Table S1. List of rare diseases endowed of an exemption code as defined by the Italian law and included in the study.

INFECTIOUS AND PARASITIC DISEASES	
Disease and/or group	*Specific diseases included in the group*
LEPROSY	
LYME DISEASE	
WHIPPLE DISEASE	
NEOPLASMS	
Disease and/or group	*Specific diseases included in the group*
CARNEY COMPLEX	
FAMILIAL ADENOMATOUS POLYPOSIS	
GARDNER SYNDROME	
GORLIN SYNDROME RETINOBLASTOMA	
HEREDITARY NONPOLYPOSIS COLON CANCER	Lynch syndrome
LYMPHANGIOLEIOMYOMATOSIS	
NEUROFIBROMATOSIS	
NEPHROBLASTOMA	
RETINOBLASTOMA	
ENDOCRINE DISEASES	
Disease and/or group	*Specific diseases included in the group*
ADRENOGENITAL SYNDROME	
AUTOIMMUNE POLYENDOCRINOPATHY	Schmidt's syndrome
CONGENITAL HYPOGONADOTROPIC HYPOGONADISM	
CONGENITAL ISOLATED ACTH DEFICIENCY	
FAMILIAL MEDULLARY THYROID CARCINOMA	
IDIOPATHIC CENTRAL PRECOCIOUS PUBERTY	
KALLMANN SYNDROME	
LARON SYNDROME	
LEPRECHAUNISM	
MULTIPLE ENDOCRINE NEOPLASIA TYPE 1	
MULTIPLE ENDOCRINE NEOPLASIA TYPE 2A	
NON-ACQUIRED ISOLATED GROWTH HORMONE DEFICIENCY	
PENDRED SYNDROME	
REFETOFF SYNDROME	

RARE PRIMARY HYPERALDOSTERONISM	Primary hyperaldosteronism-seizures-neurological abnormalities syndrome Primary unilateral adrenal hyperplasia
METABOLIC DISEASES	
Disease and/or group	*Specific diseases included in the group*
DISORDERS OF MITOCHONDRIAL METABOLISM	
CREATINE DEFICIENCY SYNDROME	
KEARNS-SAYRE SYNDROME	
ISOLATED OXIDATIVE PHOSPHORYLATION COMPLEX DISORDER	
LEIGH SYNDROME	
LEBER OPTIC ATROPHY	
LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY	
MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY	
MELAS	
MERFF	
MITOCHONDRIAL OXIDATIVE PHOSPHORYLATION DISORDER DUE TO MITOCHONDRIAL DNA ANOMALIES	
MITOCHONDRIAL OXIDATIVE PHOSPHORYLATION DISORDER DUE TO NUCLEAR DNA ANOMALIES	
OTHER DISORDER OF MITOCHONDRIAL METABOLISM	Epileptic encephalopathy with global cerebral demyelination
PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	
SHORT CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY	
VERY LONG CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY	
LYSOSOMAL STORAGE DISEASES	
GALACTOSIALIDOSIS	
GANGLIOSIDOSIS	
KRABBE DISEASE	

LIPID STORAGE DISEASE	Fabry disease Gaucher disease Niemann-Pick disease Niemann-Pick disease type C
METACHROMATIC LEUKODYSTROPHY	
MUCOLIPIDOSIS	
MUCOPOLYSACCHARIDOSIS	Mucopolysaccharidosis type 4
NEURONAL CEROID LIPOFUSCINOSIS	
OTHER LYSOSOMAL STORAGE DISEASES	Glycogen storage disease due to LAMP-2 deficiency
DISORDERS OF VITAMIN AND NON-PROTEIN COFACTOR ABSORPTION AND TRANSPORT	
BIOTINIDASE DEFICIENCY	
DISORDER OF COBALAMIN AND FOLATE METABOLISM AND TRANSPORT	Methylmalonic acidemia with homocystinuria, type cblC
DISORDER OF OTHER VITAMINS AND COFACTORS METABOLISM AND TRANSPORT* *excludes Ataxia with vitamin E deficiency	
HYPOCALCEMIC VITAMIN D-DEPENDENT RICKETS	
HYPOPHOSPHATEMIC RICKETS	
DISORDERS OF METAL METABOLISM AND TRANSPORT	
ACERULOPLASMINEMIA	
DISORDER OF IRON METABOLISM AND TRANSPORT	Rare hereditary hemochromatosis Hereditary hyperferritinemia-cataract syndrome
MENKES DISEASE	
WILSON DISEASE	
DISORDERS OF PROTEIN METABOLISM AND TRANSPORT	
CONGENITAL DISORDER OF GLYCOSYLATION	
CRIGLER-NAJJAR SYNDROME	
PRIMARY SYSTEMIC AMYLOIDOSIS	Wild type ATTR amyloidosis
OTHER METABOLIC DISEASES	
ADIPOSIS DOLOROSA	
ADRENOLEUKODYSTROPHY	

