Orphanet Report Series

Rare Diseases collection

2020

Expert reviewers for Orphanet in 2020

Inserm
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From science to health

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MINISTÈRE DES AFFAIRES SOCIALES ET DE LA SANTÉ

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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.

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, and their affiliation to a European Reference Network (ERN) if they have contributed on behalf of an ERN.

Expert networks having contributed to the update of the aforementioned data are described in a second list.

List of expert reviewers for Orphanet in 2020

Expert	ERN Affiliation	Disease name	ORPHAcode
Pr ABADIE Véronique		Isolated Pierre Robin syndrome	ORPHA:718
Dr ACCOGLI Andrea	ITHACA	THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	ORPHA:363444
Pr AMAR Laurence		Pheochromocytoma-paraganglioma	ORPHA:573163
		Non-functioning paraganglioma	ORPHA:94080
		Sporadic pheochromocytoma/secreting paraganglioma	ORPHA:276621
		OBSOLETE: Catecholamine-producing tumor	ORPHA:717
		Pheochromocytoma-paraganglioma	ORPHA:573163
Pr AMOS Christopher		Peutz-Jeghers syndrome	ORPHA:2869
Dr ANDRIEU Jean-Bernard		Juvenile neuronal ceroid lipofuscinosis	ORPHA:79264
Dr ARNAUD Eric J.		Pfeiffer syndrome type 1	ORPHA:93258
Dr ARNOUX Jean-Baptiste		Maple syrup urine disease	ORPHA:511
Pr AROCK Michel		Aggressive systemic mastocytosis	ORPHA:98850
		Mast cell leukemia	ORPHA:98851
Dr ASCH Sarah		Linear nevus sebaceus syndrome	ORPHA:2612
Dr AYUSO Carmen		X-linked retinoschisis	ORPHA:792
Dr BACROT Severine		Perinatal lethal hypophosphatasia	ORPHA:247623
		Prenatal benign hypophosphatasia	ORPHA:247638
		Odontohypophosphatasia	ORPHA:247685
		Infantile hypophosphatasia	ORPHA:247651
		Childhood-onset hypophosphatasia	ORPHA:247667
		Adult hypophosphatasia	ORPHA:247676
		Hypophosphatasia	ORPHA:436
Pr BADENS Catherine		Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	ORPHA:98791
Dr BAGOU Gilles		Behçet disease	ORPHA:117
		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Sarcoidosis	ORPHA:797
		Susac syndrome	ORPHA:838
		Common variable immunodeficiency	ORPHA:1572
		Congenital diaphragmatic hernia	ORPHA:2140
		Lennox-Gastaut syndrome	ORPHA:2382
		West syndrome	ORPHA:3451
		Gaucher disease type 3	ORPHA:77261

Expert	ERN Affiliation	Disease name	ORPHAcode
		Mastocytosis	ORPHA:98292
		Duchenne muscular dystrophy	ORPHA:98896
		Pulmonary arterial hypertension	ORPHA:182090
Pr BAHI-BUISSON Nadia		Rett syndrome	ORPHA:778
Dr BALASUBRAMANIAN Meena		Witteveen-Kolk syndrome	ORPHA:500163
Dr BANDEIRA Anabela		3-hydroxy-3-methylglutaric aciduria	ORPHA:20
Dr BARATEAU Lucie		Narcolepsy type 2	ORPHA:83465
		Narcolepsy type 1	ORPHA:2073
		Idiopathic hypersomnia	ORPHA:33208
Dr BARISIC Ingeborg		Otomandibular syndrome	ORPHA:141136
		Goldenhar syndrome	ORPHA:374
Mr BARRY Brenda		Duane retraction syndrome	ORPHA:233
Dr BATTAGLIA Agatino		Inverted duplicated chromosome 15 syndrome	ORPHA:3306
Dr BAUJAT Geneviève		Hypophosphatasia	ORPHA:436
Pr BAUMGARTNER Matthias		3-methylcrotonyl-CoA carboxylase deficiency	ORPHA:6
Dr BAYART Daniel		Juvenile neuronal ceroid lipofuscinosis	ORPHA:79264
Dr BEAUDOIN Sylvie		Omphalocele	ORPHA:660
		Gastroschisis	ORPHA:2368
Pr BECKERS Albert		Prolactinoma	ORPHA:2965
		Pituitary gigantism	ORPHA:99725
Pr BEDANE Christophe		Mucous membrane pemphigoid	ORPHA:46486
		Pemphigus vulgaris	ORPHA:704
Dr BEDESCHI Maria Francesca	ITHACA	Fraser syndrome	ORPHA:2052
Pr BEKRI Soumeya		X-linked sideroblastic anemia and spinocerebellar ataxia	ORPHA:2802
Dr BELMATOUG Nadia		Gaucher disease type 3	ORPHA:77261
Pr BENACHI Alexandra		Congenital diaphragmatic hernia	ORPHA:2140
Dr BENLIAN Pascale		Tangier disease	ORPHA:31150
Pr BERARDELLI Alfredo		Blepharospasm-oromandibular dystonia syndrome	ORPHA:93964
Pr BERGER Marc G.		Gaucher disease type 3	ORPHA:77261
Pr BERTINI Enrico		X-linked centronuclear myopathy	ORPHA:596
		Joubert syndrome	ORPHA:475
		Centronuclear myopathy	ORPHA:595
		Autosomal recessive centronuclear myopathy	ORPHA:169186
		Autosomal dominant centronuclear myopathy	ORPHA:169189
Pr BETZ Regina		Woolly hair	ORPHA:170
Dr BIESECKER Leslie		Greig cephalopolysyndactyly syndrome	ORPHA:380
Dr BLANCHET Catherine		Usher syndrome type 3	ORPHA:231183

Expert	ERN Affiliation	Disease name	ORPHAcode
		Usher syndrome type 2	ORPHA:231178
		Usher syndrome type 1	ORPHA:231169
		Usher syndrome	ORPHA:886
Pr BLAU Nenad		Maternal phenylketonuria	ORPHA:2209
		Phenylketonuria	ORPHA:716
Dr BLESSING Matthew		Saethre-Chotzen syndrome	ORPHA:794
Pr BLOCH-ZUPAN Agnès		Hypophosphatasia	ORPHA:436
Pr BLUME-PEYTAVI Ulrike		Woolly hair	ORPHA:170
Pr BOCKENHAUER Detlef	ERKNet	Gitelman syndrome	ORPHA:358
		Primary Fanconi renotubular syndrome	ORPHA:3337
		Liddle syndrome	ORPHA:526
		Nephrogenic diabetes insipidus	ORPHA:223
		Tubulointerstitial nephritis and uveitis syndrome	ORPHA:91500
Pr BODEMER Christine		OBSOLETE: Idiopathic recurrent and disabling cutaneous herpes	ORPHA:35061
Dr BOELMA Wilma		Familial exudative vitreoretinopathy	ORPHA:891
Pr BOON Laurence		Glomuvenous malformation	ORPHA:83454
Dr BOONSTRA Nienke		Familial exudative vitreoretinopathy	ORPHA:891
Dr BOSCH A.M. [Annet]		Classic phenylketonuria	ORPHA:79254
		Mild hyperphenylalaninemia	ORPHA:79651
		Riboflavin transporter deficiency	ORPHA:97229
Pr BOUILLET Laurence		Renin-angiotensin-aldosterone system-blocker-induced angioedema	ORPHA:100057
		Non-histaminic angioedema	ORPHA:658
		Hereditary angioedema type 1	ORPHA:100050
		Hereditary angioedema type 2	ORPHA:100051
		Acquired angioedema type 1	ORPHA:100056
Dr BRAIS Bernard		Oculopharyngeal muscular dystrophy	ORPHA:270
Dr BRASSIER Anaïs		Gaucher disease type 3	ORPHA:77261
Dr BRONCHARD Régis		Duchenne muscular dystrophy	ORPHA:98896
Pr BRORSON Håkan		Adiposis dolorosa	ORPHA:36397
Pr BRUE Thierry		Septo-optic dysplasia spectrum	ORPHA:3157
Pr BSCHLEIPFER Thomas	eUROGEN	Interstitial cystitis	ORPHA:37202
Dr BUFFET Alexandre		Pheochromocytoma-paraganglioma	ORPHA:573163
Dr BU'LOCK Frances		Eisenmenger syndrome	ORPHA:97214
Dr BURNICHON Nelly		Pheochromocytoma-paraganglioma	ORPHA:573163
Dr BURRAGE Lindsay		Spondyloepiphyseal dysplasia with metatarsal shortening	ORPHA:137678
Pr BURRELL James		Corticobasal syndrome	ORPHA:454887
Dr BURTON Barbara		Lysosomal acid lipase deficiency	ORPHA:275761

