Orphanet Report Series

Rare Diseases collection

2022

Expert reviewers for Orphanet in 2022

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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 to abstracts/definitions are described in a second list and third list.

List of expert reviewers for Orphanet in 2022

Expert name	Disease name	ORPHA
Dr David G. ANDERSON	Huntington disease-like 2	ORPHA:98934
Dr Aveline AOUIDAD	Non-specific autoimmune supratentorial encephalitis without characteristic antibodies	ORPHA:624178
Pr Stefan ARETZ	Familial colorectal cancer Type X	ORPHA:440437
Pr Gema ARICETA	Dent disease	ORPHA:1652
	Autosomal recessive distal renal tubular acidosis	ORPHA:402041
	Autosomal recessive proximal renal tubular acidosis	ORPHA:93607
	Proximal renal tubular acidosis	ORPHA:47159
	Distal renal tubular acidosis with anemia	ORPHA:93610
	Autosomal dominant proximal renal tubular acidosis	ORPHA:314889
	Distal renal tubular acidosis	ORPHA:18
	Autosomal dominant distal renal tubular acidosis	ORPHA:93608
Dr Jean-Baptiste ARNOUX	Congenital isolated hyperinsulinism	ORPHA:657
	Hyperinsulinism-hyperammonemia syndrome	ORPHA:35878
	Diazoxide-resistant hyperinsulinism	ORPHA:276585
	Hyperinsulinism due to UCP2 deficiency	ORPHA:276556
	Congenital hyperinsulinism due to HNF4A deficiency	ORPHA:263455
	Exercise-induced hyperinsulinism	ORPHA:165991
	Autosomal dominant hyperinsulinism due to SUR1 deficiency	ORPHA:276575
	Congenital glucokinase-related hyperinsulinism	ORPHA:79299
	Diazoxide-resistant focal hyperinsulinism	ORPHA:79298
	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	ORPHA:276598
	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	ORPHA:71212
Dr Gilles BAGOU	Primary Sjögren syndrome	ORPHA:289390
	Hemophilia	ORPHA:448
	Autoimmune polyendocrinopathy type 1	ORPHA:3453
	Rare aplastic anemia	ORPHA:182040
	Lymphangioleiomyomatosis	ORPHA:538
	Bullous pemphigoid	ORPHA:703
	Blackfan-Diamond anemia	ORPHA:124
	Hypokalemic periodic paralysis	ORPHA:681
	Crigler-Najjar syndrome	ORPHA:205
	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136

	Glanzmann thrombasthenia	ORPHA:849
	Narcolepsy type 1	ORPHA:2073
	Interstitial cystitis	ORPHA:37202
Dr Eulalia BASELGA TORRES	Autosomal dominant epidermolytic ichthyosis	ORPHA:312
Dr Daniel BATLLE	Distal renal tubular acidosis	ORPHA:18
	Proximal renal tubular acidosis	ORPHA:47159
Pr Olivier BENVENISTE	Polymyositis	ORPHA:732
Pr Enrico S. BERTINI	Walker-Warburg syndrome	ORPHA:899
	Muscle-eye-brain disease	ORPHA:588
	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	ORPHA:370997
	Congenital muscular dystrophy, Fukuyama type	ORPHA:272
Dr Gérald BERTRAND	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853
Dr Stefania BIGONI	Recessive KLHL7-related disorder	ORPHA:603699
	KLHL7-related Crisponi/cold-induced sweating-like syndrome	ORPHA:603694
	KLHL7-related Bohring-Opitz-like and Crisponi/Cold-induced sweating-like overlap syndrome	ORPHA:603684
	KLHL7-related Bohring-Opitz-like syndrome	ORPHA:603689
Pr Anthony J. BLEYER	UMOD-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88950
	REN-related autosomal dominant tubulointerstitial kidney disease	ORPHA:217330
	MUC1-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88949
Dr M.C. [Marieke] BOLLING	Punctate palmoplantar keratoderma type 1	ORPHA:79501
Dr Juli BOUTEILLER	Bullous pemphigoid	ORPHA:703
Dr Michela BRENA	Harlequin ichthyosis	ORPHA:457
Dr CatherineCARDOT-BAUTERS	Autoimmune polyendocrinopathy type 1	ORPHA:3453
Dr Pierre CATOIRE	Crigler-Najjar syndrome	ORPHA:205
Pr Hugues CHABRIAT	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
Dr Djamila CHAÏBA	Primary Sjögren syndrome	ORPHA:289390
Dr Antony CHAUVIN	Primary Sjögren syndrome	ORPHA:289390
	Hemophilia	ORPHA:448
	Autoimmune polyendocrinopathy type 1	ORPHA:3453
	Rare aplastic anemia	ORPHA:182040
	Lymphangioleiomyomatosis	ORPHA:538
	Hypokalemic periodic paralysis	ORPHA:681
	Blackfan-Diamond anemia	ORPHA:124
	Bullous pemphigoid	ORPHA:703
	Crigler-Najjar syndrome	ORPHA:205
	Glanzmann thrombasthenia	ORPHA:849