CEREBROTENDINOUS XANTHOMATOSIS	
DISORDER OF AMINO ACID ABSORPTION AND TRANSPORT	Albinism Alkaptonuria Cystinosis Cystinuria Glutaric aciduria Glycine encephalopathy Hyperprolinemia Homocystinuria Maple syrup urine disease Methylmalonic acidemia Phenylketonuria/ Hyperphenylalaninemia due to tetrahydrobiopterin deficiency Pyruvate dehydrogenase E1-alpha deficiency Tyrosinemia Other acidemias or primitive organic acidurias from metabolism defects of branched chain amino acids
DISORDER OF CARBOHYDRATE METABOLISM* *excludes Diabetes mellitus	Fructose-1,6-bisphosphatase deficiency Glucose transport disorder Galactosemia Glycogen storage disease Glycogen storage disease due to acid maltase deficiency Hereditary fructose intolerance
DISORDER OF LIPID METABOLISM* *excludes Combined familial hyperlipidemia, Dysbetalipoproteinemia, Heterozigous familial hypercholesterolemia type IIA and IIB, Polygenic hypercholesterolemia	Familial lipoprotein lipase deficiency Homozygous familial hypercholesterolemia Familial chylomicronemia syndrome Hypobetalipoproteinemia Tangier disease
DISORDER OF NEUROTRANSMITTER AND PEPTIDE METABOLISM	
DISORDER OF PURINE OR PYRIMIDINE METABOLISM	Lesch-Nyhan syndrome
DISORDER OF UREA CYCLE METABOLISM AND AMMONIA DETOXIFICATION	Citrullinemia Ornithine transcarbamylase deficiency
FAMILIAL TUMORAL CALCINOSIS	
FAMILIAL HYPERINSULINISM	
GENERALIZED LIPODYSTROPHY	
HYPOPHOSPHATASIA	

PEROXISOMAL DISEASE	
PORPHYRIA	
REFSUM DISEASE	
ZELLWEGER SYNDROME	
IMMUNE SYSTEM DISORDERS	
Disease and/or group	*Specific diseases included in the group*
ACQUIRED ANGIOEDEMA WITH C1INH DEFICIENCY	
ALPHA-1-ANTITRYPSIN DEFICIENCY	
AUTOINFLAMMATORY SYNDROME	Periodic fever syndrome Hyperimmunoglobulinemia D with periodic fever Cryopyrin-associated periodic syndrome
CHRONIC HISTIOCYTOSIS	Erdheim-Chester disease Langerhans cell histiocytosis Histiocytosis X
FAMILIAL MEDITERRANEAN FEVER	
HEREDITARY ANGIOEDEMA	
NEONATAL ANTIPHOSPHOLIPID SYNDROME	
PRIMARY IMMUNODEFICIENCY	Agammaglobulinemia DiGeorge syndrome
SCHNITZLER SYNDROME	
TUMOR NECROSIS FACTOR RECEPTOR 1 ASSOCIATED PERIODIC SYNDROME	
DISEASES OF THE BLOOD AND BLOOD-FORMING ORGANS	
Disease and/or group	*Specific diseases included in the group*
CYCLIC NEUTROPENIA	
CHRONIC GRANULOMATOUS DISEASE	
CHRONIC PRIMARY PLATELET DISORDER	Immune thrombocytopenia
HEMOLYTIC UREMIC SYNDROME	
HEREDITARY ANEMIA* *excludes Class I glucose-6-phosphate dehydrogenase deficiency	Blackfan-Diamond anemia Fanconi anemia Hereditary spherocytosis Sickle cell anemia Sideroblastic anemia Thalassemia