Expert	ERN Affiliation	Disease name	ORPHAcode
		Wolman disease	ORPHA:75233
Dr BUSA Tiffany	ITHACA	22q11.2 deletion syndrome	ORPHA:567
		22q11.2 duplication syndrome	ORPHA:1727
Pr CAGLAYAN Ahmet		Knobloch syndrome	ORPHA:1571
Pr CALENDER Alain		Sarcoidosis	ORPHA:797
Pr CALLEWAERT Bert	ERN-Skin	Menkes disease	ORPHA:565
		Occipital horn syndrome	ORPHA:198
Pr CALVAS Patrick		Norrie disease	ORPHA:649
Dr CAMPEAU Philippe		Dysosteosclerosis	ORPHA:1782
		Autosomal dominant deafness-onychodystrophy syndrome	ORPHA:79499
Dr CAPONE Valentina	ERKNet	Senior-Loken syndrome	ORPHA:3156
Dr CAPRA Valeria	ITHACA	Freeman-Sheldon syndrome	ORPHA:2053
		THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	ORPHA:363444
Dr CARAFFI Giuseppe Stefano	ITHACA	Mowat-Wilson syndrome	ORPHA:2152
		Alazami syndrome	ORPHA:319671
		B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467
Dr CARBONE Marco		Primary biliary cholangitis	ORPHA:186
Dr CASTANHOLE-NUNES Márcia Maria		GAPO syndrome	ORPHA:2067
Dr CASTEL Marion	ERN-Skin	IgA pemphigus	ORPHA:555905
		Herpetiform pemphigus	ORPHA:208524
		Pemphigus foliaceus	ORPHA:79481
		Superficial pemphigus	ORPHA:46485
		Dermatitis herpetiformis	ORPHA:1656
		Pemphigus erythematosus	ORPHA:79480
		Bullous pemphigoid	ORPHA:703
		Paraneoplastic pemphigus	ORPHA:63455
		Anti-p200 pemphigoid	ORPHA:454710
		Pemphigoid gestationis	ORPHA:63275
		Linear IgA dermatosis	ORPHA:46488
Dr CASTELLANOS Mario		Pudendal neuralgia	ORPHA:60039
Dr CATOIRE Pierre		Duchenne muscular dystrophy	ORPHA:98896
Dr CAVIRANI Benedetta	ITHACA	Basel-Vanagaite-Smirin-Yosef syndrome	ORPHA:464738
Pr CHABROL Brigitte		Duchenne muscular dystrophy	ORPHA:98896
Dr CHAIBA Djamila		Behçet disease	ORPHA:117
Pr CHANSON Philippe		Non-functioning pituitary adenoma	ORPHA:91349
Pr CHAPURLAT Roland		Fibrous dysplasia of bone	ORPHA:249
Dr CHARLES Perrine		Spinocerebellar ataxia with axonal neuropathy	ORPHA:64753

Expert	ERN Affiliation	Disease name	ORPHAcode
		type 2	
		Ataxia-oculomotor apraxia type 1	ORPHA:1168
Pr CHARRON Philippe		Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	ORPHA:300751
		Familial isolated dilated cardiomyopathy	ORPHA:154
		Familial isolated restrictive cardiomyopathy	ORPHA:75249
Dr CHAUVIN Antony		Behçet disease	ORPHA:117
		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Sarcoidosis	ORPHA:797
		Susac syndrome	ORPHA:838
		Common variable immunodeficiency	ORPHA:1572
		Congenital diaphragmatic hernia	ORPHA:2140
		Lennox-Gastaut syndrome	ORPHA:2382
		West syndrome	ORPHA:3451
		Gaucher disease type 3	ORPHA:77261
		Mastocytosis	ORPHA:98292
		Duchenne muscular dystrophy	ORPHA:98896
		Pulmonary arterial hypertension	ORPHA:182090
Dr CHONG Kim		Monosomy 5p	ORPHA:281
Pr CIMAZ Rolando		Kawasaki disease	ORPHA:2331
Pr CLARET Pierre-Gérault		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Common variable immunodeficiency	ORPHA:1572
		Pulmonary arterial hypertension	ORPHA:182090
Pr CLAYTON-SMITH Jill	ITHACA	3M syndrome	ORPHA:2616
Dr CODY Jannine		Tetrasomy 18p	ORPHA:3307
		Distal trisomy 18q	ORPHA:1716
		Monosomy 18q	ORPHA:1600
Dr COIGNARD Hélène		Mastocytosis	ORPHA:98292
Dr COLLET Corinne	ITHACA	Summitt syndrome	ORPHA:3210
		Goodman syndrome	ORPHA:65798
		Carpenter syndrome	ORPHA:65759
		Treacher-Collins syndrome	ORPHA:861
Dr COLLINS Michael		Fibrous dysplasia of bone	ORPHA:249

Expert	ERN Affiliation	Disease name	ORPHAcode
Pr COLOMBI Marina		Classical Ehlers-Danlos syndrome	ORPHA:287
Dr CONTRO Gianluca		B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:75496
Pr COPPO Paul	ERKNet	Thrombotic thrombocytopenic purpura	ORPHA:54057
		Congenital thrombotic thrombocytopenic purpura	ORPHA:93583
		Immune-mediated thrombotic thrombocytopenic purpura	ORPHA:93585
Dr COPPOLA Antonietta		Benign adult familial myoclonic epilepsy	ORPHA:86814
Pr CORMIER-DAIRE Valerie		Weill-Marchesani syndrome	ORPHA:3449
		Marshall-Smith syndrome	ORPHA:561
		Malan overgrowth syndrome	ORPHA:420179
Dr CORNU Erika		Pheochromocytoma-paraganglioma	ORPHA:573163
		Non-functioning paraganglioma	ORPHA:94080
		Sporadic pheochromocytoma/secreting paraganglioma	ORPHA:276621
		OBSOLETE: Catecholamine-producing tumor	ORPHA:717
		Pheochromocytoma-paraganglioma	ORPHA:573163
Dr CORTON PÉREZ Marta		X-linked retinoschisis	ORPHA:792
Dr COTTIN Vincent		Bronchiolitis obliterans with obstructive pulmonary disease	ORPHA:1303
Dr CRISTOFERI Laura		Primary biliary cholangitis	ORPHA:186
Pr CRUTCH Sebastian		Posterior cortical atrophy	ORPHA:54247
Dr CUNNINGHAM Michael		Saethre-Chotzen syndrome	ORPHA:794
Pr CZAUDERNA Piotr		Hepatoblastoma	ORPHA:449
Dr DALY Adrian		Prolactinoma	ORPHA:2965
		Pituitary gigantism	ORPHA:99725
Dr D'AMICO Adele		X-linked centronuclear myopathy	ORPHA:596
		Centronuclear myopathy	ORPHA:595
		Autosomal recessive centronuclear myopathy	ORPHA:169186
		Autosomal dominant centronuclear myopathy	ORPHA:169189
Pr DAUVILLIERS Yves		Familial advanced sleep-phase syndrome	ORPHA:164736
		Narcolepsy type 2	ORPHA:83465
		Narcolepsy type 1	ORPHA:2073
		Idiopathic hypersomnia	ORPHA:33208
		Non-24-hour sleep-wake syndrome	ORPHA:73267
Dr DAVID Clémence		Susac syndrome	ORPHA:838
Pr DE LONLAY Pascale		Maple syrup urine disease	ORPHA:511
Pr DE PONTUAL Loic	ITHACA	Feingold syndrome	ORPHA:1305
Pr DE BAERE Elfride		Blepharophimosis-ptosis-epicanthus inversus syndrome	ORPHA:126
Dr DE BROUWER Arjan		Phosphoribosylpyrophosphate synthetase	ORPHA:3222