	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
	Narcolepsy type 1	ORPHA:2073
	Interstitial cystitis	ORPHA:37202
Pr Vincent COTTIN	Lymphangioleiomyomatosis	ORPHA:538
	Acute interstitial pneumonia	ORPHA:79126
	Hypersensitivity pneumonitis	ORPHA:31740
	Cryptogenic organizing pneumonia	ORPHA:1302
	Lymphangioleiomyomatosis	ORPHA:538
Dr John A. COULTER	Hydatidiform mole	ORPHA:99927
Pr Yesim DARGAUD	Hemophilia	ORPHA:448
	Hemophilia A	ORPHA:98878
	Bleeding disorder in hemophilia B carriers	ORPHA:177929
	Mild hemophilia B	ORPHA:169799
	Mild hemophilia A	ORPHA:169808
	Moderate hemophilia B	ORPHA:169796
	Hemophilia B	ORPHA:98879
	Bleeding disorder in hemophilia A carriers	ORPHA:177926
	Severe hemophilia B	ORPHA:169793
	Severe hemophilia A	ORPHA:169802
	Hemophilia B Leyden	ORPHA:617930
	Hemophilia	ORPHA:448
Vincent DARROUZET	Semicircular canal dehiscence syndrome	ORPHA:420402
Pr Yves DAUVILLIERS	Narcolepsy type 1	ORPHA:2073
Pr Kumaran DEIVA	Non-specific autoimmune brainstem encephalitis with characteristic antibodies	ORPHA:624199
	Non-specific autoimmune cerebellar ataxia with characteristic antibodies	ORPHA:624259
	Paraneoplastic isolated brainstem encephalitis	ORPHA:624190
	Non-specific autoimmune supratentorial encephalitis with characteristic antibodies	ORPHA:624166
	Postinfectious cerebellitis	ORPHA:624244
	Non-specific autoimmune brainstem encephalitis without characteristic antibodies	ORPHA:624216
	OBSOLETE: Limbic encephalitis	ORPHA:163892
	OBSOLETE: Limbic encephalitis with DPP6 antibodies	ORPHA:329341
	OBSOLETE: Limbic encephalitis with caspr2 antibodies	ORPHA:276402
	OBSOLETE: Limbic encephalitis	ORPHA:163892
Dr Alessandro DEPAOLI	Congenital pseudoarthrosis of the clavicle	ORPHA:66630
Pr F.G. [Frederik] DIKKERS	Recurrent respiratory papillomatosis	ORPHA:60032
Dr Dries DOBBELAERE	Hyperammonemia due to N-acetylglutamate synthase deficiency	ORPHA:927
Dr Roni P DODIUK-GAD	Darier disease	ORPHA:218