INHERITED COAGULATION DISORDER	Hemophilia A Hemophilia B Rare hemorrhagic disorder due to a constitutional coagulation factors defect Rare hereditary thrombophilia Von Willebrand disease
MYELODYSPLASTIC SYNDROME	Refractory anemia
PAROXYSMAL NOCTURNAL HEMOGLOBINURIA	
RARE ACQUIRED APLASTIC ANEMIA* *excludes Transitory medullary aplasia	
RARE HEMORRHAGIC DISORDER DUE TO A CONSTITUTIONAL PLATELET ANOMALY	
RARE HEMORRHAGIC DISORDER DUE TO A CONSTITUTIONAL THROMBOCYTOPENIA	
SEVERE CONGENITAL NEUTROPENIA	Adult idiopathic neutropenia
SHWACHMAN-DIAMOND SYNDROME	
SYSTEMIC MASTOCYTOSIS	
PERIPHERAL AND CENTRAL NERVOUS SYSTEM DISORDERS	
Disease and/or group	*Specific diseases included in the group*
AMYOTROPHIC LATERAL SCLEROSIS	
AUTOSOMAL DOMINANT CEREBELLAR ATAXIA	Ataxia with vitamin E deficiency Cerebellar ataxia-hypogonadism syndrome Fragile X-associated tremor/ataxia syndrome Friedreich ataxia Hereditary episodic ataxia Hereditary spastic paraplegia Marie's cerebellar ataxia Marinesco-Sjögren syndrome Pantothenate kinase-associated neurodegeneration
BILATERAL STRIOPALLIDODENTATE CALCINOSIS	
CEREBRAL AUTOSOMAL DOMINANT ARTERIOPATHY-SUBCORTICAL INFARCTS-LEUKOENCEPHALOPATHY	
CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY	
CONGENITAL MYASTHENIC SYNDROME	Myasthenia gravis
CONGENITAL MYOPATHY	Central core disease Centronuclear myopathy

	Nemaline myopathy Qualitative or quantitative defects of desmin
DENTATORUBRAL PALLIDOLUYSIAN ATROPHY	
DRAVET SYNDROME	
FAMILIAL OR SPORADIC HEMIPLEGIC MIGRAINE	
GENETIC PERIPHERAL NEUROPATHY	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy Dejerine-Sottas syndrome Giant axonal neuropathy Hereditary neuropathy with liability to pressure palsies Hereditary sensory and autonomic neuropathy Roussy-Lévy syndrome
GUILLAIN-BARRÉ SYNDROME	
HUNTINGTON DISEASE	
IDIOPATHIC TORSION DYSTHONIA	
ISAAC SYNDROME	
LAMBERT-EATON MYASTHENIC SYNDROME	
LANDAU-KLEFFNER SYNDROME	
LENNOX-GASTAUT SYNDROME	
LEUKODYSTROPHY	Aicardi-Goutières syndrome Alexander disease Pelizaeus-Merzbacher disease
MELKERSSON-ROSENTHAL SYNDROME	
MULTIFOCAL MOTOR NEUROPATHY	
MULTIPLE SYSTEM ATROPHY	
MUSCULAR DYSTROPHY	Becker muscular dystrophy Calpain-3-related limb-girdle muscular dystrophy R1 Duchenne muscular dystrophy Facioscapulohumeral dystrophy Oculogastrointestinal muscular dystrophy
MYOTONIC DYSTROPHY	Paramyotonia congenita of Von Eulenburg Steinert myotonic dystrophy Thomsen and Becker disease
NARCOLEPSY	
NEURODEGENERATION WITH BRAIN IRON ACCUMULATION	Infantile neuroaxonal dystrophy

