Summary of disease expertise recorded via RD-ACTION Matchmaker under each Thematic Grouping and EURORDIS Members' Thematic Grouping

Thematic Grouping	Reported expertise of those completing the matchmaker under each heading	EURORDIS Member perspectives on Grouping RD Thematically
Rare Bone	• Achondroplasia/Hypochondroplasia • Amelia skeletal dysplasia's including cleidocranial dysostosis, arthrogryposis • Brachydactyly chondrodysplasia punctate • Collagenopathy and oncologic disease such as Li-Fraumeni syndrome • Congenital hand and fore-foot conditions • Disorders of Sex Development • Duchenne Muscular Dystrophy • Ehlers –Danlos syndrome • Fibrodysplasia Ossificans Progressiva • Growth disorders • Hypoparathyroidism • Hypophosphatemic rickets & Nutritional Rickets • Hypophosphatasia • Jeune's syndrome • Limb reduction defects • Madelung disease • Metabolic Osteoporosis • Multiple Hereditary Exostoses • Osteogenesis imperfecta • Osteoporosis • Paediatric Osteoporosis • Paget's disease • Phocomelia • Pseudohypoparathyroidism • Radial dysplasia • Skeletal dysplasia • Thanatophoric dwarfism • Ulna dysplasia	• Achondroplasia • Achondroplasia/Growth hormone deficiency/MPS/Turner • Fibrous dysplasia of bone • Fibrodysplasia ossificans progressive • Osteogenesis imperfecta • Sterno Costo Clavicular Hyperostosis
Rare Cancer and Tumours	• Adrenocortical tumours • Carcinoid tumours • Craniopharyngioma • Embryonal tumours of CNS • Ependymoma • Epithelial thymic tumours in paediatric age • Ewing Sarcoma • Gliomas • Gonadal sex cord and stromal tumours • Hereditary breast and ovarian cancer due to BRCA1, BRCA2 and other gene mutations • Hereditary Cancer • Histiocitosis • Leukaemia	• Acute monoblastic leukaemia • Brain tumour • Colon cancer, familial nonpolyposis • Craniopharyngioma • Desmoid disease • Endocrine tumour • Leukaemia • Lymphoma • Multiple endocrine neoplasia • Multiple Myeloma • Paediatric cancers • Pseudomyxoma peritonei • Retinoblastoma • Systemic mastocytosis

Co-funded by the Health Programme of the European Union

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	• Lymphoma • Lung Cancer • Malignant peripheral nerve sheet tumour • Malignant Pleural Mesothelioma • Medulloblastoma • Melanoma • Mesothelioma • Myoepithelial carcinoma in children • Nasopharyngeal carcinomas • NETs • Neuro-endocrine Tumours • Neurofibromatosis • Pancreatic tumours (Pancreatoblastoma) • Paediatric extracranial germ cell tumours • Paediatric GIST • Paediatric liver tumours: Hepatoblastoma and Hepatocellular carcinoma • Paediatric Melanoma • Paediatric vascular tumours: Kaposiform hemangioendothelioma Pheochromocytoma and Paraganglioma • Paediatric very rare tumours: Gonadal sex cord and stromal tumours • Pilocytic astrocytoma • Pleuropulmonary Blastoma • Pons glioma • Rare head and neck Neurofibromatosis • Rare tumours • Retinoblastoma • Schwannomatosis • Thymoma • Thyroid carcinomas • Tuberous Sclerosis • Uveal Melanoma • Von Hippel Lindau disease	• Von Hippel-Lindau disease • Waldenström macroglobulinemia • X-linked lymphoproliferative disease
Rare Cardiac	• Adult congenital Heart Disease • Anomalous coronary arteries/ALCAPA • Aortic Pathology • Arrhythmogenic right ventricular dysplasia (DVAD) • Atresia of cardiac valves • Brugada syndrome • Common AV canal • Coarctation of aorta • Congenital heart defects • Dilated cardiomyopathy (CMD) • Double left and right ventricles • Ebstein's anomaly • Genetic/familial cardiomyopathies (like DCM,	• Brugada syndrome • Cardiac disease, rare • Cardiomyopathy, familial dilated • Heart malformation, congenital • Long QT syndrome, familial

