Orphanet Report Series

Rare Diseases collection

2021

Expert reviewers for Orphanet in 2021

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METHODOLOGY

This document provides the list of Orphanet expert reviewers having contributed to the update and quality control of scientific information contained in the Orphanet database of Rare Diseases in 2020.

Experts were identified through their publications, and their activity related to the given disease/group of diseases (research projects, clinical trials, expert centres and dedicated networks): more information on the expert selection procedure will be available soon on www.orpha.net. Once identified, experts receive an invitation to examine, validate, correct or complete scientific information related to a given disease and produced based on peer-reviewed publications. Experts are solicited for their input on one, or a number, of the following:

- Nomenclature: preferred term and synonyms
- Related genes and the type of relationship between the gene and the diseases, namely: genes are annotated as causative, modifiers (both from germline or somatic mutations), major susceptibility factors or playing a role in the phenotype (for chromosomal anomalies). When the causative mutations are of germline origin, the functional consequences of the mutations could be documented as a loss or a gain of protein function
- Epidemiological data: point prevalence, annual incidence, prevalence at birth, lifetime prevalence, number of cases and families reported in the literature.
- Natural history data: age of onset, age of death, mode of inheritance
- An abstract structured in up to 10 sections :
    - Definition of the disease
    - Epidemiology
    - Clinical description
    - Aetiology
    - Diagnostic methods
    - Differential diagnosis
    - Genetic counseling (if relevant)
    - Antenatal diagnosis (if relevant)
    - Management and treatment
    - Prognosis
- Disability facts related to rare diseases.
- Guidelines for emergency care in rare diseases

In some cases, the experts are contacted to answer a specific question (nomenclature, genetics, classifications, or epidemiological data) in order to update the Orphanet content, or to carry out quality control of the data. Please note that the experts contacted to answer this type of question are not cited on the disease page on the Orphanet website: the expert cited on the disease page is the one having contributed to the text concerning the disease.

Data presentation

Experts are listed by alphabetical order of their name and with the expertised disease name and ORPHA code of the disease/ group of diseases.

Expert networks having contributed to the update of the nomenclature and are described in a second list.

List of expert reviewers for Orphanet in 2021

Expert	Disease name	ORPHAcode
Dr ALSAFI Ali	Pulmonary arteriovenous malformation	ORPHA:2038
Dr ANQUETIL Céline	Inclusion body myositis	ORPHA:611
	Dermatomyositis	ORPHA:221
	Juvenile dermatomyositis	ORPHA:93672
	Antisynthetase syndrome	ORPHA:81
	Overlap myositis	ORPHA:206572
	Immune-mediated necrotizing myopathy	ORPHA:206569
Pr ARICETA Gema	Dent disease type 1	ORPHA:93622
	Dent disease type 2	ORPHA:93623
	Idiopathic hypercalciuria	ORPHA:2197
Dr ARNOUX Jean-Baptiste	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
Pr ARZIMANOGLOU Alexis	Lennox-Gastaut syndrome	ORPHA:2382
	Tuberous sclerosis complex	ORPHA:805
Pr ATTIE-BITACH Tania	Meckel syndrome	ORPHA:564
	Acrocallosal syndrome	ORPHA:36
Pr BAAS Frank	Pontocerebellar hypoplasia type 2	ORPHA:2524
	Pontocerebellar hypoplasia type 1	ORPHA:2254
	Pontocerebellar hypoplasia type 3	ORPHA:97249
	Pontocerebellar hypoplasia type 6	ORPHA:166073
	Pontocerebellar hypoplasia type 4	ORPHA:166063
	Non-syndromic pontocerebellar hypoplasia	ORPHA:98523
	Pontocerebellar hypoplasia type 13	ORPHA:613267
	Pontocerebellar hypoplasia type 12	ORPHA:611256
	Pontocerebellar hypoplasia type 11	ORPHA:611247
	Pontocerebellar hypoplasia type 14	ORPHA:613274
Dr BAGOU Gilles	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
	Kawasaki disease	ORPHA:2331
	Sturge-Weber syndrome	ORPHA:3205
	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
	Dravet syndrome	ORPHA:33069
	Fragile X syndrome	ORPHA:908
	Brugada syndrome	ORPHA:130
	Hereditary hemorrhagic telangiectasia	ORPHA:774
	Non-histaminic angioedema	ORPHA:658
	Hypophosphatasia	ORPHA:436
	Alpha-1-antitrypsin deficiency	ORPHA:60
	Thrombotic microangiopathy	ORPHA:93573
	Sickle cell anemia	ORPHA:232
Pr BAHI-BUISSON	Rett syndrome	ORPHA:778