Expert	ERN Affiliation	Disease name	ORPHAcode
		superactivity	
		Mild phosphoribosylpyrophosphate synthetase superactivity	ORPHA:411536
		Severe phosphoribosylpyrophosphate synthetase superactivity	ORPHA:411543
Pr DE PONTUAL Loïc	ITHACA	Feingold syndrome type 1	ORPHA:391641
		Feingold syndrome type 2	ORPHA:391646
Dr DEBIEC Hanna		Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	ORPHA:69063
DEL POZO VALERO Marta		X-linked retinoschisis	ORPHA:792
Pr DELAPORTE Emmanuel		Sweet syndrome	ORPHA:3243
Pr DES PORTES Vincent		Fragile X syndrome	ORPHA:908
Pr DESGUERRE Isabelle		Duchenne muscular dystrophy	ORPHA:98896
Dr DESLANDRE Chantal		Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	ORPHA:85408
		Oligoarticular juvenile idiopathic arthritis	ORPHA:85410
		Systemic-onset juvenile idiopathic arthritis	ORPHA:85414
		Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	ORPHA:85435
		Psoriasis-related juvenile idiopathic arthritis	ORPHA:85436
		Enthesitis-related juvenile idiopathic arthritis	ORPHA:85438
Dr DHOOGE Patty		Stargardt disease	ORPHA:827
Pr DI FILIPPO Sylvie		Supravalvular aortic stenosis	ORPHA:3193
Dr DIGILIO Maria Cristina	ITHACA	Noonan syndrome with multiple lentigines	ORPHA:500
Dr DIVIZIA Maria Teresa		Poland syndrome	ORPHA:2911
Pr DOCO-FENZY Martine		2q37 microdeletion syndrome	ORPHA:1001
Pr DOLLFUS Hélène		Multiple syringomas	ORPHA:99156
Dr DONADIEU Jean		Langerhans cell histiocytosis	ORPHA:389
Dr DÖRR Jan-Markus		Susac syndrome	ORPHA:838
Dr DOUAY Bénédicte		Common variable immunodeficiency	ORPHA:1572
		Susac syndrome	ORPHA:838
Dr DREISLER Eva		Asherman syndrome	ORPHA:137686
Dr DUBOURG Christèle		Holoprosencephaly	ORPHA:2162
Pr EDERY Patrick	ITHACA	Microcephalic osteodysplastic primordial dwarfism types I and III	ORPHA:2636
Dr EL GHOUZZI Vincent	ITHACA	Dyggve-Melchior-Clausen disease	ORPHA:239
Pr ENG Charis		Cowden syndrome	ORPHA:201
		Bannayan-Riley-Ruvalcaba syndrome	ORPHA:109
Dr ENGLE Elizabeth		Duane retraction syndrome	ORPHA:233
Pr ENK Claes		Solar urticaria	ORPHA:97230
Pr FARR Sebastian		Ulnar hemimelia	ORPHA:93320

Expert	ERN Affiliation	Disease name	ORPHAcode
		Radial hemimelia	ORPHA:93321
		Tibial hemimelia	ORPHA:93322
		Fibular hemimelia	ORPHA:93323
Dr FASSIHI Hiva		Xeroderma pigmentosum variant	ORPHA:90342
Pr FATTORI Fabiana		X-linked centronuclear myopathy	ORPHA:596
		Centronuclear myopathy	ORPHA:595
		Autosomal recessive centronuclear myopathy	ORPHA:169186
		Autosomal dominant centronuclear myopathy	ORPHA:169189
Pr FELLMAN Vineta		GRACILE syndrome	ORPHA:53693
Dr FENOGLIO Roberta	ERKNet	Pauci-immune glomerulonephritis	ORPHA:93126
Pr FERRI Clodoveo		Mixed cryoglobulinemia type II	ORPHA:93554
		Mixed cryoglobulinemia type III	ORPHA:93555
		Cryoglobulinemic vasculitis	ORPHA:91138
Pr FIESCHI Claire		Common variable immunodeficiency	ORPHA:1572
Dr FISCHLER Björn	ERN RARE-LIVER	Isolated biliary atresia	ORPHA:30391
Pr FONTAINE Bertrand		Schwartz-Jampel syndrome	ORPHA:800
Dr FRANK Michael		Vascular Ehlers-Danlos syndrome	ORPHA:286
Dr FUJIOKA Shinsuke		Spinocerebellar ataxia type 34	ORPHA:1955
		Spinocerebellar ataxia type 30	ORPHA:211017
		Spinocerebellar ataxia type 6	ORPHA:98758
		Spinocerebellar ataxia type 37	ORPHA:363710
		Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	ORPHA:314404
		Spinocerebellar ataxia type 7	ORPHA:94147
		Spinocerebellar ataxia type 5	ORPHA:98766
		Spinocerebellar ataxia type 29	ORPHA:208513
		Spinocerebellar ataxia type 31	ORPHA:217012
		Spinocerebellar ataxia type 35	ORPHA:276193
		Spinocerebellar ataxia type 32	ORPHA:276183
		Spinocerebellar ataxia type 36	ORPHA:276198
Pr FUKAO Toshiyuki		Beta-ketothiolase deficiency	ORPHA:134
		Succinyl-CoA:3-oxoacid CoA transferase deficiency	ORPHA:832
Dr GALEOTTI Caroline		Kawasaki disease	ORPHA:2331
Dr GARAVELLI Livia	ITHACA	Spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536471
		Basel-Vanagaite-Smirin-Yosef syndrome	ORPHA:464738
		Distal arthrogryposis type 1	ORPHA:1146
		Mowat-Wilson syndrome	ORPHA:2152
		SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:157965

Expert	ERN Affiliation	Disease name	ORPHAcode
		Megalencephaly-capillary malformation-polymicrogyria syndrome	ORPHA:60040
		B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:75496
		Alazami syndrome	ORPHA:319671
		B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467
Dr GARG Abhimanyu		CANDLE syndrome	ORPHA:325004
		JMP syndrome	ORPHA:324999
		Nakajo-Nishimura syndrome	ORPHA:2615
		Proteasome-associated autoinflammatory syndrome	ORPHA:324977
		Proteasome-associated autoinflammatory syndrome	ORPHA:324977
Dr GATTORNO Marco		NLRC4-related familial cold autoinflammatory syndrome	ORPHA:576349
		CINCA syndrome	ORPHA:1451
Pr GENEVIEVE David	ITHACA	Kabuki syndrome	ORPHA:2322
Pr GENNERY Andrew		Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome	ORPHA:37042
Pr GERMAIN Dominique		Pseudoxanthoma elasticum	ORPHA:758
Dr GERUSSI Alessio		Primary biliary cholangitis	ORPHA:186
Dr GHOUMID Jamal	ITHACA	Blepharo-cheilo-odontic syndrome	ORPHA:1997
		Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	ORPHA:369891
Dr GIANI Teresa		Kawasaki disease	ORPHA:2331
Pr GIDEON Koren		Fetal valproate spectrum disorder	ORPHA:1906
Pr GIMENEZ ROQUEPLO Anne-Paule		Pheochromocytoma-paraganglioma	ORPHA:573163
		Non-functioning paraganglioma	ORPHA:94080
		Sporadic pheochromocytoma/secreting paraganglioma	ORPHA:276621
		OBSOLETE: Catecholamine-producing tumor	ORPHA:717
		Pheochromocytoma-paraganglioma	ORPHA:573163
Dr GIOVANNI Mosiello	eUROGEN	Prune belly syndrome	ORPHA:2970
Dr GIUNTA Cecilia		Arthrochalasia Ehlers-Danlos syndrome	ORPHA:1899
Dr GIUSTI Francesca		Multiple endocrine neoplasia type 2	ORPHA:653
		Multiple endocrine neoplasia type 1	ORPHA:652
Dr GLOAGUEN Aurélie		Sarcoidosis	ORPHA:797
Dr GODDET Nathalie Sybille		Rett syndrome	ORPHA:778
Pr GODEAU Bertrand		Immune thrombocytopenia	ORPHA:3002
Pr GOLFIER Francois		Gestational trophoblastic neoplasm	ORPHA:59305
		Hydatidiform mole	ORPHA:99927
Pr GOLONI-BERTOLLO Eny		GAPO syndrome	ORPHA:2067