Dr Helene DOLLFUS	Familial pterygium of the conjunctiva	ORPHA:2989
Dr Bénédicte DOUAY	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:3453
Dr Xavier DUBUCS	Narcolepsy type 1	ORPHA:2073
	Glanzmann thrombasthenia	ORPHA:849
Pr Francesco EMMA	Dent disease	ORPHA:1652
Dr Marianna FARNÈ	KLHL7-related Crisponi/cold-induced sweating-like syndrome	ORPHA:603694
	KLHL7-related Bohring-Opitz-like syndrome	ORPHA:603689
	Recessive KLHL7-related disorder	ORPHA:603699
Dr Roberta FENOGLIO	Pauci-immune glomerulonephritis with ANCA	ORPHA:97563
	Pauci-immune glomerulonephritis without ANCA	ORPHA:97564
Pr Alessandra FERLINI	KLHL7-related Bohring-Opitz-like and Crisponi/Cold-induced sweating-like overlap syndrome	ORPHA:603684
	KLHL7-related Bohring-Opitz-like syndrome	ORPHA:603689
	KLHL7-related Crisponi/cold-induced sweating-like syndrome	ORPHA:603694
	Recessive KLHL7-related disorder	ORPHA:603699
Aline FERREIRA-CORREIA	Huntington disease-like 2	ORPHA:98934
Dr Mathieu FIORE	Glanzmann thrombasthenia	ORPHA:849
Pr Judith FISCHER	Congenital ichthyosiform erythroderma	ORPHA:79394
	Mal de Meleda	ORPHA:87503
Dr Carlo FUSCO	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	ORPHA:500144
Dr Abhimanyu GARG	SAMD9L-associated autoinflammatory syndrome	ORPHA:619367
Pr Dominique P. GERMAIN	Fabry disease	ORPHA:324
Dr Laetitia GIORGI	Immune-mediated cerebellar ataxia	ORPHA:623638
Dr Sophie GUEZ	Harlequin ichthyosis	ORPHA:457
Dr Robert A. HEGELE	Homozygous familial hypercholesterolemia	ORPHA:391665
	Familial chylomicronemia syndrome	ORPHA:444490
Dr Angela HERNÁNDEZ-MARTÍN	Recessive X-linked ichthyosis	ORPHA:461
	Syndromic recessive X-linked ichthyosis	ORPHA:281090
Dr Dominique HERVÉ	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
Pr Olaf HIORT	46,XX gonadal dysgenesis	ORPHA:243
	46,XY complete gonadal dysgenesis	ORPHA:242
	45,X/46,XY mixed gonadal dysgenesis	ORPHA:1772
	46,XY partial gonadal dysgenesis	ORPHA:251510
Dr Linda HUMBERT	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:3453
Pr Pascal JOLY	Bullous pemphigoid	ORPHA:703
Dr Nathalie JONCA	Acral peeling skin syndrome	ORPHA:263534
	Peeling skin syndrome type B	ORPHA:263553
	Peeling skin syndrome type A	ORPHA:263548

Pr Guillaume JONDEAU	X-linked severe syndromic thoracic aortic aneurysm and dissection	ORPHA:622925
Dr Bastien JOUBERT	Non-specific autoimmune brainstem encephalitis with characteristic antibodies	ORPHA:624199
	Non-specific autoimmune cerebellar ataxia with characteristic antibodies	ORPHA:624259
	Paraneoplastic isolated brainstem encephalitis	ORPHA:624190
	Non-specific autoimmune supratentorial encephalitis with characteristic antibodies	ORPHA:624166
	Postinfectious cerebellitis	ORPHA:624244
	Autoimmune limbic encephalitis	ORPHA:623615
	Autoimmune encephalitis	ORPHA:622014
	Paraneoplastic cerebellar degeneration	ORPHA:623626
	Non-specific autoimmune supratentorial encephalitis without characteristic antibodies	ORPHA:624178
	Non-specific autoimmune cerebellar ataxia without characteristic antibodies	ORPHA:624268
	Immune-mediated cerebellar ataxia	ORPHA:623638
	OBSOLETE: Non-herpetic acute limbic encephalitis	ORPHA:163924
	OBSOLETE: Limbic encephalitis	ORPHA:163892
	OBSOLETE: Limbic encephalitis with DPP6 antibodies	ORPHA:329341
	OBSOLETE: Limbic encephalitis with caspr2 antibodies	ORPHA:276402
	OBSOLETE: Paraneoplastic limbic encephalitis	ORPHA:163895
	OBSOLETE: Non-paraneoplastic limbic encephalitis	ORPHA:163918
	Paraneoplastic sensory ganglionopathy	ORPHA:208999
	OBSOLETE: Limbic encephalitis	ORPHA:163892
Dr Nobuo KANAZAWA	SAMD9L-associated autoinflammatory syndrome	ORPHA:619367
Pr Jean KANITAKIS	Circumscribed palmoplantar hypokeratosis	ORPHA:69744
Dr Baljeet KAUR	Partial hydatidiform mole	ORPHA:254693
	Epithelioid trophoblastic tumor	ORPHA:254698
	Complete hydatidiform mole	ORPHA:254688
	Gestational choriocarcinoma	ORPHA:99926
	Invasive mole	ORPHA:99925
	Gestational trophoblastic neoplasm	ORPHA:59305
	Placental site trophoblastic tumor	ORPHA:99928
Pr François KERBAUL	Primary Sjögren syndrome	ORPHA:289390
	Hemophilia	ORPHA:448
	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:3453
	Rare aplastic anemia	ORPHA:182040
	Lymphangioleiomyomatosis	ORPHA:538
	Bullous pemphigoid	ORPHA:703