NEURONAL INTRANUCLEAR INCLUSION DISEASE	
PERIODIC PARALYSIS	
POEMS SYNDROME	
PRIMARY DYSTONIA	
PRIMARY LATERAL SCLEROSIS	
PROGRESSIVE MYOCLONIC EPILEPSY	
PROGRESSIVE SUPRANUCLEAR PALSY	
RETT SYNDROME	
RIBOFLAVIN TRANSPORTER DEFICIENCY	
SPINAL MUSCULAR ATROPHY	Kennedy disease Proximal spinal muscular atrophy type 1 Proximal spinal muscular atrophy type 3
STIFF PERSON SPECTRUM DISORDER	
SYRINGOMYELIA	
WEST SYNDROME	
DISORDERS OF THE EYE AND ADNEXA	
Disease and/or group	*Specific diseases included in the group*
COGAN SYNDROME	
CONGENITAL STATIONARY NIGHT BLINDNESS	
CORNEAL DYSTROPHY	Epithelial basement membrane dystrophy Fuchs endothelial corneal dystrophy Granular corneal dystrophy type II Macular corneal dystrophy Meesmann corneal dystrophy Posterior polymorphous corneal dystrophy Reis-Bücklers corneal dystrophy
FAMILIAL EXUDATIVE VITREORETINOPATHY	
ISOLATED CHORIORETINAL DYSTROPHY	
ISOLATED INHERITED RETINAL DISORDER	Best vitelliform macular dystrophy Leber congenital amaurosis Progressive cone dystrophy Retinitis pigmentosa Retinitis punctata albescens Stargardt disease Vitreoretinal distrophy

OGUCHI DISEASE	
POSTERIOR UVEITIS	
SERPIGINOUS CHOROIDITIS	
SYNDROMIC KERATOCONUS	
CIRCULATORY SYSTEM DISEASES	
Disease and/or group	*Specific diseases included in the group*
ANTI-GLOMERULAR BASEMENT MEMBRANE DISEASE	
BEHÇET DISEASE	
BUDD-CHIARI SYNDROME	
CRYOGLOBULINEMIC VASCULITIS	
EOSINOPHILIC GRANULOMATOSIS WITH POLYANGIITIS	
GIANT CELL ARTERITIS	
GRANULOMATOSIS WITH POLYANGIITIS	
HEREDITARY HEMORRHAGIC TELANGIECTASIA	
IMMUNOGLOBULIN A VASCULITIS	
MICROSCOPIC POLYANGIITIS	
POLYARTERITIS NODOSA	
PRIMARY LYMPHEDEMA	Milroy disease Meige disease Idiopathic primary lymphedema Recessive primary congenital lymphedema
RHEUMATIC HEART DISEASE	
TAKAYASU ARTERITIS	
THROMBOTIC MICROANGIOPATHY	Hemolytic uremic syndrome Thrombotic thrombocytopenic purpura
RESPIRATORY DISEASES	
Disease and/or group	*Specific diseases included in the group*
AUTOIMMUNE PULMONARY ALVEOLAR PROTEINOSIS	
IDIOPATHIC PULMONARY ARTERIAL HYPERTENSION	
IDIOPATHIC PULMONARY HEMOSIDEROSIS	
ONDINE SYNDROME	
PRIMARY CILIARY DYSKINESIA	
PRIMARY CILIARY DYSKINESIA, KARTAGENER TYPE	