	HCM, ARVC, LVNC) • Hypertrophic cardiomyopathy (CMH) • Hypoplastic left heart syndrome • Hypoplastic right heart syndrome • Inherited Cardiac diseases • Laminopathies • Long congenital QT syndrome • Paediatric Cardiology • Paediatric hypertrophic cardiomyopathy • Paediatric inherited arrhythmia syndromes • Paediatric pulmonary hypertension • Polymorphic ventricular tachycardia (TVPC) • Primary arrhythmia syndromes (like long QT syndrome, Brugada syndrome, idiopathic VF, CPVT). • Taussig-Bing anomaly • Tetralogy of Fallot • Transposition of great arteries • Turner Syndrome • Vascular Ring	
Rare Connective Tissues and Musculoskeletal	• AA amyloidosis • AL amyloidosis • ANCA vasculitis • Arthrogryposis multiplex congenital • ATTR amyloidosis • Bicuspid aortic valve with thoracic aortic aneurysm • Congenital club foot • Ectopia lentis • Ehlers-Danlos syndrome • Familial forms of BAV • Familial thoracic aortic aneurysms and dissections (FTAAD) • Giant Cell Arteritis • Goldenhar syndrome • Inflammatory myositis • Juvenile Scleroderma • Klippel-Feil syndrome • Larsen syndrome • LDS • Localized amyloidosis • Loeys-Dietz syndrome • Lupus • Lupus Nephritis • Marfan syndrome • Moebius syndrome • Neurofibromatosis • OAS • Paediatric rheumatic conditions	• Ehlers-Danlos syndrome • Marfan syndrome • Osteogenesis imperfecta • Scleroderma • Sjögren syndrome • Pseudoxanthoma elasticum

	• Pseduxanthoma Elasticum • SLE • Sprengel deformity • Stickler syndrome • Sturge-Weber • Systemic amyloidosis • Systemic Lupus Erythematosus • Systemic Sclerosis • Takayasu disease • Tuberous sclerosis	
Rare Craniofacial and ENT	• Aglossia • Anotia • Cleft lip • Cleft palate • Craniofacial tumours • Craniofacial syndromes (e.g. Crouzon, Apert etc.) • Craniosynostosis • Deafness mitochondrial • Facial microsomia • Facial Cleft • Genetic deafness: isolated and syndromic deafness • Genetic hearing loss and hearing impairment • Giant nevus and congenital cutaneous dysplasia • Hemifacial Microsomia • Isolated Craniosynostosis • Lateral Cleft • Macroglossia • Microglossia • Microdeletion syndromes connected with cleft lip and palate and with midline developmental anomalies • Microcephaly Encephaloceles including basal • Mandibulofacial Dysostosis • Microtia, Artesia and Craniofacial Microsomia • Midline dermoid • Nasal aplasia • Nose and skull base anomalies e.g. Treacher Collins, Goldenhar, Pierre robin, 22q11 microdeletion, CHARGE. • Oral commissure cleft • Orbitofacial cleft • Oto-Mandibular Dysplasia • Proboscis • Rare Maxillofacial surgical disorders • Rare otorhinolaryngologic malformations and laryngotracheal anomalies	• Apert syndrome • Crouzon disease • Pfeiffer • Esophageal atresia