Expert	ERN Affiliation	Disease name	ORPHAcode
Maria			
Pr GOUDEMAND Jenny		Congenital factor II deficiency	ORPHA:325
		Combined deficiency of factor V and factor VIII	ORPHA:35909
		Congenital factor X deficiency	ORPHA:328
		Von Willebrand disease	ORPHA:903
		Von Willebrand disease type 1	ORPHA:166078
		Congenital factor XI deficiency	ORPHA:329
Dr GREENBERG Benjamin		Acute disseminated encephalomyelitis	ORPHA:83597
Dr GREIG Aina		Pfeiffer syndrome type 1	ORPHA:93258
Dr GRELET Maude		SATB2-associated syndrome	ORPHA:576278
Dr GREVER Michael		Classic hairy cell leukemia	ORPHA:58017
		Hairy cell leukemia variant	ORPHA:300878
Dr GRIESE Matthias		Infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia	ORPHA:572428
		Hereditary pulmonary alveolar proteinosis	ORPHA:264675
Pr GRIFFITHS Paul		Schizencephaly	ORPHA:799
Pr GROOTHOFF J.W. [Jaap]	ERKNet	Primary hyperoxaluria	ORPHA:416
Pr GROSSFELD Paul		Jacobsen syndrome	ORPHA:2308
Pr GRUNEBAUM Eyal		Purine nucleoside phosphorylase deficiency	ORPHA:760
Pr GUERRINI Renzo		Rolandic epilepsy-paroxysmal exercise-induced dystonia-writer's cramp syndrome	ORPHA:163727
Pr HACHULLA Eric		CREST syndrome	ORPHA:90290
Pr HAGERMAN Randi		Fragile X syndrome	ORPHA:908
		Fragile X-associated tremor/ataxia syndrome	ORPHA:93256
		Fragile X syndrome	ORPHA:908
Dr HAMEL Valérie		Langerhans cell histiocytosis	ORPHA:389
Dr HAMM Austin		Pfeiffer syndrome type 1	ORPHA:93258
Dr HANSSON Emma		Adiposis dolorosa	ORPHA:36397
Dr HAYASHI Masahiro		Oculocutaneous albinism type 6	ORPHA:370097
		Oculocutaneous albinism type 5	ORPHA:370091
		Oculocutaneous albinism type 7	ORPHA:352745
		Temperature-sensitive oculocutaneous albinism type 1	ORPHA:352737
		Minimal pigment oculocutaneous albinism type 1	ORPHA:352734
		Oculocutaneous albinism type 1	ORPHA:352731
		Oculocutaneous albinism	ORPHA:55
		Oculocutaneous albinism type 1A	ORPHA:79431
		Oculocutaneous albinism type 2	ORPHA:79432
		Oculocutaneous albinism type 3	ORPHA:79433
		Oculocutaneous albinism type 1B	ORPHA:79434

Expert	ERN Affiliation	Disease name	ORPHAcode
		Oculocutaneous albinism type 4	ORPHA:79435
Dr HEATH Karen		OBSOLETE: Madelung deformity	ORPHA:35688
		OBSOLETE: Madelung deformity	ORPHA:35688
		LÚri-Weill dyschondrosteosis	ORPHA:240
		Langer mesomelic dysplasia	ORPHA:2632
		Hypochondroplasia	ORPHA:429
		Rhizomelic chondrodysplasia punctata	ORPHA:177
Dr HEIDET Laurence	ERKNet	Galloway-Mowat syndrome	ORPHA:2065
		Alport syndrome	ORPHA:63
Dr HEIDET Laurence	ERKNet	X-linked Alport syndrome-diffuse leiomyomatosis	ORPHA:1018
Dr HEIKE Carrie		Otomandibular syndrome	ORPHA:141136
		Goldenhar syndrome	ORPHA:374
		Oculo-auriculo-vertebral spectrum	ORPHA:141132
Pr HENNEKAM R.C. [Raoul]	ITHACA	Hennekam syndrome	ORPHA:2136
		Pitt-Hopkins syndrome	ORPHA:2896
Pr HERMINE Olivier		Mastocytosis	ORPHA:98292
Dr HILGER Alina		Atresia of urethra	ORPHA:105
Dr HOLLAND Steven		Chronic granulomatous disease	ORPHA:379
Dr HONJO KAWAHIRA Rachel		Monosomy 5p	ORPHA:281
Pr HOPPE Bernd	ERKNet	Primary hyperoxaluria type 1	ORPHA:93598
		Primary hyperoxaluria type 2	ORPHA:93599
		Primary hyperoxaluria	ORPHA:416
Pr HOUYEL Lucile		Complete atrioventricular septal defect	ORPHA:1329
		Partial atrioventricular septal defect	ORPHA:1330
		Intermediate atrioventricular septal defect	ORPHA:576242
		Complete atrioventricular septal defect	ORPHA:1329
		Atrioventricular septal defect	ORPHA:98722
		Partial atrioventricular septal defect	ORPHA:1330
		Complete atrioventricular septal defect	ORPHA:1329
		Interatrial communication	ORPHA:1478
Pr HOYNG Carel		Stargardt disease	ORPHA:827
Dr HOYTEMA VAN KONIJNENBURG Eva		Riboflavin transporter deficiency	ORPHA:97229
Dr INNES A.Micheil		Bowen-Conradi syndrome	ORPHA:1270
Pr INVERNIZZI Pietro	ERN RARE-LIVER	Primary biliary cholangitis	ORPHA:186
Dr JAEGER Bregje		Riboflavin transporter deficiency	ORPHA:97229
Dr JANECKE Andreas		Congenital sodium diarrhea	ORPHA:103908
Dr JAROUSSIE Marianne		Maple syrup urine disease	ORPHA:511
Dr JENY Florence		Sarcoidosis	ORPHA:797