	Hypokalemic periodic paralysis	ORPHA:681
	Interstitial cystitis	ORPHA:37202
	Blackfan diamond anemia	ORPHA:124
	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
	Thrombasthénie de Glanzmann	ORPHA:849
	Narcolepsy type 1	ORPHA:2073
	Crigler-Najjar syndrome	ORPHA:205
Dr Stanislav KMOCH	REN-related autosomal dominant tubulointerstitial kidney disease	ORPHA:217330
	UMOD-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88950
	MUC1-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88949
Dr Stefan KOHL	Renal dysplasia, unilateral	ORPHA:93172
Pr Philippe LABRUNE	Crigler-Najjar syndrome	ORPHA:205
Dr Pierrick LE BORGNE	Bullous pemphigoid	ORPHA:703
Pr Thierry LEBLANC	Blackfan-Diamond anemia	ORPHA:124
Dr Sylvain LECAILTEL	Rare aplastic anemia	ORPHA:182040
Pr Juliane LEGER	Congenital adrenal hyperplasia	ORPHA:418
	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	ORPHA:95699
	Congenital lipoid adrenal hyperplasia due to STAR deficency	ORPHA:90790
	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	ORPHA:90793
	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	ORPHA:90794
	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	ORPHA:90795
Dr Dorothee LEROUX	Familial pterygium of the conjunctiva	ORPHA:2989
Dr Christophe LEROY	Primary Sjögren syndrome	ORPHA:289390
	Hemophilia	ORPHA:448
	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:3453
	Rare aplastic anemia	ORPHA:182040
	Lymphangioleiomyomatosis	ORPHA:538
	Hypokalemic periodic paralysis	ORPHA:681
	Bullous pemphigoid	ORPHA:703
	Interstitial cystitis	ORPHA:37202
	Blackfan diamond	ORPHA:124
	Glanzmann thrombasthenia	ORPHA:
	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
	Narcolepsy type 1	ORPHA:2073
	Crigler-Najjar syndrome	ORPHA:205