	• Recurrent respiratory/Tracheal papillomatosis (RRP) • Syndrome Craniosynostosis • Van der Woude Syndrome • Vascular malformations	
Rare Endocrine	• 5 alpha reductase deficiency • Acromegaly • Addison's disease • Adrenal hormone deficiency • Adrenal disorders including Cushing syndrome, hypoadrenalism and Congenital Adrenal Hyperplasia • Adrenocortical cancer • Bone Disorders • Carney complex • Complete Androgen Insensitivity • Congenital Adrenal Hyperplasie • Cushing's Disease • Disorders of puberty • Disorders of Sex Development • Endocrine Hypertension • Familial Endocrine Tumour syndromes • Growth disorders including GH deficiency, Hypopituitarism and Septa-optic dysplasia • Hereditary endocrine tumours and tumour syndromes • Homozygous Familial Hypercholesterolemia • Klinefelter Syndrome • Lysosomal Acid Lipase deficiency • Macronodular adrenal hyperplasia • Monogenic diabetes • Multiple Endocrine Neoplasia Type 1 and Type 2 • Neonatal hyperinsulinism • Non-functioning pituitary adenoma • Paraganglioma • Partial Androgen insensitivity • Pheochromocytoma • Pituitary Diseases • Prader-Willi Syndrome • Rare causes of Primary Hyperaldosteronism • Rare congenital syndromes requiring endocrine treatment (growth hormone): Turner syndrome, Noonan, • Rare disorders of calcium metabolism • Rare forms of Diabetes • Rare neuroendocrine disorders such as late effects of brain tumours pheochromocytoma/paraganglioma- primary	• Acquired generalized lipodystrophy • Acromegaly • Addison's disease • Albright hereditary osteodystrophy • Alström syndrome • Cushing disease • Growth hormone deficiency, isolated, genetic forms • Hypoparathyroidism familial isolated • Hypothyroidism, congenital • Hypoparathyroidism familial isolated • Pituitary deficiency • Short stature due to growth hormone resistance • Turner syndrome

	aldosteronism- adrenal incidentaloma • Russell Silver Syndrome. • Sporadic Medullary Thyroid cancer • Tangier Disease • Thyroid disorders - thyroid dyshormonogenesis • Von Hippel Lindau	
Rare Eye Diseases	• Anophtalmos/microphthalmos • Anti-MOG optic neuritis and other forms of autoimmune mediated aetiologies of optic neuropathies • Chronic relapsing inflammatory optic neuropathy (CRION) • Congenital cataract • Congenital glaucoma • Congenital stationary night blindness • Genetic vitreous retinal disease • Inflammatory eye diseases with a special interest in non-infectious and infectious uveitis in adults and children • Isolated optic neuritis (ION) • Multiple sclerosis optic neuritis (MSON) • Neuromyelitis optica optic neuritis (NMO-ON) • Neurophthalmological disorders • Ocular myasthenia gravis • Oculocutaneous and ocular albinism and cerebral visual impairment in children • Optic neuropathies • Relapsing optic neuritis (RION) • Systemic diseases or genetic predisposition like Birdshot chorioretinopathy and HLA B27 associated uveitis	• Aniridia • WAGR syndrome • Blindness • Leber hereditary optic neuropathy • Retinitis pigmentosa • Septooptic dysplasia • Usher syndrome • Uveitis, adult
Rare Gastrointestinal	• ACNES • All Atresia's (oesophagus, duodenum, small and large intestine, anal) • Chronic pseudo-obstruction • Diaphragmatic hernia • Functional short bowel syndrome due to intestinal fistula following abdominal infection or surgery gastroschisis • M. Hirschsprung • Inflammatory bowel disease • Motility disorder • Necrotizing enterocolitis • Omphalocele • Radiation enteritis • Short bowel syndrome	• Crohn disease • Oesophageal Atresia • Familial adenomatous polyposis • Intestinal pseudoobstruction chronic idiopathic • Pancreatitis
Rare Gynaecological and Obstetric	• Adenocarcinoma • Malignant epithelial tumour of the ovary • Rare cancer of the cervix uteri, including	• Mayer-Rokitansky-Küster-Hauser syndrome