PRIMARY INTERSTITIAL LUNG DISEASE IN CHILDHOOD AND ADULTHOOD	Acute interstitial pneumonia Idiopathic pulmonary fibrosis
SARCOIDOSIS	
DIGESTIVE DISORDERS	
Disease and/or group	*Specific diseases included in the group*
CHRONIC INTESTINAL PSEUDOOBSTRUCTION	
CONGENITAL CHLORIDE DIARRHEA	
EOSINOPHILIC GASTROENTERITIS	
ISOLATED ACHALASIA AND SYNDROMES-ASSOCIATED ACHALASIA	Triple A syndrome
PRIMARY INTESTINAL LYMPHANGIECTASIA	
PRIMARY SCLEROSING CHOLANGITIS	
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS	Progressive familial intrahepatic cholestasis type 3
DISEASES OF THE GENITOURINARY SYSTEM	
Disease and/or group	*Specific diseases included in the group*
ALPORT SYNDROME	
IGG4-RELATED RETROPERITONEAL FIBROSIS	
INTERSTITIAL CYSTITIS	
NEPHROGENIC DIABETES INSIPIDUS	
PRIMARY GLOMERULAR DISEASE	
RARE RENAL TUBULAR DISEASE	Bartter syndrome Dent disease Gitelman syndrome
DISEASES OF THE SKIN AND SUBCUTANEOUS TISSUE	
Disease and/or group	*Specific diseases included in the group*
APLASIA CUTIS CONGENITA	
BULLOUS PEMPHIGOID	
CONGENITAL NON-BULLOUS ICHTHYOSIFORM ERYTHRODERMA	
CUTIS LAXA	
DARIER DISEASE	
DIFFUSE CUTANEOUS SYSTEMIC SCLEROSIS	

DYSKERATOSIS CONGENITA	
EEC SYNDROME	
ECTODERMAL DYSPLASIA SYNDROME	
EPIDERMAL NEVUS SYNDROME	
ERYTHROKERATODERMIA VARIABILIS	
FAMILIAL ATYPICAL MULTIPLE MOLE MELANOMA SYNDROME	
HEREDITARY PALMOPLANTAR KERATODERMA	
HYPOMELANOSIS OF ITO	
IBIDS SYNDROME	
INCONTINENTIA PIGMENTI	
INHERITED EPIDERMOLYSIS BULLOSA	
INHERITED ICHTHYOSIS	Autosomal recessive congenital ichthyosis Lamellar ichthyosis Netherton syndrome X-linked ichthyosis
KERATOSIS FOLLICULARIS SPINULOSA DECALVANS	
LICHEN SCLEROSUS ET ATROPHICUS	
MUCOUS MEMBRANE PEMPHIGOID	
PEMPHIGUS	
PSEUDOXANTHOMA ELASTICUM	
PYODERMA GANGRENOSUM	
SJÖGREN-LARSSON SYNDROME	

DISEASES OF THE MUSCULOSKELETAL SYSTEM AND CONNECTIVE TISSUE

Disease and/or group	*Specific diseases included in the group*
ANTISYNTHETASE SYNDROME	
DERMATOMYOSITIS	
DIFFUSE FASCIITIS	
EOSINOPHILIC FASCIITIS	
FIBRODYSPLASIA OSSIFICANS PROGRESSIVA	
GORHAM-STOUT DISEASE	
INCLUSION BODY MYOSITIS	
MIXED CONNECTIVE TISSUE DISEASE	
POLYMYOSITIS	
RELAPSING POLYCHONDRITIS	
SAPHO SYNDROME	

SYSTEMIC SCLEROSIS	

CONGENITAL ANOMALIES, CHROMOSOMAL ABERRATIONS AND GENETIC SYNDROMES

Disease and/or group	*Specific diseases included in the group*
CONGENITAL MALFORMATIONS OF THE NERVOUS SYSTEM	
ARNOLD-CHIARI MALFORMATION	
CEREBELLAR AGENESIS	
GERSTMANN SYNDROME	
ISOLATED OR SYNDROMIC HOLOPROSENCEPHALY	
ISOLATED OR SYNDROMIC MICROCEPHALY	
JOUBERT SYNDROME AND RELATED DISORDERS	
LISSENCEPHALY	
SYNDROME WITH CORPUS CALLOSUM AGENESIS/DYSGENESIS AS A MAJOR FEATURE	Dandy-Walker sydrome
WALKER-WARBURG SYNDROME	
OTHER SEVERE AND DISABLING SYNDROMES WITH A CENTRAL NERVOUS SYSTEM MALFORMATION AS A MAJOR FEATURE	
CONGENITAL MALFORMATIONS OF THE EYE	
ANIRIDIA	
AXENFELD-RIEGER SYNDROME	
COLOBOMA OF THE OPTIC NERVE	
ISOLATED OR SYNDROMIC CONGENITAL OCULAR COLOBOMA	
MICROPHTHALMIA-ANOPHTHALMIA-COLOBOMA	Microphthalmia, Lenz type
PETERS ANOMALY	
RIEGER ANOMALY	
SEPTO-OPTIC DYSPLASIA SPECTRUM	
VOGT-KOYANAGI-HARADA DISEASE	
ISOLATED OR SYNDROMIC CONGENITAL CRANIOFACIAL ANOMALIES	
ACROCEPHALOSYNDACTYLY	
ANTLEY-BIXLER SYNDROME	
APERT SYNDROME	