Expert	Disease name	ORPHAcode
Nadia	Microcephalic cortical malformations-short stature due to RTTN deficiency	ORPHA:468631
	Polymicrogyria due to TUBB2B mutation	ORPHA:300573
Dr BARCIA Giulia	Malignant migrating focal seizures of infancy	ORPHA:293181
Pr BARIC Ivo	S-adenosylhomocysteine hydrolase deficiency	ORPHA:88618
Dr BARTULI Andrea	8p inverted duplication/deletion syndrome	ORPHA:96092
Pr BATTAGLIA Agatino	Wolf-Hirschhorn syndrome	ORPHA:280
	1p36 deletion syndrome	ORPHA:1606
Dr BAUJAT Geneviève	Antley-Bixler syndrome	ORPHA:83
	Hypophosphatasia	ORPHA:436
Dr BEDESCHI Maria Francesca	Moebius syndrome	ORPHA:570
	Campomelic dysplasia	ORPHA:140
Pr BENVENISTE Olivier	Inclusion body myositis	ORPHA:611
	Dermatomyositis	ORPHA:221
	Antisynthetase syndrome	ORPHA:81
	Juvenile dermatomyositis	ORPHA:93672
	Immune-mediated necrotizing myopathy	ORPHA:206569
	Overlap myositis	ORPHA:206572
Dr BERNIT Emmanuelle	Sickle cell anemia	ORPHA:232
Pr BETZ Regina	Marie Unna hereditary hypotrichosis	ORPHA:444
Dr BIALAS Rebecca	Hypoplasminogenemia	ORPHA:722
Pr BIESECKER Leslie	Microphthalmia-ankyloblepharon-intellectual disability syndrome	ORPHA:85275
Dr BIGONI Stefania	Coffin-Lowry syndrome	ORPHA:192
	Bohring-Opitz syndrome	ORPHA:97297
	KLHL7-related disorder	ORPHA:603699
Dr BISDORFF-BRESSON Annouk	Sturge-Weber syndrome	ORPHA:3205
Pr BISHOP Nick	Osteogenesis imperfecta	ORPHA:666
	Osteogenesis imperfecta type 2	ORPHA:216804
	Osteogenesis imperfecta type 5	ORPHA:216828
	Osteogenesis imperfecta type 3	ORPHA:216812
	Osteogenesis imperfecta type 1	ORPHA:216796
Pr BJORNSSON Hans	Wiedemann-Steiner syndrome	ORPHA:319182
Pr BLAU Nenad	Classic phenylketonuria	ORPHA:79254
	Mild hyperphenylalaninemia	ORPHA:79651
	Tetrahydrobiopterin-responsive hyperphenylalaninemia/phenylketonuria	ORPHA:293284
Pr BLOCH-ZUPAN Agnès	Hypophosphatasia	ORPHA:436
Dr BOBER Michael	Thanatophoric dysplasia type 1	ORPHA:1860
	Thanatophoric dysplasia type 2	ORPHA:93274
Dr BOCCARA Olivia	Sturge-Weber syndrome	ORPHA:3205
Dr BOCCON-GIBOD Isabelle	Non-histaminic angioedema	ORPHA:658

Expert	Disease name	ORPHAcode
Pr BOCKENHAUER Detlef	Tubular renal disease-cardiomyopathy syndrome	ORPHA:73224
Dr BOUCHEREAU Juliette	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
Pr BOUILLET Laurence	Non-histaminic angioedema	ORPHA:658
	OBSOLETE: ANGPT1-related hereditary angioedema with normal C1Inh	ORPHA:537891
	Hereditary angioedema with normal C1Inh not related to F12 or PLG variant	ORPHA:599418
Dr BOUNAUD Nicolas	Alpha-1-antitrypsin deficiency	ORPHA:60
Pr BOVÉE Judith	Multiple osteochondromas	ORPHA:321
Pr BOYER Olivia	Genetic steroid-resistant nephrotic syndrome	ORPHA:656
Pr BRANDI Maria Luisa	Familial medullary thyroid carcinoma	ORPHA:99361
	Multiple endocrine neoplasia type 2	ORPHA:653
	Multiple endocrine neoplasia type 2A	ORPHA:247698
Dr BRENA Michela	Severe generalized junctional epidermolysis bullosa	ORPHA:79404
	Autosomal dominant generalized dystrophic epidermolysis bullosa	ORPHA:231568
	Junctional epidermolysis bullosa with pyloric atresia	ORPHA:79403
Dr BROOKS Alice	Hirschsprung disease	ORPHA:388
Dr BRUNELLE Perrine	Ulnar-mammary syndrome	ORPHA:3138
Dr BRUNKLAUS Andreas	Rett syndrome	ORPHA:778
Pr CAMPEAU Philippe	CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	ORPHA:599082
Pr CAREL Jean-Claude	Turner syndrome	ORPHA:881
Dr CHAUVIN Antony	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
	Kawasaki disease	ORPHA:2331
	Sturge-Weber syndrome	ORPHA:3205
	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
	Dravet syndrome	ORPHA:33069
	Fragile X syndrome	ORPHA:908
	Brugada syndrome	ORPHA:130
	Hereditary hemorrhagic telangiectasia	ORPHA:774
	Non-histaminic angioedema	ORPHA:658
	Alpha-1-antitrypsin deficiency	ORPHA:60
	Hypophosphatasia	ORPHA:436
	Thrombotic microangiopathy	ORPHA:93573
	Sickle cell anemia	ORPHA:232
Dr CHEMALY Nicole	Dravet syndrome	ORPHA:33069
Pr CHRISTIN-MAITRE Sophie	Turner syndrome	ORPHA:881
Pr CHRZANOWSKA Krystyna	Nijmegen breakage syndrome	ORPHA:647
Dr CLARET Pierre-Géraud	Hypophosphatasia	ORPHA:436

Expert	Disease name	ORPHAcode
Dr COARELLI Giulia	Spastic paraplegia type 7	ORPHA:99013
	Autosomal dominant spastic paraplegia type 38	ORPHA:171617
	Autosomal dominant spastic paraplegia type 48	ORPHA:306511
	Autosomal dominant spastic paraplegia type 30	ORPHA:101010
	Autosomal dominant spastic paraplegia type 13	ORPHA:100994
	Autosomal dominant spastic paraplegia type 10	ORPHA:100991
	Autosomal dominant spastic paraplegia type 8	ORPHA:100989
	Autosomal dominant spastic paraplegia type 6	ORPHA:100988
	Autosomal dominant spastic paraplegia type 3	ORPHA:100984
Dr CONTRO Gianluca	Nicolaides-Baraitser syndrome	ORPHA:3051
Pr COPPO Paul	Thrombotic thrombocytopenic purpura	ORPHA:54057
Pr CORMIER-DAIRE Valérie	Weaver syndrome	ORPHA:3447
Pr CORMIER-DAIRE, Pr JACKSON, Pr ROBERTSON	HEYN-SPROUL-JACKSON SYNDROME	#N/A
Dr COUCHONNAL Edouardo	Wilson disease	ORPHA:905
	MEDNIK syndrome	ORPHA:171851
Dr DAINA Erica	Immunoglobulin-mediated membranoproliferative glomerulonephritis	ORPHA:329903
	C3 glomerulopathy	ORPHA:329918
	Dense deposit disease	ORPHA:93571
	Primary membranoproliferative glomerulonephritis	ORPHA:54370
Pr DARDIS Andrea	Mesomelia-synostoses syndrome	ORPHA:2496
Pr DARGAUD Yesim	Moderate hemophilia B	ORPHA:169796
	Moderate hemophilia A	ORPHA:169805
	Severe hemophilia A	ORPHA:169802
	Severe hemophilia B	ORPHA:169793
	Factor V Atlanta bleeding disorder	ORPHA:600194
	Combined deficiency of factor VII and factor X	ORPHA:600691
	Factor V short isoforms-related bleeding disorder	ORPHA:599519
	Acquired factor XIII deficiency	ORPHA:599513
	Acquired factor XI deficiency	ORPHA:599507
	Acquired factor X deficiency	ORPHA:599501
	Acquired factor VII deficiency	ORPHA:599495
	Acquired factor V deficiency	ORPHA:599490
	OBSOLETE: Acquired hemophilia	ORPHA:73274
	Acquired hemophilia B	ORPHA:599485
	Acquired hemophilia A	ORPHA:599480
	Factor V Amsterdam bleeding disorder	ORPHA:599579
Pr DE LONLAY Pascale	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
Pr DEBRAY Dominique	Wilson disease	ORPHA:905
Dr DEGIORGIS Valentina	Ring chromosome 20 syndrome	ORPHA:1444