Expert	ERN Affiliation	Disease name	ORPHAcode
		Sarcoidosis	ORPHA:797
Dr JOKELA Manu		Lower motor neuron syndrome with late-adult onset	ORPHA:276435
Pr JOLY Pascal	ERN-Skin	IgA pemphigus	ORPHA:555905
		Herpetiform pemphigus	ORPHA:208524
		Pemphigus foliaceus	ORPHA:79481
		Superficial pemphigus	ORPHA:46485
		Dermatitis herpetiformis	ORPHA:1656
		Pemphigus erythematosus	ORPHA:79480
		Bullous pemphigoid	ORPHA:703
		Paraneoplastic pemphigus	ORPHA:63455
		Linear IgA dermatosis	ORPHA:46488
		Pemphigoid gestationis	ORPHA:63275
		Anti-p200 pemphigoid	ORPHA:454710
Pr JONDEAU Guillaume		OBSOLETE: Aneurysm or dilatation of ascending aorta	ORPHA:95484
Dr JUNGK Christine		Meningioma	ORPHA:2495
Dr KAMIHARA Junne		Li-Fraumeni syndrome	ORPHA:524
Dr KANAZAWA Nobuo		CANDLE syndrome	ORPHA:325004
		JMP syndrome	ORPHA:324999
		Nakajo-Nishimura syndrome	ORPHA:2615
		Proteasome-associated autoinflammatory syndrome	ORPHA:324977
Pr KANITAKIS Jean		Disseminated superficial actinic porokeratosis	ORPHA:79152
		Porokeratosis of Mibelli	ORPHA:735
		Porokeratosis plantaris palmaris et disseminata	ORPHA:737
Pr KAVAMURA Maria Ines		Cardiofaciocutaneous syndrome	ORPHA:1340
Pr KAZUMOTO Iijima		Pierson syndrome	ORPHA:2670
Pr KEIJZER Richard		Congenital diaphragmatic hernia	ORPHA:2140
Pr KERBAUL François		Behçet disease	ORPHA:117
		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Sarcoidosis	ORPHA:797
		Susac syndrome	ORPHA:838
		Common variable immunodeficiency	ORPHA:1572
		Congenital diaphragmatic hernia	ORPHA:2140
		Lennox-Gastaut syndrome	ORPHA:2382
		West syndrome	ORPHA:3451

Expert	ERN Affiliation	Disease name	ORPHAcode
		Gaucher disease type 3	ORPHA:77261
		Mastocytosis	ORPHA:98292
		Duchenne muscular dystrophy	ORPHA:98896
		Pulmonary arterial hypertension	ORPHA:182090
Pr KLEEFSTRA Tjitske	ITHACA	Witteveen-Kolk syndrome	ORPHA:500163
Dr KOHL Stefan	ERKNet	Renal hypoplasia	ORPHA:93101
		Renal dysplasia	ORPHA:93108
		Renal agenesis	ORPHA:411709
Pr KOHLSCHÜTTER Alfried		Juvenile neuronal ceroid lipofuscinosis	ORPHA:79264
Pr KONE-PAUT Isabelle		Kawasaki disease	ORPHA:2331
Dr KÖNIG Jens	ERKNet	Senior-Loken syndrome	ORPHA:3156
Pr KONRAD Martin	ERKNet	Transient pseudohypoaldosteronism	ORPHA:93164
Dr KOPACOVA Marcela		Blue rubber bleb nevus	ORPHA:1059
Dr KUSEYRI HÜBSCHMANN Oya		Aromatic L-amino acid decarboxylase deficiency	ORPHA:35708
Pr LACOUR-GAYET François		Double outlet right ventricle	ORPHA:3426
Pr LAPUNZINA Pablo	ITHACA	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	ORPHA:300496
		Simpson-Golabi-Behmel syndrome	ORPHA:373
Dr LATOURTE Augustin		Familial calcium pyrophosphate deposition	ORPHA:1416
Dr LAVILLAUREIX Alinoë		Holoprosencephaly	ORPHA:2162
Dr LEBLANC Thierry	ITHACA	Fanconi anemia	ORPHA:84
Pr LECONTE Philippe		Maple syrup urine disease	ORPHA:511
Dr LEEHEY Maureen		Fragile X-associated tremor/ataxia syndrome	ORPHA:93256
Pr LENNON Rachel	ERKNet	Alport syndrome	ORPHA:63
		X-linked Alport syndrome-diffuse leiomyomatosis	ORPHA:1018
Dr LERONE Margherita		Poland syndrome	ORPHA:2911
Dr LEROY Christophe		Behçet disease	ORPHA:117
		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Sarcoidosis	ORPHA:797
		Susac syndrome	ORPHA:838
		Common variable immunodeficiency	ORPHA:1572
		Congenital diaphragmatic hernia	ORPHA:2140
		Lennox-Gastaut syndrome	ORPHA:2382
		West syndrome	ORPHA:3451
		Gaucher disease type 3	ORPHA:77261
		Mastocytosis	ORPHA:98292

Expert	ERN Affiliation	Disease name	ORPHAcode
		Duchenne muscular dystrophy	ORPHA:98896
		Pulmonary arterial hypertension	ORPHA:182090
Dr LEVESQUE Amélie		Pudendal neuralgia	ORPHA:60039
Dr LEVI Assi		Solar urticaria	ORPHA:97230
Pr LEVTCHENKO Elena	ERKNet	Cystinosis	ORPHA:213
		Dominant hypophosphatemia with nephrolithiasis or osteoporosis	ORPHA:244305
Pr LI Jun		Hereditary neuropathy with liability to pressure palsies	ORPHA:640
Dr LIEBAU Max	ERKNet	Fraser syndrome	ORPHA:2052
		Galloway-Mowat syndrome	ORPHA:2065
		Atresia of urethra	ORPHA:105
		Renal dysplasia	ORPHA:93108
		Renal agenesis	ORPHA:411709
		Renal hypoplasia	ORPHA:93101
Pr LINGLART Agnès		Hypophosphatasia	ORPHA:436
Dr LIOU Theodore		Cystic fibrosis	ORPHA:586
Pr LIPSKA-ZIETKIEWICZ Beata S.		Frasier syndrome	ORPHA:347
Dr Lívia GALBIATTI-DIAS Ana		GAPO syndrome	ORPHA:2067
Pr LORTHOLARY Olivier		Mastocytosis	ORPHA:98292
Dr LUCENDO Alfredo		Eosinophilic esophagitis	ORPHA:73247
Pr Luisa BRANDI Maria		Multiple endocrine neoplasia type 2	ORPHA:653
		Multiple endocrine neoplasia type 1	ORPHA:652
Dr LUQUETTI Daniela		Otomandibular syndrome	ORPHA:141136
		Goldenhar syndrome	ORPHA:374
		Oculo-auriculo-vertebral spectrum	ORPHA:141132
Dr MAKI Robert		Dedifferentiated liposarcoma	ORPHA:99970
Dr MALAN Valérie	ITHACA	Marshall-Smith syndrome	ORPHA:561
		Malan overgrowth syndrome	ORPHA:420179
Dr MALCHOFF Carl		Differentiated thyroid carcinoma	ORPHA:146
Dr MALTRET Alice		Histiocytoid cardiomyopathy	ORPHA:137675
Pr MANIÈRE Marie-Cécile		Hypophosphatasia	ORPHA:436
Dr MARCIANO Emmanuel		Congenital glaucoma	ORPHA:98976
Dr MARINI Francesca		Multiple endocrine neoplasia type 2	ORPHA:653
		Multiple endocrine neoplasia type 1	ORPHA:652
Pr Martin DAS Anibh		Congenital sucrase-isomaltase deficiency	ORPHA:35122
Dr MARTINACHE Isabelle		Behçet disease	ORPHA:117
		Hypophosphatasia	ORPHA:436
		Rett syndrome	ORPHA:778

Expert	ERN Affiliation	Disease name	ORPHAcode
		Susac syndrome	ORPHA:838
Dr MARTINEZ Mikaël		West syndrome	ORPHA:3451
Dr MATSUMOTO Hideki		Succinyl-CoA:3-oxoacid CoA transferase deficiency	ORPHA:832
Dr MAZZANTI Andrea		Brugada syndrome	ORPHA:130
Pr MAZZANTI Laura	ITHACA	Noonan syndrome-like disorder with loose anagen hair	ORPHA:2701
Dr MECKERT Francine		Behçet disease	ORPHA:117
		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Sarcoidosis	ORPHA:797
		Susac syndrome	ORPHA:838
		Common variable immunodeficiency	ORPHA:1572
		Congenital diaphragmatic hernia	ORPHA:2140
		Lennox-Gastaut syndrome	ORPHA:2382
		West syndrome	ORPHA:3451
		Gaucher disease type 3	ORPHA:77261
		Mastocytosis	ORPHA:98292
		Duchenne muscular dystrophy	ORPHA:98896
		Pulmonary arterial hypertension	ORPHA:182090
Pr MEKAHLI Djalila	ITHACA	Autosomal dominant polycystic kidney disease	ORPHA:730
Dr MENEGHESSO Davide	ERKNet	Infantile nephronophthisis	ORPHA:93591
Pr MEOLA Giovanni		Steinert myotonic dystrophy	ORPHA:273
		Proximal myotonic myopathy	ORPHA:606
Dr MERCIER Sandra		Alobar holoprosencephaly	ORPHA:93925
Dr MERCIER Marie-France		Behçet disease	ORPHA:117
		Langerhans cell histiocytosis	ORPHA:389
		Hypophosphatasia	ORPHA:436
		Maple syrup urine disease	ORPHA:511
		Rett syndrome	ORPHA:778
		Common variable immunodeficiency	ORPHA:1572
Dr MESULAM Marek-Marsel		Frontotemporal dementia, right temporal atrophy variant	ORPHA:293848
Pr MEUNIER Isabelle		Usher syndrome type 3	ORPHA:231183
		Usher syndrome type 2	ORPHA:231178
		Usher syndrome type 1	ORPHA:231169
		Usher syndrome	ORPHA:886
Pr MILEA Dan		Autosomal dominant optic atrophy plus syndrome	ORPHA:1215