Dr Anne LIENHART	Hemophilia	ORPHA:448
Pr Agnès LINGLART	Hypophosphatemic rickets	ORPHA:437
	X-linked hypophosphatemia	ORPHA:89936
	Hypocalcemic vitamin D-dependent rickets	ORPHA:289157
	Hypocalcemic vitamin D-resistant rickets	ORPHA:93160
	Autosomal dominant hypophosphatemic rickets	ORPHA:89937
	Autosomal recessive hypophosphatemic rickets	ORPHA:289176
	Hypocalcemic rickets	ORPHA:289103
	Hereditary hypophosphatemic rickets with hypercalciuria	ORPHA:157215
Dr Christianne LOK	Gestational choriocarcinoma	ORPHA:99926
	Partial hydatidiform mole	ORPHA:254693
	Gestational trophoblastic neoplasm	ORPHA:59305
	Invasive mole	ORPHA:99925
	Epithelioid trophoblastic tumor	ORPHA:254698
	Complete hydatidiform mole	ORPHA:254688
	Placental site trophoblastic tumor	ORPHA:99928
Pr Michelangelio MANCUSO	MERRF	ORPHA:551
Pr Xavier MARIETTE	Primary Sjögren syndrome	ORPHA:289390
Pr Juliette MAZEREEUW-HAUTIER	Peeling skin syndrome type B	ORPHA:263553
	Generalized peeling skin syndrome	ORPHA:263543
	Peeling skin syndrome type A	ORPHA:263548
	Acral peeling skin syndrome	ORPHA:263534
	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	ORPHA:444138
	Peeling skin syndrome	ORPHA:817
Dr Francine MECKERT	Primary Sjögren syndrome	ORPHA:289390
	Hemophilia	ORPHA:448
	Polyendocrinopathie auto-immune type 1 / APECED	ORPHA:3453
	Rare aplastic anemia	ORPHA:182040
	Lymphangioleiomyomatosis	ORPHA:538
	Hypokalemic periodic paralysis	ORPHA:681
	Bullous pemphigoid	ORPHA:703
	Crigler-Najjar syndrome	ORPHA:205
	Blackfan-Diamond anemia	ORPHA:124
	Glanzmann thrombasthenia	ORPHA:849
	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
	Narcolepsy type 1	ORPHA:2073
	Interstitial cystitis	ORPHA:37202
Dr Camilla MEOSSI	Ochoa syndrome	ORPHA:2704
Dr Marie-France MERCIER	Primary Sjögren syndrome	ORPHA:289390
	Blackfan-Diamond anemia	ORPHA:124

	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
	Rare aplastic anemia	ORPHA:182040
Dr Donatella MILANI	Ochoa syndrome	ORPHA:2704
Dario G. MINOLI	Ochoa syndrome	ORPHA:2704
Dr Fanny MORICE-PICARD	Trichothiodystrophy	ORPHA:33364
Dr Giovanni MOSIELLO	Caudal regression syndrome	ORPHA:3027
	Familial caudal dysgenesis	ORPHA:1768
	Sirenomelia	ORPHA:3169
	Caudal regression-sirenomelia spectrum	ORPHA:444941
Pr Rima NABBOUT	Infantile spasms syndrome	ORPHA:3451
Dr Eithne M. NIC AN RÍOGH	Drug-induced vasculitis	ORPHA:251325
Dr Gaëtane NOCTURNE	Primary Sjögren syndrome	ORPHA:289390
Dr Mathieu OBERLIN	Interstitial cystitis	ORPHA:37202
Pr Saskia ORO	Stevens-Johnson syndrome/toxic epidermal necrolysis overlap syndrome	ORPHA:506784
	Acute generalized exanthematous pustulosis	ORPHA:293173
	Fixed drug eruption	ORPHA:293812
	Erythema multiforme major	ORPHA:502499
	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	ORPHA:95455
	Toxic epidermal necrolysis	ORPHA:537
	Drug reaction with eosinophilia and systemic symptoms	ORPHA:139402
Pr Edel A O'TOOLE	Pachyonychia congenita	ORPHA:2309
Dr Myriam PASTURAL	Narcolepsy type 1	ORPHA:2073
	Glanzmann thrombasthenia	ORPHA:849
Pr Régis PEFFAULT DE LATOUR	Rare aplastic anemia	ORPHA:182040
Arnaud PERAMO	Hypophosphatemic rickets	ORPHA:437
	Hereditary hypophosphatemic rickets with hypercalciuria	ORPHA:157215
	Autosomal recessive hypophosphatemic rickets	ORPHA:289176
	Hypocalcemic rickets	ORPHA:289103
Pr Nicolas PESCHANSKI	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	ORPHA:136
Dr Matthieu PEYRE	Mosaic Legius syndrome	ORPHA:634511
	Mosaic schwannomatosis	ORPHA:634492
	Mosaic NF2-related schwannomatosis	ORPHA:634475
	Mosaic neurofibromatosis type 1	ORPHA:634461
	Neurofibromatosis/schwannomatosis	ORPHA:634518
Dr Aldona PIETRZAK	CHILD syndrome	ORPHA:139
Dr Marta PINEDA RÍU	Familial colorectal cancer Type X	ORPHA:440437
Pr Susana QUIJANO-ROY	Muscle-eye-brain disease	ORPHA:588
	Congenital muscular dystrophy, Fukuyama type	ORPHA:272
	Muscle-eye-brain disease with bilateral	ORPHA:370997