Expert	Disease name	ORPHAcode
Dr DEGRUGILLIER-CHOPINET Caroline	Sturge-Weber syndrome	ORPHA:3205
Pr DEJACO Christian	Giant cell arteritis	ORPHA:397
Dr DENJOY Isabelle	Brugada syndrome	ORPHA:130
Pr DES PORTES Vincent	Fragile X syndrome	ORPHA:908
Pr DEVUYST Olivier	Autosomal dominant tubulointerstitial kidney disease	ORPHA:34149
Dr DIAS Patricia	Branchio-oculo-facial syndrome	ORPHA:1297
Dr DIAS Patricia	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	ORPHA:457193
Dr DONADILLE Bruno	Turner syndrome	ORPHA:881
Dr DOUILLET Delphine	Sturge-Weber syndrome	ORPHA:3205
Dr DRAAISMA J.M.T.	Brain malformations-musculoskeletal abnormalities-facial dysmorphism-intellectual disability syndrome	ORPHA:500150
Dr DUIJKERS Floor	Wiedemann-Steiner syndrome	ORPHA:319182
Dr DUPUIS-GIROD Sophie	Hereditary hemorrhagic telangiectasia	ORPHA:774
Pr DURR Alexandra	Spastic paraplegia type 7	ORPHA:99013
	Autosomal dominant spastic paraplegia type 38	ORPHA:171617
	Autosomal dominant spastic paraplegia type 48	ORPHA:306511
	Autosomal dominant spastic paraplegia type 30	ORPHA:101010
	Autosomal dominant spastic paraplegia type 13	ORPHA:100994
	Autosomal dominant spastic paraplegia type 10	ORPHA:100991
	Autosomal dominant spastic paraplegia type 8	ORPHA:100989
	Autosomal dominant spastic paraplegia type 6	ORPHA:100988
	Autosomal dominant spastic paraplegia type 3	ORPHA:100984
Pr EDERY Patrick	Roifman syndrome	ORPHA:353298
Pr El-Ghoneimi Alaa	Isolated female hypospadias	ORPHA:603515
Pr EMMA Francesco	Dent disease type 1	ORPHA:93622
	Dent disease type 2	ORPHA:93623
Dr ENGELEN Marc	X-linked adrenoleukodystrophy	ORPHA:43
	Adrenomyeloneuropathy	ORPHA:139399
	X-linked cerebral adrenoleukodystrophy	ORPHA:139396
Dr ENGWERDA Aafke	Polyvalvular heart disease syndrome	ORPHA:228410
Pr F.SCHREUDER Michiel	Oligomeganephronia	ORPHA:2260
Dr FAERGEMAN Soren	Hajdu-Cheney syndrome	ORPHA:955
Dr FASCETTILEON Francesco	Atresia of small intestine	ORPHA:1201
Pr FERLINI Alessandra	Coffin-Lowry syndrome	ORPHA:192
	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	ORPHA:502423
	Bohring-Opitz syndrome	ORPHA:97297
	KLHL7-related disorder	ORPHA:603699
Dr FRANÇOIS Mathilde	Hereditary hemorrhagic telangiectasia	ORPHA:774

Expert	Disease name	ORPHAcode
Dr GAIGNARD Pauline	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	ORPHA:217371
Pr GALACTEROS Frédéric	Sickle cell anemia	ORPHA:232
	Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome	ORPHA:251380
	Sickle cell-hemoglobin E disease syndrome	ORPHA:251375
	Sickle cell-hemoglobin D disease syndrome	ORPHA:251370
Pr GAMBARO Giovanni	Medullary sponge kidney	ORPHA:1309
Dr GARAVELLI Livia	Myhre syndrome	ORPHA:2588
	Blepharophimosis-intellectual disability syndrome, SBBYS type	ORPHA:3047
	Nicolaides-Baraitser syndrome	ORPHA:3051
	Blepharophimosis-intellectual disability syndrome/genitopatellar overlap syndrome	ORPHA:597746
	KAT6B-related multiple congenital anomalies syndrome	ORPHA:597749
	Reunion Island Larsen-like syndrome	ORPHA:294049
Pr GIBBONS Richard	Alpha-thalassemia-X-linked intellectual disability syndrome	ORPHA:847
Dr GIL-JARDINE Cédric	Sickle cell anemia	ORPHA:232
Dr GIUSTI Francesca	Multiple endocrine neoplasia type 2	ORPHA:653
Pr GOADSBY Peter	SUNCT syndrome	ORPHA:57145
	Hemicrania continua	ORPHA:443070
Dr GOLDENBERG Alice	Cornelia de Lange syndrome	ORPHA:199
Pr GONZALES Emmanuel	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	ORPHA:217371
Pr GOUDEMAND Jenny	Von Willebrand disease type 3	ORPHA:166096
	Von Willebrand disease type 2	ORPHA:166081
	Von Willebrand disease type 2B	ORPHA:166087
	Von Willebrand disease type 2A	ORPHA:166084
	Von Willebrand disease type 2M	ORPHA:166090
	Von Willebrand disease type 2N	ORPHA:166093
Dr GRANATA Tiziana	Progressive myoclonic epilepsy type 1	ORPHA:308
Dr GREGERSEN Pernille	Williams syndrome	ORPHA:904
	Hajdu-Cheney syndrome	ORPHA:955
Pr GRIESE Matthias	Autoimmune pulmonary alveolar proteinosis	ORPHA:747
Pr GRUEL Yves	Heparin-induced thrombocytopenia	ORPHA:3325
Dr GUENEZAN Jeremy	Kawasaki disease	ORPHA:2331
Dr GUEZ Sophie	Severe generalized junctional epidermolysis bullosa	ORPHA:79404
	Autosomal dominant generalized dystrophic epidermolysis bullosa	ORPHA:231568
	Junctional epidermolysis bullosa with pyloric atresia	ORPHA:79403
Pr HABIBI Anoosha	Sickle cell anemia	ORPHA:232
Pr HACHULA Eric	Antiphospholipid syndrome	ORPHA:80
Dr HANDRUP Mette	Williams syndrome	ORPHA:904
Dr HARDY Gaëlle	OBSOLETE: ANGPT1-related hereditary angioedema with normal C1Inh	ORPHA:537891
Dr HARDY Gaëlle	Hereditary angioedema with normal C1Inh not related to F12 or PLG variant	ORPHA:599418