Expert	ERN Affiliation	Disease name	ORPHAcode
Pr MILEA Dan		Autosomal dominant optic atrophy, classic form	ORPHA:98673
Dr MILLÁN SALVADOR José María		X-linked retinoschisis	ORPHA:792
Pr MITANCHEZ Delphine		Omphalocele	ORPHA:660
Pr MOLINA-MOLINA María		Idiopathic pulmonary fibrosis	ORPHA:2032
Pr MONTINI Giovanni	ERKNet	Senior-Loken syndrome	ORPHA:3156
Dr MOOSAJEE Mariya		Congenital primary aphakia	ORPHA:83461
Dr MORNET Etienne		Perinatal lethal hypophosphatasia	ORPHA:247623
		Prenatal benign hypophosphatasia	ORPHA:247638
		Infantile hypophosphatasia	ORPHA:247651
		Childhood-onset hypophosphatasia	ORPHA:247667
		Adult hypophosphatasia	ORPHA:247676
		Odontohypophosphatasia	ORPHA:247685
		Hypophosphatasia	ORPHA:436
Dr MORROW Eric		Christianson syndrome	ORPHA:85278
Pr MÜLLER Thomas		Congenital sodium diarrhea	ORPHA:103908
Dr MUR Sébastien		Congenital diaphragmatic hernia	ORPHA:2140
Dr MURAWSKI Maciej		Hepatoblastoma	ORPHA:449
Dr MURER Luisa	ERKNet	Nephronophthisis	ORPHA:655
		Infantile nephronophthisis	ORPHA:93591
Dr MUSSA Alessandro	ITHACA	Sotos syndrome	ORPHA:821
Pr NABBOUT Rima		Lennox-Gastaut syndrome	ORPHA:2382
		West syndrome	ORPHA:3451
Dr NEGRISOLO Susanna	ERKNet	Nephronophthisis	ORPHA:655
		Infantile nephronophthisis	ORPHA:93591
Dr NESTI Claudia		NARP syndrome	ORPHA:644
Dr NICO Marcello		Cheilitis glandularis	ORPHA:1221
		Cheilitis glandularis	ORPHA:1221
Pr NORDENSKJÖLD Agneta		46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	ORPHA:753
		Posterior hypospadias	ORPHA:95706
Pr NOZU Kandai		Pierson syndrome	ORPHA:2670
Pr NUNES Hilario		Sarcoidosis	ORPHA:797
Dr OBERLIN Mathieu		Gaucher disease type 3	ORPHA:77261
Pr ODENT Sylvie	ITHACA	Alobar holoprosencephaly	ORPHA:93925
		Alobar holoprosencephaly	ORPHA:93925
		Lobar holoprosencephaly	ORPHA:93924
		Semilobar holoprosencephaly	ORPHA:220386
		Microform holoprosencephaly	ORPHA:280200
		Septopreoptic holoprosencephaly	ORPHA:280195

Expert	ERN Affiliation	Disease name	ORPHAcode
		Holoprosencephaly	ORPHA:2162
Pr OLIVIER-FAIVRE Laurence	ITHACA	Nasopalpebral lipoma-coloboma syndrome	ORPHA:2399
		White-Sutton syndrome	ORPHA:468678
		Tall stature-intellectual disability-renal anomalies syndrome	ORPHA:500095
Pr OPLADEN Thomas		Aromatic L-amino acid decarboxylase deficiency	ORPHA:35708
Pr ORNOY Asher		Fetal valproate spectrum disorder	ORPHA:1906
Dr ORO Saskia		Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	ORPHA:95455
Dr ORSUCCI Daniele		Familial paroxysmal ataxia	ORPHA:97
Dr PAISAN-RUIZ Coro		Young-onset Parkinson disease	ORPHA:2828
Pr PAPO Thomas		Susac syndrome	ORPHA:838
Dr PAVARINO Érika Cristina		GAPO syndrome	ORPHA:2067
Dr PELLISE Maria		Serrated polyposis syndrome	ORPHA:157798
Dr PERRI Annamaria		Noonan syndrome-like disorder with loose anagen hair	ORPHA:2701
Dr PERRIN Laurence	ITHACA	Smith-Magenis syndrome	ORPHA:819
Pr PESCHANSKI Nicolas		Lennox-Gastaut syndrome	ORPHA:2382
Pr PETERSEN Claus	ERN RARE-LIVER	Isolated biliary atresia	ORPHA:30391
Pr PETIT Florence	ITHACA	Blepharo-cheilo-odontic syndrome	ORPHA:1997
Pr PHILIP Nicole	ITHACA	SATB2-associated syndrome	ORPHA:576278
		22q11.2 deletion syndrome	ORPHA:567
		22q11.2 duplication syndrome	ORPHA:1727
		KBG syndrome	ORPHA:2332
Dr PICCOLO Gianluca	ITHACA	Freeman-Sheldon syndrome	ORPHA:2053
		THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome	ORPHA:363444
Dr PICHARD Samia		Gaucher disease type 3	ORPHA:77261
Dr PILLEBOUT Evangeline		Immunoglobulin A vasculitis	ORPHA:761
Dr POLLAZZON Marzia	ITHACA	Distal arthrogryposis type 1	ORPHA:1146
Dr PONDARRE Corinne		Hemoglobin H disease	ORPHA:93616
Dr POWELL Cynthia		Townes-Brocks syndrome	ORPHA:857
Pr PRIORI Silvia		Brugada syndrome	ORPHA:130
Dr PUECHAL Xavier		Cutaneous small vessel vasculitis	ORPHA:889
		Eosinophilic granulomatosis with polyangiitis	ORPHA:183
		Microscopic polyangiitis	ORPHA:727
		Polyarteritis nodosa	ORPHA:767
		Primary angiitis of the central nervous system	ORPHA:140989
Dr PUJALTE Mathilde	ITHACA	Distal Xq28 microduplication syndrome	ORPHA:293939
Dr PULA Shpresa		Duchenne muscular dystrophy	ORPHA:98896