	squamous cell carcinoma • Rare vulvovaginal tumour • Squamous cell carcinoma of vulva	
Rare Haematological	• Aceruloplasminemia • All rare non-oncological/benign haematological diseases • Atransferrinemia • DMT-1 deficiency • Endoplasmic reticulum related defects leading to neutropenia • Genetic hyperferritinemia without iron overload • Haemophilia • Hereditary Hemochromatosis, with particular reference to rare forms of "non-HFE" Hemochromatosis • Hereditary Hyperferritinemia-Cataract Syndrome • Iron Loading Anaemias • Iron Refractory Anaemia (IRIDA) due to mutations in TMPRSS-6 • Multiple myeloma • Myelodysplastic syndrome • Polycythaemia • Rare anaemias • Severe chronic neutropenia type IV - G6PC3 deficiency • Sickle cell disease • Systemic mastocytosis • Thalassemia • Thrombotic microangiopathies such as thrombotic thrombocytopenic purpura (TTP) and haemolytic uremic syndrome (HUS) • Von Gierke disease type I • Von Willebrand Disease and other allied bleeding disorders	• Alpha-thalassemia / Beta-thalassemia • Angioedema, hereditary • Aplastic anaemia • Dyserythropoietic anaemia, congenital • Fanconi anaemia • Haematological diseases • Hemochromatosis • Haemophilia • Langerhans cell histiocytosis • Myeloproliferative disease, adult, rare • Paroxysmal nocturnal haemoglobinuria • Shwachman-diamond syndrome • Sickle cell anaemia • Thrombocytopenic purpura, autoimmune
Rare Hepatic	• Atresia of bile ducts • Autoimmune liver disease, to include PBS/PSC and AIH/AISC • Genetic liver disease, to include cholestasis in children and adults, Biliary atresia and ALF in Infants. Non-alcoholic fatty liver disease (NAFLD) • Rare liver cancers, to include cholangiocarcinoma and fibrolamellar HCC • Wilson's Disease	• Hepatic diseases • Metabolic liver disease, paediatric, rare • Primary sclerosing cholangitis • Wilson's disease

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Rare Hereditary Metabolic	• 3-methylcrotonyl glycinuria • 3-methylglutaconic aciduria • 3-oxothiolase deficiency • Biotinidase deficiency • Carnitine transport disorders (carnitine uptake deficiency, CPT I-II, SCAD, MCAD, LCHAD, VLCADCGD • Citrullinemia • Disorders of mitochondrial fatty acid oxidation • Fabry disease • Galactosaemia • Gaucher's disease • Hepatic glycogen storage disorders • Hepatic metabolic diseases • Homocystinuria • Lysosomal Acid Lipase Deficit • Lysosomal storage disorders • Metabolic myopathies • Niemann-Pick A/B • Niemann-Pick C • Organic acidemias (propionic academia) • Organic acidurias (propionic aciduria, methylmalonic aciduria, glutaric acidurias, HMG-CoA lyase deficiency). • Peroxisomal defects • Phenylketonuria • Sanfilippo Syndrome • Smith-Lemli Opitz syndrome • Tetrahydrobiopterin defects • Tyrosinemia • Urea Cycle Disorders • Von Gierke disease	• Acyl-CoA dehydrogenase, medium chain, deficiency • Adrenoleukodystrophy, X-linked • Alkaptonuria • CDG syndrome • Cystinosis • Fabry disease • Gaucher's disease • Glycogen storage disease • Hypophosphatasia • Hyperglycinemia, isolated nonketotic • Lesch-Nyhan syndrome • Lysosomal disease • Mitochondrial disease • Mucopolysaccharidosis • Niemann-Pick • Phenylketonuria • Sanfilippo syndrome type C (Mucopolysaccharidose type 3C) • Tay-Sachs disease/Sandhoff disease • Trimethylaminuria
Rare Immunological and Auto-inflammatory	• ANCA vasculitis • Auto inflammatory diseases such as FMF, HIDS, TRAPS, CAPS, angioedema, hereditary angioedema (HAE). • Autoimmune bullous dermatoses • Behcet's Disease • Benign Hypermobility syndrome • C1-inhibitor deficiency • Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) • Connectives tissues diseases, particularly systemic sclerosis inflammatory myositis • Erythromelalgia • Freiberg's disease • Giant Cell Arteritis • Growing Pains/'recurrent nocturnal limb pain in children'	• Amyloidosis • Familial hemophagocytic lymphohistiocytosis (FHL) • Immunodeficiency, primary • Juvenile arthritis, idiopathic • Lupus erythematosus • Mediterranean fever, familial • Muckle-Wells syndrome • Relapsing polychondritis • SAPHO syndrome • Scleroderma • Sjögren syndrome