	multicystic leucodystrophy	
	Walker-Warburg syndrome	ORPHA:899
Dr Nur Villar QUILES	Hypokalemic periodic paralysis	ORPHA:681
Pr Niels QVIST	Esophageal atresia	ORPHA:1199
	Esophageal duplication cyst	ORPHA:100047
	Congenital esophageal diverticulum	ORPHA:91358
	Tubular duplication of the esophagus	ORPHA:100048
Dr Melissa RIEGER	Isolated childhood apraxia of speech	ORPHA:209908
Dr Susanna RIZZI	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	ORPHA:500144
Pr Dario ROCCATELLO	Pauci-immune glomerulonephritis with ANCA	ORPHA:97563
	Pauci-immune glomerulonephritis without ANCA	ORPHA:97564
Dr Anya ROTHENBUHLER	Hypophosphatemic rickets	ORPHA:437
	X-linked hypophosphatemia	ORPHA:89936
	Hereditary hypophosphatemic rickets with hypercalciuria	ORPHA:157215
	Hypocalcemic vitamin D-dependent rickets	ORPHA:289157
	Autosomal recessive hypophosphatemic rickets	ORPHA:289176
	Autosomal dominant hypophosphatemic rickets	ORPHA:89937
	Hypocalcemic vitamin D-resistant rickets	ORPHA:93160
	Hypocalcemic rickets	ORPHA:289103
Dr Geoffroy ROUSSEAU	Hypokalemic periodic paralysis	ORPHA:681
Dr Cécile SAINT-MARTIN	Congenital isolated hyperinsulinism	ORPHA:657
Dr Emmanuelle SAMSON	Caudal regression syndrome	ORPHA:3027
	Familial caudal dysgenesis	ORPHA:1768
	Caudal regression-sirenomelia spectrum	ORPHA:444941
	Sirenomelia	ORPHA:3169
Pr Christian SAUSSINE	Interstitial cystitis	ORPHA:37202
Ariane SCHMETZ	BOR syndrome	ORPHA:107
Pr Matthias SCHMUTH	CHILD syndrome	ORPHA:139
Pr M.F. [Michiel] SCHREUDER	Idiopathic steroid-resistant nephrotic syndrome	ORPHA:567548
	Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	ORPHA:567546
Pr Savino SCIASCIA	Idiopathic non-lupus full-house nephropathy	ORPHA:567544
Dr Flore SICRE DE FONTBRUNE	Rare aplastic anemia	ORPHA:182040
Dr Jacqueline SILLERAN CHASSANY	Autoimmune polyendocrinopathy type 1	ORPHA:3453
	Bullous pemphigoid	ORPHA:703
	Lymphangioleiomyomatosis	ORPHA:538
Dr Kira SÜßMUTH	Lamellar ichthyosis	ORPHA:313
Dr Laure THOMAS	OBSOLETE: Classic paraneoplastic limbic encephalitis	ORPHA:163898
Dr Antonio TORRELO	SAMD9L-associated autoinflammatory syndrome	ORPHA:619367
Dr Giovanni TRISOLINO	Congenital pseudoarthrosis of the clavicle	ORPHA:66630
Dr Laura VALLE	Familial colorectal cancer Type X	ORPHA:440437