Expert	Disease name	ORPHAcode
Pr HAS Cristina	Epidermolysis bullosa simplex	ORPHA:304
	Dystrophic epidermolysis bullosa pruriginosa	ORPHA:89843
	Intermediate generalized junctional epidermolysis bullosa	ORPHA:79402
	Localized dystrophic epidermolysis bullosa	ORPHA:595356
	Epidermolysis bullosa simplex with muscular dystrophy	ORPHA:257
	Kindler epidermolysis bullosa	ORPHA:2908
	Junctional epidermolysis bullosa	ORPHA:305
	Epidermolysis bullosa simplex due to BP230 deficiency	ORPHA:412181
	Intermediate epidermolysis bullosa simplex with cardiomyopathy	ORPHA:508529
	Epidermolysis bullosa simplex due to exophilin 5 deficiency	ORPHA:412189
	Autosomal recessive generalized epidermolysis bullosa simplex	ORPHA:89838
	Epidermolysis bullosa simplex with pyloric atresia	ORPHA:158684
	Epidermolysis bullosa simplex with circinate migratory erythema	ORPHA:158681
	Epidermolysis bullosa simplex with mottled pigmentation	ORPHA:79397
	PLEC-related intermediate epidermolysis bullosa simplex without extracutaneous involvement	ORPHA:79401
	Late-onset junctional epidermolysis bullosa	ORPHA:79406
	Junctional epidermolysis bullosa inversa	ORPHA:79405
	Localized junctional epidermolysis bullosa	ORPHA:251393
	Localized dystrophic epidermolysis bullosa, pretibial form	ORPHA:79410
	Localized dystrophic epidermolysis bullosa, acral form	ORPHA:158673
	Localized dystrophic epidermolysis bullosa, nails only	ORPHA:158676
Dr HAYASHI Masahiro	Oculocutaneous albinism type 8	ORPHA:597733
Dr HEGER Bob	Brugada syndrome	ORPHA:130
Dr HEIDET Laurence	Renal tubular dysgenesis	ORPHA:3033
	Renal Tubular Dysgenesis due to Twin to twin transfusion	ORPHA:97367
Pr HENNEKAM Raoul	Wiedemann-Rautenstrauch syndrome	ORPHA:3455
Dr HERON Delphine	Fragile X syndrome	ORPHA:908
Dr HETZELT Katalin	CNTNAP2-related developmental and epileptic encephalopathy	ORPHA:163681
	NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	ORPHA:600663
Pr HÖGLINGER Günter	Progressive supranuclear palsy	ORPHA:683
	Classic progressive supranuclear palsy syndrome	ORPHA:240071
	Progressive supranuclear palsy-pure akinesia with gait freezing syndrome	ORPHA:240094
	Atypical progressive supranuclear palsy syndrome	ORPHA:99750
	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	ORPHA:240112
	Progressive supranuclear palsy-corticobasal syndrome	ORPHA:240103
	Progressive supranuclear palsy-parkinsonism syndrome	ORPHA:240085
Pr HOUYEL Lucile	Ebstein malformation of the tricuspid valve	ORPHA:1880
	Tricuspid atresia	ORPHA:1209
	Congenital pulmonary valvar stenosis	ORPHA:3189

Expert	Disease name	ORPHAcode
Dr ILLOUZ Frédéric	Syndrome of reduced sensitivity to thyroid hormone	ORPHA:596426
	Euthyroid dysprealbuminemic hyperthyroxinemia	ORPHA:597939
Dr JAIS Xavier	Chronic thromboembolic pulmonary hypertension	ORPHA:70591
Dr JANECKE Andreas	Syndromic congenital sodium diarrhea	ORPHA:563708
Dr JANNONE-PEDRO Nicolas	Myoclonic-astatic epilepsy	ORPHA:1942
Dr JAROUSSIE Marianne	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
Dr JEVNIKAR Mitja	Chronic thromboembolic pulmonary hypertension	ORPHA:70591
Dr JONES Wendy	Wiedemann-Steiner syndrome	ORPHA:319182
Dr JORIOT Sylvie	Sturge-Weber syndrome	ORPHA:3205
Pr KÄLVIÄINEN Reetta	Progressive myoclonic epilepsy type 1	ORPHA:308
Dr KAMINSKA Anna	Myoclonic-astatic epilepsy	ORPHA:1942
Pr KATOULIS Alexander	Syringocystadenoma papilliferum	ORPHA:840
Pr KERBAUL François	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
	Kawasaki disease	ORPHA:2331
	Sturge-Weber syndrome	ORPHA:3205
	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
	Dravet syndrome	ORPHA:33069
	Fragile X syndrome	ORPHA:908
	Brugada syndrome	ORPHA:130
	Hereditary hemorrhagic telangiectasia	ORPHA:774
	Non-histaminic angioedema	ORPHA:658
	Hypophosphatasia	ORPHA:436
	Alpha-1-antitrypsin deficiency	ORPHA:60
	Thrombotic microangiopathy	ORPHA:93573
	Sickle cell anemia	ORPHA:232
Dr KERSTJENS-FREDERIKSE Wilhelmina	Polyvalvular heart disease syndrome	ORPHA:228410
Pr KHELLAF Mehdi	Sickle cell anemia	ORPHA:232
Pr KLEEFSTRA Tjitske	Kleefstra syndrome	ORPHA:261494
Pr KONE PAUT Isabelle	Multisystem inflammatory syndrome in children and adults	ORPHA:598363
	Kawasaki disease	ORPHA:2331
Pr KONRAD Martin	Infantile Bartter syndrome with sensorineural deafness	ORPHA:89938
	Classic Bartter syndrome	ORPHA:93605
	Bartter syndrome type 5	ORPHA:570371
Pr KOTULSKA-JOZWIAK Katarzyna	Sturge-Weber syndrome	ORPHA:3205
	Tuberous sclerosis complex	ORPHA:805
Dr KUISMIN Outi	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	ORPHA:480907
Dr KUKAVICA Deni	Catecholaminergic polymorphic ventricular tachycardia	ORPHA:3286