	• Guillain-Barre Syndrome (GBS) • Henoch-Scholein Purpura • Juvenile Dermatomyositis • Juvenile Idiopathic Arthritis (JIA) • Juvenile Spondyloarthritis/Enthesitis Related Arthritis (Spa-Era) • Kawasaki disease • Legg-Calvé-Perthes disease • Limb Pain syndromes (Fibromyalgia syndrome) • Localized Idiopathic Musculoskeletal Pain syndrome • Lyme disease • MALT lymphoma associated with Sjogren's disease • Multifocal Motor Neuropathy (MMN) • Myasthenia Gravis (MG) • Osgood-Schlatter Disease • Osteochondrosis • Patellofemoral Pain • Primary Immunodeficiencies • Rare Juvenile Primary Systemic Vasculitis • Rare systemic autoimmune vasculitis, including ANCA vasculitis, EGPA, anti-GBM disease, Takayasu's arteritis, Cogan's syndrome • Rheumatic Fever • Scheuermann's disease • Sever's disease • Sjogren's disease (including MALT lymphoma) • Systemic amyloidosis • Slipped Capital Femoral Epiphysis • Systemic autoimmune disease • Systemic lupus erythematosus (SLE) • Systemic sclerosis (SSC) • Takayasu's disease • Transient Synovitis • Tumour of hematopoietic and lymphoid tissues	
Rare Malformations, Congenital Anomalies and rare intellectual disorders	• Anorectal Malformations • Asperger syndrome • BAF complex disorders (e.g. Coffin-Siris) • Biliairy Atresia • Beckwith-Wiedemann syndrome • Bowel Atresia • CHARGE syndrome • Cohesinopathies • Cornelia de Lange syndrome • Developmental anomalies • Down syndrome • Filippi syndrome • Fragile-X syndrome	• Angelman syndrome • CHARGE association • Chromosome anomalies • Ciliopathies • Cornelia de Lange syndrome • Costello syndrome • Embryopathy • Foetal valproic syndrome • Fragile X syndrome • Intellectual deficiency with developmental anomaly, rare • Jacobsen syndrome • Joubert syndrome

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	• Gastroschisis • Genetic Intellectual Disability • Loeys-Dietz syndrome • Marshall-Smith syndrome • Microdeletion syndromes in connection with developmental anomalies & rare intellectual disabilities • Multiple anomaly syndromes (RAS-MAP, Cohen, Williams, Ohdo, Genitopatellar, Kabuki, Treacher Collins and other spliceosome disorders). • Ochoa syndrome • Perrault syndrome, • Phelan-McDermid syndrome • Pitt-Hopkins syndrome • Rare chromosome disorders, in particular Phelan-McDermid syndrome • Rare intellectual disability syndromes including Angelman, Pitt Hopkins, Rett, Mowat-Wilson, X linked ID. • RASopathies • Rubinstein-Taybi syndrome • Undiagnosed multiple anomaly syndromes	• Klinefelter syndrome • Limb hypoplasia - limb reduction defect • Lowe syndrome • Marshall-Smith syndrome • McCune-Albright syndrome • Moebius syndrome • Moebius syndrome, Monosomy 22q11, Monosomy 5p • Noonan syndrome • Poland anomaly • Prader-Willi syndrome • Ring chromosome 20 • Spina bifida • Syringomyelia • Tetrasomy 12p • Thalidomide • Trisomy 18 • Williams syndrome
Rare Multisystemic Vascular	• Aneurysm Osteoarthritis syndrome • Aortopathies • Arterial Tortuosity syndrome • Beals syndrome • Bicuspid Aortic Valve disease • Buerger Arteritis • Congenital Heart disease • Cutis Laxa syndrome • Ectopia Lentis • Fabry disease • Familial thoracic aortic aneurysms • Heritable Thoracic Aortic disease • Kawasaki disease • Klippel–Trénaunay syndrome • Klinefelter syndrome • Inborn lymphatic diseases • Loeys Dietz syndrome • Marfan syndrome • OAS • Primary lymphedema • Primitive Cardiomyopathy • Pulmonary Hypertension • Takayasu disease • Turner syndrome • Vascular Ehlers Danlos syndrome	• Behcet disease • Cerebral cavernous malformations • Familial aortic dissection • Rendu-Osler-Weber disease • Sturge-Weber syndrome • Vascular disease, rare Vasculitis
Rare	• 4H syndrome	• Aicardi syndrome