Expert	ERN Affiliation	Disease name	ORPHAcode
		Duchenne and Becker muscular dystrophy	ORPHA:262
		Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers	ORPHA:206546
		Becker muscular dystrophy	ORPHA:98895
Dr PULITI Aldamaria		Poland syndrome	ORPHA:2911
Dr PUTOUX Audrey		Microcephalic osteodysplastic primordial dwarfism types I and III	ORPHA:2636
Pr QUIJANO-ROY Susana		Congenital muscular dystrophy due to LMNA mutation	ORPHA:157973
Pr QUINLIVAN Rosaline		Duchenne muscular dystrophy	ORPHA:98896
		Duchenne and Becker muscular dystrophy	ORPHA:262
		Symptomatic form of muscular dystrophy of Duchenne and Becker in female carriers	ORPHA:206546
		Becker muscular dystrophy	ORPHA:98895
Pr RADLER Christof		Ulnar hemimelia	ORPHA:93320
		Radial hemimelia	ORPHA:93321
		Tibial hemimelia	ORPHA:93322
		Fibular hemimelia	ORPHA:93323
Pr RAHMAN Shamima		WARS2-related combined oxidative phosphorylation defect	ORPHA:572798
Dr RAJAN Neil		Brooke-Spiegler syndrome	ORPHA:79493
Dr RAMOND Francis		AICA-ribosiduria	ORPHA:250977
Pr RAWASHDEH Yazan	eUROGEN	Posterior urethral valve	ORPHA:93110
		Anterior urethral valve	ORPHA:435372
Dr REPKA Michael		Retinopathy of prematurity	ORPHA:90050
Pr REYNIER Pascal		Autosomal dominant optic atrophy plus syndrome	ORPHA:1215
		Autosomal dominant optic atrophy, classic form	ORPHA:98673
Dr RHYS Evans	ERKNet	IgG4-related kidney disease	ORPHA:449395
Dr RIBRAG Vincent		Mantle cell lymphoma	ORPHA:52416
		MALT lymphoma	ORPHA:52417
		Follicular lymphoma	ORPHA:545
Dr RICCARDI Florence	ITHACA	KBG syndrome	ORPHA:2332
Pr RICHETTE Pascal		Familial calcium pyrophosphate deposition	ORPHA:1416
Dr RIEGER Melissa	ITHACA	Childhood apraxia of speech	ORPHA:209908
Dr ROBAK Tadeusz		Classic hairy cell leukemia	ORPHA:58017
Dr ROMANINI Maria Victoria		Poland syndrome	ORPHA:2911
Pr ROCCATELLO Dario	ERKNet	Anti-glomerular basement membrane disease	ORPHA:375
		Pauci-immune glomerulonephritis	ORPHA:93126
Pr RONCO Pierre	ERKNet	Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	ORPHA:69063

Expert	ERN Affiliation	Disease name	ORPHAcode
Dr ROSSIGNOL Julien		Mastocytosis	ORPHA:98292
Dr ROTHENBUHLER Anya		Hypophosphatasia	ORPHA:436
Dr ROUSSEAU Geoffroy		Congenital diaphragmatic hernia	ORPHA:2140
Dr ROUX Anne Françoise		Usher syndrome type 3	ORPHA:231183
		Usher syndrome type 2	ORPHA:231178
		Usher syndrome type 1	ORPHA:231169
		Usher syndrome	ORPHA:886
Dr RUSHING Elisabeth		Focal myositis	ORPHA:48918
Pr RUTKOWSKI Stefan		Medulloblastoma	ORPHA:616
Pr SAADOUN David		Behçet disease	ORPHA:117
		Hereditary pediatric Behþet-like disease	ORPHA:476102
Pr SACCONI Sabrina	EURO-NMD	Facioscapulohumeral dystrophy	ORPHA:269
Pr SACRE Karim		Susac syndrome	ORPHA:838
Dr SAHM Felix		Meningioma	ORPHA:2495
Dr SANCHEZ FERNAN Ivan		Landau-Kleffner syndrome	ORPHA:98818
Dr SANCHEZ FERNANDEZ Mayka		Congenital dyserythropoietic anemia type I	ORPHA:98869
Dr SANGIORGI Luca		Nail-patella syndrome	ORPHA:2614
Pr SANLAVILLE Damien	ITHACA	Distal Xq28 microduplication syndrome	ORPHA:293939
Pr SANTORELLI Filippo		NARP syndrome	ORPHA:644
SARKAR Hajrah		Congenital primary aphakia	ORPHA:83461
Dr SASAI Hideo		Beta-ketothiolase deficiency	ORPHA:134
Dr SAVALE Laurent		Pulmonary arterial hypertension	ORPHA:182090
Dr SCALA Marcello		Freeman-Sheldon syndrome	ORPHA:2053
Dr SCARANO Emanuela		Noonan syndrome-like disorder with loose anagen hair	ORPHA:2701
Dr SCHLUTH-BOLARD Caroline	ITHACA	Distal Xq28 microduplication syndrome	ORPHA:293939
SCHNEIDER Katherine		Li-Fraumeni syndrome	ORPHA:524
Dr SCHREUDER Michiel	eUROGEN & ERKNet	Multicystic dysplastic kidney	ORPHA:1851
		Unilateral multicystic dysplastic kidney	ORPHA:97363
		Bilateral multicystic dysplastic kidney	ORPHA:97364
Dr SCHUHMANN Sarah	ITHACA	CTCF-related neurodevelopmental disorder	ORPHA:363611
Pr SCHUSTER Volker		Ligneous conjunctivitis	ORPHA:97231
Pr SCHWARTZ Peter		Timothy syndrome	ORPHA:65283
		Romano-Ward syndrome	ORPHA:101016
		Jervell and Lange-Nielsen syndrome	ORPHA:90647
Dr SENTARO Kusuhara		Pierson syndrome	ORPHA:2670
Dr SHERR Elliott		Temtamy syndrome	ORPHA:1777
Dr SILVE Caroline		Acrodysostosis with multiple hormone resistance	ORPHA:280651
Dr SMOL Thomas	ITHACA	Developmental delay-facial dysmorphism	ORPHA:369891

Expert	ERN Affiliation	Disease name	ORPHAcode
		syndrome due to MED13L deficiency	
Dr SNAST Igor		Solar urticaria	ORPHA:97230
Dr SNEAD Martin		Stickler syndrome	ORPHA:828
Dr SOLDNER Roland		Congenital diaphragmatic hernia	ORPHA:2140
Pr STEINMANN Beat		Arthrochalasia Ehlers-Danlos syndrome	ORPHA:1899
Dr STRUB Marion		Dental ankylosis	ORPHA:1077
Pr STUMPEL C.T.R.M. [Connie]	ITHACA	L1 syndrome	ORPHA:275543
		Hydrocephalus with stenosis of the aqueduct of Sylvius	ORPHA:2182
		MASA syndrome	ORPHA:2466
		X-linked complicated spastic paraplegia type 1	ORPHA:306617
		X-linked complicated corpus callosum dysgenesis	ORPHA:1497
Dr SUGARMAN Jeffrey		Linear nevus sebaceus syndrome	ORPHA:2612
Pr SUNADA Yoshihide		MELAS	ORPHA:550
Pr SUSEN Sophie		Von Willebrand disease	ORPHA:903
		Von Willebrand disease type 1	ORPHA:166078
Dr SUZUKI Tamio		Oculocutaneous albinism type 6	ORPHA:370097
		Oculocutaneous albinism type 5	ORPHA:370091
		Oculocutaneous albinism type 7	ORPHA:352745
		Temperature-sensitive oculocutaneous albinism type 1	ORPHA:352737
		Minimal pigment oculocutaneous albinism type 1	ORPHA:352734
		Oculocutaneous albinism type 1	ORPHA:352731
		Oculocutaneous albinism	ORPHA:55
		Oculocutaneous albinism type 1A	ORPHA:79431
		Oculocutaneous albinism type 2	ORPHA:79432
		Oculocutaneous albinism type 3	ORPHA:79433
		Oculocutaneous albinism type 1B	ORPHA:79434
		Oculocutaneous albinism type 4	ORPHA:79435
Dr SZAKSZON Katalin	ITHACA	Oculocerebrofacial syndrome, Kaufman type	ORPHA:2707
Dr TAMBURRINO Federica		Noonan syndrome-like disorder with loose anagen hair	ORPHA:2701
Pr TAZI Abdellatif		Langerhans cell histiocytosis	ORPHA:389
Dr TENORIO CASTAÑO Jair Antonio		Simpson-Golabi-Behmel syndrome	ORPHA:373
Pr TERRIER Benjamin		Cutaneous small vessel vasculitis	ORPHA:889
		Eosinophilic granulomatosis with polyangiitis	ORPHA:183
		Microscopic polyangiitis	ORPHA:727
		Polyarteritis nodosa	ORPHA:767
		Primary angiitis of the central nervous system	ORPHA:140989
Dr TESTONI Alberto		NON RARE IN EUROPE: Recurrent acute	ORPHA:64740