Expert	Disease name	ORPHAcode
Dr KUSEYRIHÜBSCHMANN Oya	Dopamine beta-hydroxylase deficiency	ORPHA:230
Pr LABRUNE Philippe	Crigler-Najjar syndrome	ORPHA:205
	Crigler-Najjar syndrome type 1	ORPHA:79234
	Crigler-Najjar syndrome type 2	ORPHA:79235
Pr LACHAUX Alain	Wilson disease	ORPHA:905
	MEDNIK syndrome	ORPHA:171851
Pr LAGAE Lieven	Epilepsy with myoclonic absences	ORPHA:86911
Pr LAIMER Martin	Dystrophic epidermolysis bullosa	ORPHA:303
	Autosomal recessive generalized dystrophic epidermolysis bullosa, severe form	ORPHA:79408
	Recessive dystrophic epidermolysis bullosa inversa	ORPHA:79409
Dr LE BORGNE Pierrick	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
Pr LECONTE Philippe	Fragile X syndrome	ORPHA:908
Dr LEEGAARD Anne	Williams syndrome	ORPHA:904
Pr LEENHARDT Antoine	Brugada syndrome	ORPHA:130
Pr LEES Andrew	Atypical progressive supranuclear palsy syndrome	ORPHA:99750
	Progressive supranuclear palsy-corticobasal syndrome	ORPHA:240103
	Classic progressive supranuclear palsy syndrome	ORPHA:240071
	Progressive supranuclear palsy-parkinsonism syndrome	ORPHA:240085
	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	ORPHA:240112
Pr LEGER Juliane	Turner syndrome	ORPHA:881
	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	ORPHA:315311
	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	ORPHA:315306
	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency	ORPHA:90791
Dr LEROY Christophe	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
	Kawasaki disease	ORPHA:2331
	Sturge-Weber syndrome	ORPHA:3205
	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
	Dravet syndrome	ORPHA:33069
	Fragile X syndrome	ORPHA:908
	Brugada syndrome	ORPHA:130
	Hereditary hemorrhagic telangiectasia	ORPHA:774
	Non-histaminic angioedema	ORPHA:658
	Hypophosphatasia	ORPHA:436
	Alpha-1-antitrypsin deficiency	ORPHA:60
	Thrombotic microangiopathy	ORPHA:93573
	Sickle cell anemia	ORPHA:232
Pr LEVY Philippe	IgG4-related systemic disease	ORPHA:596448

Expert	Disease name	ORPHAcode
Pr LINGLART Agnès	Hypophosphatasia	ORPHA:436
Pr LIPSKA-ZIETKIEWICZ Beata	Schimke immuno-osseous dysplasia	ORPHA:1830
	Genetic steroid-resistant nephrotic syndrome	ORPHA:656
Dr LUZI Paola	Krabbe disease	ORPHA:487
Dr MA Elise	Leber hereditary optic neuropathy	ORPHA:104
Dr MACCHIAIOLO Marina	8p inverted duplication/deletion syndrome	ORPHA:96092
Dr MAILLARD Helene	Antiphospholipid syndrome	ORPHA:80
	IgG4-related systemic disease	ORPHA:596448
	Mevalonate kinase deficiency	ORPHA:309025
Dr MALINA Michal	Atypical hemolytic uremic syndrome	ORPHA:2134
Pr MANIÈRE Marie-Cécile	Hypophosphatasia	ORPHA:436
Dr MARINI Francesca	Multiple endocrine neoplasia type 2	ORPHA:653
Dr MARTINACHE Isabelle	Hypophosphatasia	ORPHA:436
Dr MARTINEZ Mikaël	Dravet syndrome	ORPHA:33069
Dr MATRICARDI Sara	SYNGAP1-related developmental and epileptic encephalopathy	ORPHA:544254
Dr MATULEVICIENE Ausra	Oculodentodigital dysplasia	ORPHA:2710
Dr MCTAGUE Amy	Malignant migrating focal seizures of infancy	ORPHA:293181
Dr MECKERT Francine	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
	Kawasaki disease	ORPHA:2331
	Sturge-Weber syndrome	ORPHA:3205
	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
	Dravet syndrome	ORPHA:33069
	Fragile X syndrome	ORPHA:908
	Brugada syndrome	ORPHA:130
	Hereditary hemorrhagic telangiectasia	ORPHA:774
	Non-histaminic angioedema	ORPHA:658
	Hypophosphatasia	ORPHA:436
	Alpha-1-antitrypsin deficiency	ORPHA:60
	Thrombotic microangiopathy	ORPHA:93573
	Sickle cell anemia	ORPHA:232
Dr MERCIER Marie-France	Medium chain acyl-CoA dehydrogenase deficiency	ORPHA:42
	Non-histaminic angioedema	ORPHA:658
	Alpha-1-antitrypsin deficiency	ORPHA:60
	Hypophosphatasia	ORPHA:436
Pr MERCURI Eugenio	Proximal spinal muscular atrophy type 3	ORPHA:83419
	Proximal spinal muscular atrophy type 2	ORPHA:83418
	Proximal spinal muscular atrophy type 1	ORPHA:83330
	Proximal spinal muscular atrophy type 4	ORPHA:83420
Dr MIGNOT Cyril	Lamb-Shaffer syndrome	ORPHA:530983