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Neurological	• ADEM • Autoimmune encephalitis • Autoimmune epilepsies • Brain Arteriovenous malformation (BAVM) • Cerebrovascular malformations • Chronic relapsing inflammatory optic neuropathy (CRION) • Complex occlusive cerebral arteriopathies • Complex paediatric cerebrovascular disease • Disorders mimicking cerebral palsy • Dural Arteriovenous Fistula (DAVF) • Dystonia • Early Infantile Epileptic Encephalopathies • Epilepsy with continuous spike-waves during sleep • Functional movement disorders • Genetic epilepsies • Hereditary ataxias • Hereditary spastic paraplegias • Huntington disease • Hyperekplexia • Hypomyelination and LBSL • Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum (H-ABC) • Infantile epilepsy syndromes • Infantile Spasms • Inherited white matter disorders • Isolated optic neuritis (ION) • Lambert-Eaton syndrome • Lennox-Gastaut syndrome • Leukodystrophies • Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC) • Metachromatic Leukodystrophy (MLD) • Mitochondrial disorders • Motor neuron disorders • Movement disorders • Moyamoya • Multiple sclerosis associated optic neuritis (MSON) • Muscle disorders • Myasthenia gravis • Myoclonus • Narcolepsy-catalepsy • NBIA • Neurodegeneration with brain iron accumulation • Neurodegenerative disorders (early onset Parkinson disease and rare dementias)	• Alternating hemiplegia • Amyotrophic lateral sclerosis • Anoxic seizures • Arachnoid cyst • Arnold-Chiari malformation • Aromatic L-aminoacid decarboxylase deficiency • Ataxia, adult • Arnold-Chiari malformation • Cerebellar ataxia, autosomal dominant • Ceroid lipofuscinosis, neuronal • Complex regional pain syndrome • Dravet syndrome • Dystonia • Facioscapulohumeral muscular dystrophy • Friedreich ataxia • Gelineau disease • Hereditary spastic paraplegia, autosomal dominant complex • Huntington disease • Juvenile neuronal ceroid lipofuscinosis (Batten disease, Spielmeyer-Vogt, disease, CLN3) • Kleine-Levin syndrome • Leukodystrophy • Locked-in syndrome • Neurofibromatosis • Multiple sclerosis • Multiple system atrophy • Myasthenia gravis • Neuroacanthocytosis • Neurodegeneration with brain iron accumulation (NBIA) • Ondine-Hirschsprung disease • Opsoclonus-myoclonus syndrome • PANDAS • Perineural cyst • Post-poliomyelitic syndrome • Proximal spinal muscular atrophy • Rett syndrome • Ring chromosome 14 • Spastic paraplegia, familial • Stiff-man syndrome • Syringomyelia • Tourette syndrome • Transmissible spongiform encephalopathies