PFEIFFER SYNDROME	
PIERRE ROBIN SYNDROME	
OTHER SEVERE AND DISABLING ANOMALIES OF SKULL AND FACE BONES, INTEGUMENTS AND MUCOSA	Crouzon disease Frontofacionasal dysplasia Isolated craniosynostosis Isolated or syndromic cleft palate
CONGENITAL FACIAL ABNORMALITIES	
GOLDENHAR SYNDROME	
MOEBIUS SYNDROME	
SCHINZEL-GIEDION SYNDROME	
CONGENITAL LIMB MALFORMATIONS	
ADAMS-OLIVER SYNDROME	
ARTHROGRYPOSIS MULTIPLEX CONGENITA	
CONGENITAL ABSENCE OF UPPER ARM AND FOREARM WITH HAND PRESENT	
FEMORAL-FACIAL SYNDROME	
FREEMAN-SHELDON SYNDROME	
POLAND SYNDROME	
CONGENITAL HEART DISEASE	
KLIPPEL-TRÉNAUNAY SYNDROME	
RARE CONGENITAL NON-SYNDROMIC HEART MALFORMATION* *excludes Isolated atrial septal defect, Isolated pulmonary valve stenosis, Isolated ventricular septal defect, Patent ductus arteriosus	Ebstein malformation of the tricuspid valve Hypoplastic left heart syndrome
OTHER SEVERE AND DISABLING CONGENITAL SYNDROMIC VASCULAR DISEASE	CLOVES syndrome Capillary malformation-arteriovenous malformation
ABDOMINAL WALL DEFECTS	
ISOLATED KLIPPEL-FEIL SYNDROME	
CONGENITAL ANOMALIES OF THE GASTROINTESTINAL TRACT	
ANORECTAL MALFORMATION	
CAROLI DISEASE	
DUPLICATION OF THE GASTROINTESTINAL TRACT	

ESOPHAGEAL ATRESIA AND/OR ISOLATED TRACHEOESOPHAGEAL FISTULA	
ISOLATED BILIARY ATRESIA	
ISOLATED POLYCYSTIC LIVER DISEASE	
HIRSCHSPRUNG DISEASE	
PERSISTENT CLOACA	
CONGENITAL UROGENITAL ANOMALIES	
AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE	
BLADDER EXSTROPHY	
DISORDER OF SEXUAL DEVELOPMENT AND/OR AMBIGUOUS GENITALIA AND/OR KARIOTYPE DISCORDANCE/GONADAL DEVELOPMENT AND/OR PHENOTYPE	Gonadal dysgenesis Complete androgen insensitivity syndrome Partial androgen insensitivity syndrome
MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME	
FAMILIAL CYSTIC RENAL DISEASE	
MEDULLARY SPONGE KIDNEY	
PSEUDOHERMAPHRODITISM	
OTHER SEVERE AND DISABLING DEFECTS OF SEXUAL DEVELOPMENT AND/OR AMBIGUOUS GENITALIA AND/OR KARIOTYPE DISCORDANCE/GONADAL DEVELOPMENT AND/OR PHENOTYPE	
GENETIC SKELETAL DISORDERS	
ACRODYSOSTOSIS	
CONGENITAL CHONDRODISPLASIAS	Achondroplasia Jeune syndrome Multiple osteochondromas
CONGENITAL ISOLATED OR SYNDROMIC CHONDRODISPLASIAS	Camurati-Engelmann disease Craniometaphyseal dysplasia Fibrous dysplasia Léri-Weill dyschondrosteosis Multiple epiphyseal dysplasia McCune-Albright syndrome Osteogenesis imperfecta Osteopetrosis and related disorders Spondyloepiphyseal dysplasia tarda
MAFFUCCI SYNDROME	

SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA	
OTHER SYNDROMES AND COMPLEX CONGENITAL MALFORMATIONS	
22q11.2 DELETION SYNDROME	
AARSKOG-SCOTT SYNDROME	
ALAGILLE SYNDROME	
ALSTRÖM SYNDROME	
ANGELMAN SYNDROME	
BARDET-BIEDL SYNDROME	
BECKWITH-WIEDEMANN SYNDROME	
BOR SYNDROME	
BORJESON-FORSSMAN-LEHMANN SYNDROME	
BRANCHIO-OCULO-FACIAL SYNDROME	
CARDIOFACIOCUTANEOUS SYNDROME	
CHAR SYNDROME	
CHARGE SYNDROME	
COCKAYNE SYNDROME	
COFFIN-SIRIS SYNDROME	
COHEN SYNDROME	
CORNELIA DE LANGE SYNDROME	
DUBOWITZ SYNDROME	
EHLERS-DANLOS SYNDROME	
FRAGILE X SYNDROME	
HOLT-ORAM SYNDROME	
ISOLATED HEMIHYPERPLASIA	
KABUKI SYNDROME	
LOEYS-DIETZ SYNDROME	
MARFAN SYNDROME	
MARSHALL SYNDROME	
MARSHALL-SMITH SYNDROME	
MONOSOMY 5P	
MOSAIC VARIEGATED ANEUPLOIDY SYNDROME	
MULTIPLE CONGENITAL ANOMALIES/DYSMORPHIC SYNDROME-INTELLECTUAL DISABILITY	KBG syndrome
MULTIPLE HAMARTOMAS	Cowden syndrome Hepatic cystic hamartoma

NAIL-PATELLA SYNDROME	
NOONAN SYNDROME AND NOONAN-RELATED SYNDROME	
NOONAN SYNDROME WITH MULTIPLE LENTIGINES	
OCULODENTODIGITAL DYSPLASIA	
OPITZ GBBB SYNDROME	
PEUTZ-JEGHERS SYNDROME	
PRADER-WILLI SYNDROME	
PROGRESSIVE HEMIFACIAL ATROPHY	
RUBINSTEIN-TAYBI SYNDROME	
SALDINO-MAINZER SYNDROME	
SECKEL SYNDROME	
SILVER-RUSSELL SYNDROME	
SMITH-MAGENIS SYNDROME	
SOTOS SYNDROME	
STICKLER SYNDROME	
STURGE-WEBER SYNDROME	
SYNDROMIC CHROMOSOMAL AND GENOMIC UNBALANCED REARRANGEMENTS	Distal 22q11.2 microdeletion syndrome
TETRASOMY 12P	
TOWNES-BROCKS SYNDROME	
TUBEROUS SCLEROSIS COMPLEX	
TURNER SYNDROME	
VACTERL/VATER ASSOCIATION	
VON HIPPEL-LINDAU DISEASE	
WAGR SYNDROME	
WAARDENBURG SYNDROME	
WILLIAMS SYNDROME	
WOLF-HIRSCHHORN SYNDROME	
WOLFRAM SYNDROME	
NEONATAL MORBITIES OF PERINATAL ORIGIN	
Disease and/or group	*Specific diseases included in the group*
BILIRUBIN ENCEPHALOPATHY	
FETAL ALCOHOL SYNDROME	
HEPATIC FIBROSIS-RENAL CYSTS-INTELLECTUAL DISABILITY SYNDROME	

For each nosological group, groups of diseases are reported in bold and, if available, specific diseases belonging to the groups are indicated.