Expert	Disease name	ORPHAcode
Dr MOORTGAT Stéphanie	MEHMO syndrome	ORPHA:85282
Pr MORNEX Jean-François	Alpha-1-antitrypsin deficiency	ORPHA:60
Dr MOTTA Francesca	Reactive arthritis	ORPHA:29207
Dr MULLER Jean	Bardet-Biedl syndrome	ORPHA:110
Dr MUSSA Alessandro	Luscan-Lumish syndrome	ORPHA:597738
Pr NABBOUT Rima	Rett syndrome	ORPHA:778
	Progressive myoclonic epilepsy type 1	ORPHA:308
	Sturge-Weber syndrome	ORPHA:3205
	Lennox-Gastaut syndrome	ORPHA:2382
	Ring chromosome 20 syndrome	ORPHA:1444
	SYNGAP1-related developmental and epileptic encephalopathy	ORPHA:544254
	Epilepsy with myoclonic absences	ORPHA:86911
	Dravet syndrome	ORPHA:33069
	Febrile infection-related epilepsy syndrome	ORPHA:163703
	Tuberous sclerosis complex	ORPHA:805
	Malignant migrating focal seizures of infancy	ORPHA:293181
	Dravet syndrome	ORPHA:33069
Dr NERI Marcella	Mitochondrial myopathy-cerebellar ataxia-pigmentary retinopathy syndrome	ORPHA:502423
Dr NEVES Mariana	Branchio-oculo-facial syndrome	ORPHA:1297
	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	ORPHA:457193
Pr NGUYENTHETICH Sylvie	Epilepsy with myoclonic absences	ORPHA:86911
Dr NIGWEKAR Sagar	Calciphylaxis	ORPHA:280062
	Visceral calciphylaxis	ORPHA:280068
	Calciphylaxis cutis	ORPHA:280065
Dr NISHIMURA Gen	Hajdu-Cheney syndrome	ORPHA:955
Dr OBERLIN Mathieu	Brugada syndrome	ORPHA:130
Pr OPLADEN Thomas	Dopamine beta-hydroxylase deficiency	ORPHA:230
Dr OUSSOREN Esmeralda	Mucolipidosis type II	ORPHA:576
	Mucolipidosis type IV	ORPHA:578
	Mucolipidosis type III	ORPHA:577
Pr PAKARINEN Mikko	Hirschsprung disease	ORPHA:388
Pr PALLADINI Giovanni	Wild type ATTR amyloidosis	ORPHA:330001
	AA amyloidosis	ORPHA:85445
	AL amyloidosis	ORPHA:85443
	AH amyloidosis	ORPHA:442582
	Light chain deposition disease	ORPHA:93558
	Light and heavy chain deposition disease	ORPHA:93557
	Heavy chain deposition disease	ORPHA:93556
	Non-amyloid monoclonal immunoglobulin deposition disease	ORPHA:86861

Expert	Disease name	ORPHAcode
Dr PASSEMARD Sandrine	DONSON-related microcephaly-short stature-limb abnormalities spectrum	ORPHA:572761
Dr PASTURAL Myriam	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
Dr PEREIRA Elaine M.	Thanatophoric dysplasia type 2	ORPHA:93274
	Thanatophoric dysplasia type 1	ORPHA:1860
Pr PESCHANSKI Nicolas	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
Dr PICCIOLINI Odoardo	Moebius syndrome	ORPHA:570
Dr PLESSIER Aurélie	Budd-Chiari syndrome	ORPHA:131
	Hepatic veno-occlusive disease	ORPHA:890
	Incomplete septal cirrhosis	ORPHA:596941
	Hepatoportal sclerosis	ORPHA:64743
	Nodular regenerative hyperplasia of the liver	ORPHA:48372
Dr PONDARRE Corinne	Alpha-thalassemia	ORPHA:846
	Hemoglobin H disease	ORPHA:93616
	Hb Bart's hydrops fetalis	ORPHA:163596
Dr POUJOIS Aurélia	Wilson disease	ORPHA:905
Dr PRADERE Benjamin	Upper tract urothelial carcinoma	ORPHA:598216
Pr PRIORI Silvia	Catecholaminergic polymorphic ventricular tachycardia	ORPHA:3286
Dr PUECHAL Xavier	Hypocomplementemic urticarial vasculitis	ORPHA:36412
	Anti-neutrophil cytoplasmic antibody-associated vasculitis	ORPHA:156152
Dr PUJALTE Mathilde	Proximal Xq28 duplication syndrome	ORPHA:1762
Pr QUERE Isabelle	ANGPT2-related primary lymphoedema	#N/A
Pr QVIST Niels	Atresia of small intestine	ORPHA:1201
	Duodenal atresia	ORPHA:1203
Pr RALLIS Efstathios	Syringocystadenoma papilliferum	ORPHA:840
Dr RANCAN Alessandra	Atresia of small intestine	ORPHA:1201
Pr REBOURS Vinciane	IgG4-related systemic disease	ORPHA:596448
Dr REFETOFF Samuel	Euthyroid dysprealbuminemic hyperthyroxinemia	ORPHA:597939
Pr REIS André	CNTNAP2-related developmental and epileptic encephalopathy	ORPHA:163681
Dr RENOU Pauline	Sickle cell anemia	ORPHA:232
Dr ROBERT Matthieu	Sturge-Weber syndrome	ORPHA:3205
Dr RODRIGUES Marcia	X-linked intellectual disability, Cabezas type	ORPHA:85293
	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	ORPHA:496641
Pr ROORYCK-THAMBO Caroline	3MC syndrome	ORPHA:293843
Dr ROSATO Simonetta	Myhre syndrome	ORPHA:2588
Dr ROSENBERG Avi	Collagen type III glomerulopathy	ORPHA:84087
Dr ROTHENBUHLER Anya	Hypophosphatasia	ORPHA:436
Dr ROUSSEAU Geoffroy	Hereditary hemorrhagic telangiectasia	ORPHA:774
Dr RUIZ Mathias	Alpha-1-antitrypsin deficiency	ORPHA:60
Pr SADUN Alfredo	Leber hereditary optic neuropathy	ORPHA:104