Dr P.C. [Peter] VAN DEN AKKER	Punctate palmoplantar keratoderma type 1	ORPHA:79501
Pr Marie-Christine VANTYGEM	Autoimmune polyendocrinopathy type 1	ORPHA:3453
Dr Savine VICART	Hypokalemic periodic paralysis	ORPHA:681
Dr Barbara VILLOING	Rare aplastic anemia	ORPHA:182040
	Blackfan-Diamond anemia	ORPHA:124
	Lymphangioleiomyomatosis	ORPHA:538
Dr Marina VIVARELLI	Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	ORPHA:567546
	Idiopathic steroid-resistant nephrotic syndrome	ORPHA:567548
Pr Dagmar WIECZOREK	BOR syndrome	ORPHA:107
Pr Norbert WINER	Fetal and neonatal alloimmune thrombocytopenia	ORPHA:853
Pr Pierre WOLKENSTEIN	Mosaic Legius syndrome	ORPHA:634511
	Mosaic schwannomatosis	ORPHA:634492
	Mosaic NF2-related schwannomatosis	ORPHA:634475
	Mosaic neurofibromatosis type 1	ORPHA:634461
	Neurofibromatosis/schwannomatosis	ORPHA:634518
Dr Ekaterina ZAKHAROVA	Chronic neurovisceral acid sphingomyelinase deficiency	ORPHA:618891
	Acid sphingomyelinase deficiency	ORPHA:618899
Dr Paola ZARANTONELLO	Congenital pseudoarthrosis of the clavicle	ORPHA:66630
Dr Martina ZIVNA	REN-related autosomal dominant tubulointerstitial kidney disease	ORPHA:217330
Pr Christiane ZWEIER	Isolated childhood apraxia of speech	ORPHA:209908

List of expert networks reviewing the Orphanet nomenclature in 2022

Name of expert network	Disease name	ORPHAcode
ERKNet	Primary hypomagnesemia-generalized seizures-intellectual disability-obesity syndrome	ORPHA:620363
ERKNet	EGF-related primary hypomagnesemia with intellectual disability	ORPHA:620368
ERKNet	Gitelman-like kidney tubulopathy due to mitochondrial DNA mutation	ORPHA:620371
ERKNet	Genetic primary hypomagnesemia	ORPHA:34526
ERKNet	Genetic primary hypomagnesemia with hypocalciuria	ORPHA:306519
ERKNet	Genetic primary hypomagnesemia with normocalciuria	ORPHA:306522
ERKNet	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis	ORPHA:306516
ERKNet	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement	ORPHA:2196
ERKNet	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement	ORPHA:31043
ERKNet	Familial primary hypomagnesemia with normocalciuria and normocalcemia	ORPHA:34527
ERKNet	Tubular renal disease-cardiomyopathy syndrome	ORPHA:73224
ERKNet	Primary hypomagnesemia with secondary hypocalcemia	ORPHA:30924
ERKNet	Primary hypomagnesemia with refractory seizures and intellectual disability	ORPHA:564178
ERKNet	Hypertrophic cardiomyopathy and renal tubular disease due to mitochondrial DNA mutation	ORPHA:324525
ERKNet	Bartter syndrome type 1	ORPHA:620217
ERKNet	Bartter syndrome type 2	ORPHA:620220
ERKNet	Antenatal Bartter syndrome	ORPHA:93604
ERKNet	Bartter syndrome type 5	ORPHA:570371
ERKNet	Tubulointerstitial nephritis and uveitis syndrome	ORPHA:91500
ERKNet	Pseudohypoaldosteronism type 2	ORPHA:757
ERKNet	Nephrogenic diabetes insipidus-intracranial calcification-short stature-facial dysmorphism syndrome	ORPHA:3145
ERKNet	Autosomal dominant primary hypomagnesemia with hypocalciuria	ORPHA:34528
ERKNet	Primary hypomagnesemia with secondary hypocalcemia	ORPHA:30924
ERKNet	Isolated autosomal dominant hypomagnesemia, Glaudemans type	ORPHA:199326
ERKNet	Primary hypomagnesemia-refractory seizures-intellectual disability syndrome	ORPHA:564178
ERKNet	Primary hypomagnesemia-generalized seizures-intellectual disability-obesity syndrome	ORPHA:620363
ERKNet	EGF-related primary hypomagnesemia with intellectual disability	ORPHA:620368
ERKNet	Distal renal tubular acidosis	ORPHA:18
ERKNet	Autosomal dominant polycystic kidney disease	ORPHA:730
ERKNet	Senior-Boichis syndrome	ORPHA:84081
ERKNet	Hereditary renal hypouricemia	ORPHA:94088
ERKNet	RHYNS syndrome	ORPHA:140976