Expert	ERN Affiliation	Disease name	ORPHAcode
		pancreatitis	
Dr TIMOTHY Katherine		Timothy syndrome	ORPHA:65283
Pr TOMATSU Shunji		Mucopolysaccharidosis type 4	ORPHA:582
Dr TORRE Michele		Poland syndrome	ORPHA:2911
Dr TREMOLIERES François		Tetanus	ORPHA:3299
Dr TREPICCIONE Francesco	ERKNet	Fanconi-Bickel syndrome	ORPHA:2088
Dr TRIMARCHI Gabriele	ITHACA	SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:157965
Dr TSENG MinHua	ERKNet	Transient pseudohypoaldosteronism	ORPHA:93164
Dr TSUBOI Yoshio		Spinocerebellar ataxia type 34	ORPHA:1955
		Spinocerebellar ataxia type 30	ORPHA:211017
		Spinocerebellar ataxia type 6	ORPHA:98758
		Spinocerebellar ataxia type 37	ORPHA:363710
		Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	ORPHA:314404
		Spinocerebellar ataxia type 7	ORPHA:94147
		Spinocerebellar ataxia type 5	ORPHA:98766
		Spinocerebellar ataxia type 29	ORPHA:208513
		Spinocerebellar ataxia type 31	ORPHA:217012
		Spinocerebellar ataxia type 32	ORPHA:276183
		Spinocerebellar ataxia type 35	ORPHA:276193
		Spinocerebellar ataxia type 36	ORPHA:276198
Pr TÜMER Zeynep	ITHACA	Menkes disease	ORPHA:565
		Occipital horn syndrome	ORPHA:198
Dr TURLEAU Catherine		Cat-eye syndrome	ORPHA:195
Pr VALENTE Enza Maria		Joubert syndrome	ORPHA:475
Dr VALENZUELA Irene	ITHACA	Short stature-brachydactyly-obesity-global developmental delay syndrome	ORPHA:464288
		Cerebellar-facial-dental syndrome	ORPHA:444072
Dr VAN DER SPEK Jet		Witteveen-Kolk syndrome	ORPHA:500163
Dr VAN EERDE A.M. [Albertien]	ERKNet	Fraser syndrome	ORPHA:2052
Dr VAN WIJK R. [Richard]		Hemolytic anemia due to red cell pyruvate kinase deficiency	ORPHA:766
Dr VAULOUP-FELLOUS Christelle		Congenital rubella syndrome	ORPHA:290
		Congenital enterovirus infection	ORPHA:292
Dr VAUX Julien		Hypophosphatasia	ORPHA:436
Dr VERDIN Hannah		Blepharophimosis-ptosis-epicanthus inversus syndrome	ORPHA:126
Pr VERLOES Alain	ITHACA & ERKNet	Neu-Laxova syndrome	ORPHA:2671
		Serine biosynthesis pathway deficiency, infantile/juvenile form	ORPHA:583595

Expert	ERN Affiliation	Disease name	ORPHAcode
		Neurometabolic disorder due to serine deficiency	ORPHA:35705
		Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	ORPHA:363972
		Galloway-Mowat syndrome	ORPHA:2065
		Hypertelorism, Teebi type	ORPHA:1519
		Noonan syndrome	ORPHA:648
		Opitz G/BBB syndrome	ORPHA:2745
Dr VICART Savine		Hypokalemic periodic paralysis	ORPHA:681
Pr VIGOUROUX Corinne		Insulin-resistance syndrome type B	ORPHA:2298
		Congenital generalized lipodystrophy	ORPHA:528
		Familial partial lipodystrophy, Dunnigan type	ORPHA:2348
		Familial partial lipodystrophy	ORPHA:98306
Pr VIKKULA Miikka		Glomuvenous malformation	ORPHA:83454
Dr VILAIN Catheline	ITHACA	Hartsfield syndrome	ORPHA:2117
Dr VILARINHO Laura		3-hydroxy-3-methylglutaric aciduria	ORPHA:20
Dr VIVARELLI Marina	ERKNet	Idiopathic steroid-sensitive nephrotic syndrome	ORPHA:69061
		Congenital membranous nephropathy due to fetomaternal anti-neutral endopeptidase alloimmunization	ORPHA:69063
Dr VOCKLEY Gerard		Isovaleric acidemia	ORPHA:33
Pr VOCKLEY Jerry		Isovaleric acidemia	ORPHA:33
Dr VOS Y.J. [Yvonne]	ITHACA	Hydrocephalus with stenosis of the aqueduct of Sylvius	ORPHA:2182
		MASA syndrome	ORPHA:2466
		X-linked complicated spastic paraplegia type 1	ORPHA:306617
		X-linked complicated corpus callosum dysgenesis	ORPHA:1497
		L1 syndrome	ORPHA:275543
Pr WALLON Martine		Congenital toxoplasmosis	ORPHA:858
Dr WANG Heng		Cohen syndrome	ORPHA:193
Pr WIECZOREK Dagmar	ITHACA	Postaxial acrofacial dysostosis	ORPHA:246
		Burn-McKeown syndrome	ORPHA:1200
		Mandibulofacial dysostosis-microcephaly syndrome	ORPHA:79113
Pr WIERSINGA W.J. [Joost]		Melioidosis	ORPHA:31202
Dr WIJSENBEEK M.S. [Marlies]		Idiopathic pulmonary fibrosis	ORPHA:2032
Dr WILKINSON Mark		Stargardt disease	ORPHA:827
Dr WOLF Barry		Multiple carboxylase deficiency	ORPHA:148
		Biotinidase deficiency	ORPHA:79241
		Holocarboxylase synthetase deficiency	ORPHA:79242
Dr WOLF Nicole		4H leukodystrophy	ORPHA:289494
Dr WORTMANN-HAGEMANN		3-methylglutaconic aciduria type 1	ORPHA:67046

Expert	ERN Affiliation	Disease name	ORPHAcode
Saskia			
Pr WOUTERS Carine H.		Blau syndrome	ORPHA:90340
Dr WSZOLEK Zbigniew		Spinocerebellar ataxia type 34	ORPHA:1955
		Spinocerebellar ataxia type 30	ORPHA:211017
		Spinocerebellar ataxia type 6	ORPHA:98758
		Spinocerebellar ataxia type 37	ORPHA:363710
		Autosomal dominant cerebellar ataxia-deafness-narcolepsy syndrome	ORPHA:314404
		Spinocerebellar ataxia type 5	ORPHA:98766
		Spinocerebellar ataxia type 29	ORPHA:208513
		Spinocerebellar ataxia type 32	ORPHA:276183
		Spinocerebellar ataxia type 35	ORPHA:276193
		Spinocerebellar ataxia type 36	ORPHA:276198
		Spinocerebellar ataxia type 31	ORPHA:217012
		Spinocerebellar ataxia type 7	ORPHA:94147
Dr ZACCHIA Miriam	ERKNet	Hereditary renal hypouricemia	ORPHA:94088
Dr ZANGEN David		Congenital lipoid adrenal hyperplasia due to STAR deficency	ORPHA:90790
Pr ZWEIER Christiane		CTCF-related neurodevelopmental disorder	ORPHA:363611
		Childhood apraxia of speech	ORPHA:209908

List of expert networks reviewing Orphanet data in 2020

Name of expert network	Disease name	ORPHA code
EpiCARE	FOXG1 syndrome	ORPHA:561854
ERKNet	Hypertension due to gain-of-function mutations in the mineralocorticoid receptor	ORPHA:88660
ERN-Skin	Inherited epidermolysis bullosa	ORPHA:79361

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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