Expert	Disease name	ORPHAcode
Pr SALAVASTRU Carmen	Dystrophic epidermolysis bullosa pruriginosa	ORPHA:89843
	Autosomal recessive generalized dystrophic epidermolysis bullosa, intermediate form	ORPHA:89842
	Self-improving dystrophic epidermolysis bullosa	ORPHA:79411
Dr SANDAHL Thomas Damgaard	Wilson disease	ORPHA:905
Pr SANLAVILLE Damien	Proximal Xq28 duplication syndrome	ORPHA:1762
Pr SCHLUTHBOLARD Caroline	Proximal Xq28 duplication syndrome	ORPHA:1762
Pr SCHWARTZ Peter	Familial long QT syndrome	ORPHA:768
	Andersen-Tawil syndrome	ORPHA:37553
Dr SEBODE Marcial	Autoimmune hepatitis	ORPHA:2137
	Autoimmune hepatitis type 1	ORPHA:563576
	Seronegative autoimmune hepatitis	ORPHA:563589
Pr SELMI Carlo	Reactive arthritis	ORPHA:29207
Dr SHAPIRO Amy	Hypoplasminogenemia	ORPHA:722
Pr SHOVLIN Claire	Pulmonary arteriovenous malformation	ORPHA:2038
Dr SILLERAN CHASSANY Jacqueline	Kawasaki disease	ORPHA:2331
	Brugada syndrome	ORPHA:130
Pr SLAVOTINEK Anne	Fryns syndrome	ORPHA:2059
	Multiple congenital anomalies - PIGN	ORPHA:280633
	Multiple congenital anomalies-hypotonia-seizures syndrome	ORPHA :280633
Dr SLOOTS Pim	Hirschsprung disease	ORPHA:388
Dr SNEAD Martin	Stickler syndrome	ORPHA:828
Dr SOARES Marta	X-linked intellectual disability, Cabezas type	ORPHA:85293
	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	ORPHA:496641
Pr SOCHA Piotr	Wilson disease	ORPHA:905
Dr SOIKKONEN Leila	X-linked intellectual disability-global development delay-facial dysmorphism-sacral caudal remnant syndrome	ORPHA:480907
Pr SPECCHIO Nicola	Febrile infection-related epilepsy syndrome	ORPHA:163703
Dr SPINELLI Alessandro	Mesomelia-synostoses syndrome	ORPHA:2496
Pr STÖDBERG Tommy	Febrile infection-related epilepsy syndrome	ORPHA:163703
Dr STOLK Jan	Alpha-1-antitrypsin deficiency	ORPHA:60
Pr STRNAD Pavel	Alpha-1-antitrypsin deficiency	ORPHA:60
Pr SURGES Rainer	Lennox-Gastaut syndrome	ORPHA:2382
Pr SUSEN Sophie	Von Willebrand disease type 3	ORPHA:166096
	Von Willebrand disease type 2	ORPHA:166081
Dr SUZUKI Tamio	Oculocutaneous albinism type 8	ORPHA:597733
Dr TADINI Gianluca	Autosomal dominant generalized dystrophic epidermolysis bullosa	ORPHA:231568
	Junctional epidermolysis bullosa with pyloric atresia	ORPHA:79403
	Severe generalized junctional epidermolysis bullosa	ORPHA:79404
Pr TERRIER Benjamin	Hypocomplementemic urticarial vasculitis	ORPHA:36412
	Multisystem inflammatory syndrome in children and adults	ORPHA:598363

Expert	Disease name	ORPHAcode
Dr TRIMARCHI Gabriele	Blepharophimosis-intellectual disability syndrome, SBBYS type	ORPHA:3047
Pr TÜMER Zeynep	DLG4-Intellectual developmental disorder	#N/A
	ELOVL4-related ichthyosis without ID and spastic quadriplegia	#N/A
Dr VALENZUELA Irene	Bainbridge-Ropers syndrome	ORPHA:352577
Dr VAN DIJK Tessa	Pontocerebellar hypoplasia type 13	ORPHA:613267
	Pontocerebellar hypoplasia type 12	ORPHA:611256
	Pontocerebellar hypoplasia type 11	ORPHA:611247
	Pontocerebellar hypoplasia type 14	ORPHA:613274
Dr VANBON Bregje	DYRK1A-related intellectual disability syndrome	ORPHA:464306
Dr VANDIJK Tessa	Pontocerebellar hypoplasia type 2	ORPHA:2524
	Pontocerebellar hypoplasia type 3	ORPHA:97249
	Non-syndromic pontocerebellar hypoplasia	ORPHA:98523
	Pontocerebellar hypoplasia type 6	ORPHA:166073
	Pontocerebellar hypoplasia type 4	ORPHA:166063
	Pontocerebellar hypoplasia type 1	ORPHA:2254
Dr VANLERBERGHE Clémence	Okihiro syndrome	ORPHA:93293
Pr VANRAVENSWAAIJ-ARTS Conny	Mosaic trisomy 8	ORPHA:96061
Dr VAUX Julien	Hypophosphatasia	ORPHA:436
Dr VECCHIO Davide	8p inverted duplication/deletion syndrome	ORPHA:96092
Dr VERA Gabriella	Cornelia de Lange syndrome	ORPHA:199
Pr VERKADE Henkjan	Wilson disease	ORPHA:905
Pr VIGOUROUX Corinne	Insulin-resistance syndrome type B	ORPHA:2298
Pr VILAIN Catheline	Zimmermann-Laband syndrome	ORPHA:3473
	Temple-Baraitser syndrome	ORPHA:420561
Dr VILLANUEVA Vicente	Myoclonic-astatic epilepsy	ORPHA:1942
Pr VISSING John	FKRP-related limb-girdle muscular dystrophy R9	ORPHA:34515
Pr VISSING John	Oculopharyngeal muscular dystrophy	ORPHA:270
Dr VIVARELLI Marina	Immunoglobulin-mediated membranoproliferative glomerulonephritis	ORPHA:329903
	C3 glomerulopathy	ORPHA:329918
	Primary membranoproliferative glomerulonephritis	ORPHA:54370
	Dense deposit disease	ORPHA:93571
Dr WANG Aijing	Stickler syndrome	ORPHA:828
Dr WASSENBERG Tessa	Dopamine beta-hydroxylase deficiency	ORPHA:230
Ms WATSON Allison	Ring chromosome 20 syndrome	ORPHA:1444
Pr WEBER Stefanie	HNF1B-related autosomal dominant tubulointerstitial kidney disease	ORPHA:93111
Pr WEINSHENKER Brian	Neuromyelitis optica spectrum disorder	ORPHA:71211
Pr WEISS Karl Heinz	Wilson disease	ORPHA:905
Pr WENGER David	Krabbe disease	ORPHA:487
Pr WESTER Tomas	Hirschsprung disease	ORPHA:388

