 4 [Disorder - Disease] ORPHA:79095
Congenital disorder of glycosylation [Group of disorders - Category] ORPHA:137
Creatine deficiency syndrome [Group of disorders - Clinical group] ORPHA:79172
Disorder of purine or pyrimidine metabolism [Group of disorders - Category] ORPHA:79224
Disorder of mineral absorption and transport [Group of disorders - Category] ORPHA:309836
Classic glucose transporter type 1 deficiency syndrome [Disorder - Disease] ORPHA:71277
Disorder of neurotransmitter metabolism and transport [Group of disorders - Category] ORPHA:79169
Disorder of folate metabolism and transport [Group of disorders - Category] ORPHA:285657
Disorder of gamma-aminobutyric acid metabolism [Group of disorders - Category] ORPHA:79175
Porphyria due to ALA dehydratase deficiency [Disorder - Disease] ORPHA:100924
Hereditary coproporphyria [Disorder - Disease] ORPHA:79273
Myeloid hemopathy [Group of disorders - Category] ORPHA:171895
Lymphoid hemopathy [Group of disorders - Category] ORPHA:171898
Rare tumor of neuroepithelial tissue [Group of disorders - Category] ORPHA:251558
Neuroblastoma [Disorder - Disease] ORPHA:635
Retinoblastoma [Disorder - Disease] ORPHA:790
Nephroblastoma [Disorder - Disease] ORPHA:654
Hepatoblastoma [Disorder - Disease] ORPHA:449
Skeletal Ewing sarcoma [Disorder - Disease] ORPHA:319
Soft tissue sarcoma [Group of disorders - Clinical group] ORPHA:3394
Germ cell tumor [Group of disorders - Category] ORPHA:3399
Malignant melanoma of the mucosa [Disorder - Disease] ORPHA:168999 Melanoma of soft tissue [Disorder - Disease] ORPHA:97338 Primary melanoma of the central nervous system [Disorder - Disease] ORPHA:252050
Langerhans cell histiocytosis [Disorder - Disease] ORPHA:389
Primary biliary cholangitis [Disorder - Disease] ORPHA:186
Autoimmune hepatitis [Disorder - Disease] ORPHA:2137
Primary sclerosing cholangitis [Disorder - Disease] ORPHA:171
IgG4-related sclerosing cholangitis [Disorder - Disease] ORPHA:447764
Genetic biliary tract disease [Group of disorders - Category] ORPHA:156607 Alagille syndrome [Disorder - Malformation syndrome] ORPHA:52
Isolated biliary atresia [Disorder - Morphological anomaly] ORPHA:30391 Syndromic biliary atresia [Group of disorders - Clinical group] ORPHA:498350
Choledochal cyst [Disorder - Morphological anomaly] ORPHA:480501

Alpha-1-antitrypsin deficiency [Disorder - Disease] ORPHA:60
Wilson disease [Disorder - Disease] ORPHA:905
Isolated polycystic liver disease [Disorder - Malformation syndrome] ORPHA:2924 Caroli disease [Disorder - Malformation syndrome] ORPHA:53035
Rare vascular liver disease [Group of disorders - Category] ORPHA:101938
Rare hepatic and biliary tract tumor [Group of disorders - Category] ORPHA:101943
Acute liver failure [Disorder - Clinical syndrome] ORPHA:90062
Intrahepatic cholestasis of pregnancy [Disorder - Disease] ORPHA:69665
HELLP syndrome [Disorder - Disease] ORPHA:244242
Acute fatty liver of pregnancy [Disorder - Disease] ORPHA:243367
Systemic sclerosis [Disorder - Disease] ORPHA:90291
Mixed connective tissue disease [Disorder - Disease] ORPHA:809
Polymyositis [Disorder - Disease] ORPHA:732 Dermatomyositis [Disorder - Disease] ORPHA:221 Antisynthetase syndrome [Disorder - Disease] ORPHA:81
Antiphospholipid syndrome [Disorder - Disease] ORPHA:80 Catastrophic antiphospholipid syndrome [Disorder - Disease] ORPHA:464343 Neonatal antiphospholipid syndrome [Disorder - Disease] ORPHA:398097
Undifferentiated connective tissue syndrome [Disorder - Disease] ORPHA:90002
IgG4-related disease [Group of disorders - Clinical group] ORPHA:284264
Relapsing polychondritis [Disorder - Disease] ORPHA:728
Systemic lupus erythematosus [Disorder - Disease] ORPHA:536
Primary Sjögren syndrome [Disorder - Disease] ORPHA:289390
Ehlers-Danlos syndrome [Group of disorders - Clinical group] ORPHA:98249
Primary immunodeficiency [Group of disorders - Category] ORPHA:101997
Autoinflammatory syndrome [Group of disorders - Category] ORPHA:93665
Vasculitis [Group of disorders - Category] ORPHA:52759 Rare pediatric vasculitis [Group of disorders - Category] ORPHA:280369 Rare neuroinflammatory or neuroimmunological disease [Group of disorders - Category] ORPHA:182064
Juvenile idiopathic arthritis [Group of disorders - Clinical group] ORPHA:92 Juvenile idiopathic inflammatory myopathy [Group of disorders - Category] ORPHA:329888 Pediatric systemic lupus erythematosus [Disorder - Disease] ORPHA:93552 Overlapping connective tissue disease [Group of disorders - Clinical group] ORPHA:251312 Rheumatic fever [Disorder - Disease] ORPHA:3099 Scleroderma [Group of disorders - Clinical group] ORPHA:801
nvt
Rare disorder potentially indicated for liver transplant [Group of disorders - Category] ORPHA:506210
Rare disorder potentially indicated for kidney transplant [Group of disorders - Category] ORPHA:506213
Rare disorder potentially indicated for lung transplant [Group of disorders - Category] ORPHA:506222
Rare disorder potentially indicated for heart transplant [Group of disorders - Category] ORPHA:506225
Rare disorder potentially indicated for bowel transplant [Group of disorders - Category] ORPHA:506216