	• Neurometabolic disease related to movement disorders • Neuromyelitis optica optic neuritis (NMO-ON, AQP4) • Neuro-ophthalmological disorders • Neurosarcoidosis • Niemann Pick C • Opsoclonus-myoclonus syndrome • Optic neuropathies • Paroxysmal dyskinesias- polyradiculitis • Rare cerebrovascular disorders • Rare Neuroimmune disorders • Rare neuropathies • Relapsing isolated optic neuritis (RION) • Spectrum (MOG) • Spinal Cord Vascular malformation • Stiff-person syndrome • Subarachnoid haemorrhage (SAH) • Tuberose sclerosis • Vanishing White Matter (VWM) • Vein of Galen malformation • Venous malformation (Cavernomas/DVA) • West syndrome	• Tremor hereditary essential • Tuberous sclerosis • undiagnosed neurological • West syndrome
Rare Neuromuscular	• Anterior Cutaneous Nerve Entrapment syndrome • Amyotrophic lateral sclerosis • Arthrogryposis • Charcot-Marie-Tooth • Congenital myopathies • Congenital myasthenia • Congenital hydrocephalus • Dandy-Walker • Distal myopathies • Friedreich-ataxia • Hereditary myopathies • Immune-mediated conditions such as myasthenia gravis • Inflammatory neuropathies (e.g. CIDP) • Inherited muscle disease; - e.g. Duchenne Muscular Dystrophy, Other LGMD, Congenital Muscular Dystrophies, Congenital Myopathies, Congenital Myasthenic Syndromes, Myotonia Congenita and other myotonias and paramyotonias. • Lissencephaly • Mitochondrial muscle diseases such as Leigh, Alpers, Kearns-Sayre, MELAS, and MERRF syndromes • Myofibrillar myopathies	• Amyotrophic lateral sclerosis • Becker MD • Charcot-Marie-Tooth disease (generic term) • Chronic inflammatory demyelinating polyneuropathy • Duchenne Muscular Dystrophy • Myotubular myopathy • Neuromuscular disease

	• Neural tube defects (encephalocele, spina bifida) • Paediatric and adult neuromuscular disorders • Peripheral neuropathies • Rimmed vacuolar myopathies • Spinal Muscular Atrophies • Transthyretin Familial Polyneuropathy • Tendinopathy	
Rare Pulmonary	• ARDS • Chronic thromboembolic pulmonary hypertension • Congenital Cystic Adenomatoid malformations • Cystic fibrosis • Idiopathic Pulmonary Fibrosis Lymphangioleiomyomatosis • Interstitial Lung diseases • Langerhans Histiocytosis • Paediatric Pulmonary Hypertension • Primary ciliary dyskinesia • Pulmonary alveolar proteinosis • Pulmonary Arterial Hypertension • Pulmonary Metastases • Pulmonary Thrombo Endarterectomy (PTE) surgery • Sarcoidosis • Severe Acute Respiratory Failure in Adults (Respiratory ECMO)	• Alpha-1 antitrypsin and alpha-1-antichymotrypsin deficiencies • Cystic fibrosis • Gorham-Stout disease • Lymphangioleiomyomatosis • Pulmonary arterial hypertension • Pulmonary fibrosis, idiopathic • Sarcoidosis
Rare Renal	• Alport Syndrome • Atypical haemolytic uremic syndrome • Chronic kidney disease and renal replacement therapy in children • Ciliopathies • Cystinosis • Dens Deposit disease • Hereditary kidney diseases as congenital nephrotic syndrome, nephronophthisis, cystinosis, Oxalosis Inherited glomerulopathies (podocytopathies, Alport syndrome, MPGN); Fabry, Tuberous Sclerosis, FSGS, ARPKD, OFD, UMOD, MUC1, HNF1b, REN, HANAC. • Inherited Glomerulopathies • Inherited tubulopathies • Meckel-Gruber syndrome • Metabolic diseases affecting the kidney (hyperoxaluria, cystinosis, mitochondriopathies). • Multicystic renal dysplasia • Nephronophthisis and other inherited and sporadic forms of kidney dysplasia	• Cystinosis • Nephrotic syndrome congenital and infant form • Nephrotic syndrome, idiopathic, steroid-resistant, familial • Polycystic kidney disease • Renal disease, genetic