Expert	Disease name	ORPHAcode
Pr WILLIAMS David	Atypical progressive supranuclear palsy syndrome	ORPHA:99750
Pr YOUNG Jacques	Isolated congenital hypogonadotropic hypogonadism	ORPHA:238666
Dr YU Run	Insulinoma	ORPHA:97279
Pr ZACCHIA Miriam	Bardet-Biedl syndrome	ORPHA:110
Dr ZAKHAROVA Ekaterina	Niemann-Pick disease type C	ORPHA:646
	Niemann-Pick disease type A	ORPHA:77292
	Niemann-Pick disease type B	ORPHA:77293
Dr ZANKER Caroline	Non-histaminic angioedema	ORPHA:658
Dr ZENNARO Maria-Christina	Familial hyperaldosteronism type I	ORPHA:403
	Pseudohypoaldosteronism type 1	ORPHA:756
	Familial hyperaldosteronism type II	ORPHA:404
	Familial hyperaldosteronism type III	ORPHA:251274
	Generalized pseudohypoaldosteronism type 1	ORPHA:171876
	Renal pseudohypoaldosteronism type 1	ORPHA:171871
Pr ZWEIER Christiane	CNTNAP2-related developmental and epileptic encephalopathy	ORPHA:163681
	NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	ORPHA:600663
	CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	ORPHA:599082
	NRXN1-related severe neurodevelopmental disorder-motor stereotypies-chronic constipation-sleep-wake cycle disturbance	ORPHA:600663

List of expert networks having reviewed the Orphanet nomenclature in 2021

Name of expert network	Disease name	ORPHAcode
EpiCARE	FOXG1 syndrome due to intragenic alteration	ORPHA:598164
ERN RARE-LIVER	Portosinusoidal vascular disease	ORPHA:596937
	Incomplete septal cirrhosis	ORPHA:596941
	Early-onset familial noncirrhotic portal hypertension	ORPHA:494348
eUROGEN	Anorectal malformation	ORPHA:96346
	Non-syndromic anorectal malformation with H-type fistula	ORPHA:601033
	Non-syndromic anorectal malformation with rectovaginal fistula	ORPHA:601028
	Non-syndromic anorectal malformation with rectal stenosis	ORPHA:601023
	Non-syndromic anorectal malformation with rectal atresia	ORPHA:601018
	Non-syndromic anorectal malformation with pouch colon	ORPHA:601013
	Non-syndromic anorectal malformation with anal stenosis	ORPHA:601008
	Non-syndromic anorectal malformation without fistula	ORPHA:601002
	Non-syndromic cloacal malformation	ORPHA:600998
	Non-syndromic anorectal malformation with vestibular fistula	ORPHA:600993
	Non-syndromic anorectal malformation with rectovesical fistula	ORPHA:600984
	Non-syndromic anorectal malformation with rectourethral fistula, prostatic type	ORPHA:600975
	Non-syndromic anorectal malformation with rectourethral fistula, bulbar type	ORPHA:600966
	Non-syndromic anorectal malformation with rectourethral fistula	ORPHA:600961
	Non-syndromic anorectal malformation with perineal fistula	ORPHA:600952
	OBSOLETE: Low isolated anorectal malformation	ORPHA:171215
	OBSOLETE: Intermediate isolated anorectal malformation	ORPHA:171208
	OBSOLETE: High isolated anorectal malformation	ORPHA:171201
ITHACA	X-linked intellectual disability, Turner type	ORPHA:85328
	Juberg-Marsidi syndrome	ORPHA:93972
	X-linked intellectual disability, Brooks type	ORPHA:3056

List of abbreviations

ERN : European Reference Network
Endo-ERN: European Reference Network on endocrine conditions
ERKNet: European Reference Network on kidney diseases
ERN BOND: European Reference Network on bone disorders
ERN CRANIO: European Reference Network on craniofacial anomalies and ENT disorders
ERN EpiCARE: European Reference Network on epilepsies
ERN EURACAN: European Reference Network on adult cancers (solid tumours)
ERN EuroBloodNet: European Reference Network on haematological diseases
ERN eUROGEN: European Reference Network on urogenital diseases and conditions
ERN EURO-NMD: European Reference Network on neuromuscular diseases
ERN EYE: European Reference Network on eye diseases
ERN GENTURIS: European Reference Network on genetic tumour risk syndromes
ERN GUARD-HEART: European Reference Network on diseases of the heart
ERNICA: European Reference Network on inherited and congenital anomalies
ERN ITHACA: European Reference Network on congenital malformations and rare intellectual disability
ERN LUNG: European Reference Network on respiratory diseases
ERN PaedCan: European Reference Network on paediatric cancer (haemato-oncology)
ERN RARE-LIVER: European Reference Network on hepatological diseases
ERN ReCONNET: European Reference Network on connective tissue and musculoskeletal diseases
ERN RITA: European Reference Network on immunodeficiency, autoinflammatory and autoimmune diseases
ERN-RND: European Reference Network on neurological diseases
ERN Skin: European Reference Network on skin disorders
ERN TRANSPLANT-CHILD: European Reference Network on transplantation in children
MetabERN: European Reference Network on hereditary metabolic disorders
VASCERN: European Reference Network on multisystemic vascular diseases

For any questions or comments, please contact us: contact.orphanet@inserm.fr

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