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Rare disorder potentially indicated for hematopoietic stem cell transplant [Group of disorders - Category] ORPHA:506219
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Hereditary hemorrhagic telangiectasia [Disorder - Disease] ORPHA:774
Marfan syndrome [Disorder - Disease] ORPHA:558
Loeys-Dietz syndrome [Disorder - Malformation syndrome] ORPHA:60030
Aneurysm-osteoarthritis syndrome [Disorder - Disease] ORPHA:284984
Arterial tortuosity syndrome [Disorder - Malformation syndrome] ORPHA:3342
Familial thoracic aortic aneurysm and aortic dissection [Disorder - Disease] ORPHA:91387
Multisystemic smooth muscle dysfunction syndrome [Disorder - Disease] ORPHA:404463
Autosomal recessive cutis laxa type 1 [Disorder - Disease] ORPHA:90349
Rare venous malformation [Group of disorders - Category] ORPHA:211252
Rare lymphatic malformation [Group of disorders - Category] ORPHA:2415
Rare capillary malformation [Group of disorders - Category] ORPHA:211247
Rare arteriovenous malformation [Group of disorders - Category] ORPHA:211266
Complex vascular malformation with associated anomalies [Group of disorders - Category] ORPHA:211277
Rare vascular tumor [Group of disorders - Category] ORPHA:211237
Primary lymphedema [Group of disorders - Category] ORPHA:77240
Congenital primary lymphedema without systemic or visceral involvement [Group of disorders - Clinical group] ORPHA:2416 Milroy disease [Disorder - Disease] ORPHA:79452
Late-onset primary lymphedema without systemic or visceral involvement [Group of disorders - Clinical group] ORPHA:289825
Meige disease [Disorder - Disease] ORPHA:90186
Lymphedema-distichiasis syndrome [Disorder - Malformation syndrome] ORPHA:33001 Deafness-lymphedema-leukemia syndrome [Disorder - Malformation syndrome] ORPHA:3226
Primary lymphedema with systemic or visceral involvement [Group of disorders - Category] ORPHA:568044
Hennekam syndrome [Disorder - Malformation syndrome] ORPHA:2136
PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis [Disorder - Disease] ORPHA:568062

PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis [Disorder - Disease] ORPHA:568062 EPHB4-related lymphatic-related hydrops fetalis [Disorder - Disease] ORPHA:568065
Primary lymphedema with systemic or visceral involvement [Group of disorders - Category] ORPHA:568044
Door wijzigingen in de classificaties is erkenning voor deze groep niet meer aan te vragen
Noonan syndrome [Disorder - Malformation syndrome] ORPHA:648 Cardiofaciocutaneous syndrome [Disorder - Malformation syndrome] ORPHA:1340
Turner syndrome [Disorder - Malformation syndrome] ORPHA:881
Monosomy 22q13.3 [Disorder - Malformation syndrome] ORPHA:48652
Microcephaly-lymphedema-chorioretinopathy syndrome [Disorder - Malformation syndrome] ORPHA:2526
Complex vascular malformation with associated anomalies [Group of disorders - Category] ORPHA:211277
Vascular Ehlers-Danlos syndrome [Disorder - Disease] ORPHA:286
Niet in de Orphanet database
Idiopathic spontaneous coronary artery dissection [Disorder - Disease] ORPHA:458718