	• Nephrotic Syndrome • Oxalosis • Polycystic kidney Disease • Renal malformations • Renovascular hypertension in children • Syndromic and non-syndromic kidney malformations, including autosomal recessive and dominant polycystic kidney disease (ARPKD; ADPKD), • Tuberous sclerosis complex • Tubular diseases • Tubulopathies	
Rare Skin	• Anhidrotic Ectodermal Dysplasia with Immunodeficiency • Aplasia cutis congenital • ARCI • Atypical Fibroxanthoma • Autoimmune Skin diseases: lupus, scleroderma • Auto inflammatory skin disease • Basal Cell Nevus syndrome • Birt-Hogg-Dubé syndrome • Bullous autoimmune diseases • Bullous dermatoses • Collagen vascular disorders • Connective tissue disorders • Cutaneous diseases related to DNA Repair disorders • Cutaneous localized overgrowth syndromes • Cutaneous Lymphoma • Cutaneous Mosaic disorders • Cutaneous Vascular anomalies • Cutis Laxa • Cutis Marmorata Telangiectasia • Dermatitis Herpetiformis • Dermatofibrosarcoma protuberans • Ectodermal Dysplasias • Ehlers-Danlos syndromes • Epidermolysis bullosa (adult and paediatric) • Genodermatoses: Netherton Syndrome, Darier Disease, Gorlin Syndrome, CEB. • Hemangioma vascular formations • Hereditary and Acquired Angioedema • Hereditary tumour syndromes • Histaminergic and recurrent idiopathic Angioedema • Ichthyosis • Incontinentia pigmenti • Inflammatory and Granulomatous Dermatoses • KLICK	• Cutis laxa • Cutis marmorata telangiectatica congenital • Ectodermal dysplasia syndrome • EEC syndrome • Epidermolysis bullosa • Giant pigmented hairy nevus • Ichthyosis • Incontinentia pigmenti • Oculocutaneous albinism • Papulosis, malignant atrophic • Partington disease • Pemphigus vulgaris • Pemphigus, benign chronic familial • Porphyria • Pseudoxanthoma elasticum • Xeroderma pigmentosum

	• Keratinizing Disorders: Mb Darier, Connexin disorders, Peeling Skin disorders. • Linear Morphea • Mal de Meleda • Marie-Unna hypotrichosis • Mastocytosis • Mendelian Disorders of Cornification (MEDOC) • Mendelian Skin and Nail diseases • Merkel Cell Carcinoma • Monilethrix • Monogenic connective tissue disorders • Morbus Darier • Morbus Hailey-Hailey • Neurocutaneous disorders: NF1, NF2, Schwannomatosis, Tuberous sclerosis complex. • Neurofibromatosis • Nevus Andreu nevoid conditions • Nevi & Nevoid Skin conditions • Non-Histaminergic Angioedema • Pachyonychia Congenita • Palmoplantar keratoderma • PIK3CA related disorders and interrelated pathways • Pigmentary and Vascular Keratodermas • Phacomatoses • Porphyria vascular formations • Premature skin ageing • Pseudoxanthoma elasticum • Rare skin tumours • Sebaceous Gland Carcinoma • Skin Fragility disorders • Toxic epidermal necrolysis • Tuberous Sclerosis Complex • Valvular malformations • Vascular tumours: Kaposiform hemangioendothelioma and Tufted Angioma. • Palmoplantar keratodermas (PPKs)	
Rare Urogenital	• Bladder exstrophy • Cloacal exstrophy • Complicated and complex pelvic floor disorders (including POP and male and female incontinence) • Cystinuria • Failed secondary urethral surgery • Female strictures • Functional urology requiring highly specialised surgery • Indeterminate sex • Lynch syndrome	• Anorectal malformation • Bladder exstrophy • Epispadias • Interstitial cystitis

	• Mesh repairs • Non-syndromic urogenital tract malformation of male and female e.g. in spina bifida • Oxaluria • Penile cancer • Polycystic disease • Post pelvic fracture surgery • Posterior urethral valves • Rare congenital urogenital anomalies, including anorectal malformations and the ARM network • Rare Urogenital tumours • Recto urinary fistulae • Renal, ureteral and bladder amyloidosis • Retroperitoneal Fibrosis • Retroperitoneal Sarcomas • Severe types of Hypospadias • Sex disorder developments • Testicular cancer • Ureterolysis • Urethral diverticula in women • Urethroplasty • Vesicovaginal fistulae • Von Hippel Lindau disease	