ERKNet	Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome	ORPHA:528105
ERKNet	Renal ciliopathy	ORPHA:156162
ERKNet	Adult familial nephronophthisis-spastic quadriparesia syndrome	ORPHA:2666
ERKNet	Autosomal dominant polycystic kidney disease	ORPHA:730
ERKNet	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	ORPHA:88924
ERKNet	MUC1-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88949
ERKNet	UMOD-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88950
ERKNet	Autosomal recessive polycystic kidney disease	ORPHA:731
ERKNet	Joubert syndrome with renal defect	ORPHA:220497
ERKNet	Oncogenic osteomalacia	ORPHA:352540
ERKNet	Tuberous sclerosis complex	ORPHA:805
ERKNet	Von Hippel-Lindau disease	ORPHA:892
ERKNet	Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation	ORPHA:324525
ERKNet	Autosomal dominant polycystic kidney disease	ORPHA:730
ERKNet	Saldino-Mainzer syndrome	ORPHA:140969
ERKNet	IgG4-related kidney disease	ORPHA:449395
ERKNet	Genetic cystic renal disease	ORPHA:93587
ERKNet	Autosomal dominant tubulointerstitial kidney disease	ORPHA:34149
ERKNet	MUC1-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88949
ERKNet	UMOD-related autosomal dominant tubulointerstitial kidney disease	ORPHA:88950
ERKNet	HNF1B-related autosomal dominant tubulointerstitial kidney disease	ORPHA:93111
ERKNet	REN-related autosomal dominant tubulointerstitial kidney disease	ORPHA:217330
ERKNet	Cranioectodermal dysplasia	ORPHA:1515
ERKNet	Joubert syndrome with oculorenal defect	ORPHA:2318
ERKNet	Nephronophthisis	ORPHA:655
ERKNet	Late-onset nephronophthisis	ORPHA:93589
ERKNet	Infantile nephronophthisis	ORPHA:93591
ERKNet	Juvenile nephronophthisis	ORPHA:93592
ERKNet	RHYNS syndrome	ORPHA:140976
ERKNet	Senior-Loken syndrome	ORPHA:3156
ERKNet	Senior-Boichis syndrome	ORPHA:84081
ERKNet	Ellis Van Creveld syndrome	ORPHA:289
ERN CRANIO	Non-syndromic unisutural craniosynostosis	ORPHA:620096
ERN CRANIO	Non-syndromic unicoronal craniosynostosis	ORPHA:620102
ERN CRANIO	Non-syndromic unilambdoid craniosynostosis	ORPHA:620113
ERN CRANIO	Non-syndromic unifrontosphenoidal craniosynostosis	ORPHA:620139
ERN CRANIO	Non-syndromic unisquamosal craniosynostosis	ORPHA:620146
ERN CRANIO	Non-syndromic multisutural craniosynostosis	ORPHA:620152
ERN CRANIO	Non-syndromic non-specific multisutural craniosynostosis	ORPHA:620158
ERN CRANIO	Non-syndromic bilambdoid craniosynostosis	ORPHA:620178
ERN CRANIO	Non-syndromic unicoronal and sagittal craniosynostosis	ORPHA:620186

ERN CRANIO	Non-syndromic metopic and sagittal craniosynostosis	ORPHA:620192
ERN CRANIO	Non-syndromic bicoronal and metopic craniosynostosis	ORPHA:620198
ERN CRANIO	Non-syndromic bicoronal and sagittal craniosynostosis	ORPHA:620205
ERN CRANIO	Non-syndromic pansynostosis	ORPHA:620212
ERN CRANIO	Isolated cloverleaf skull syndrome	ORPHA:2343
ERN CRANIO	Familial lambdoid synostosis	ORPHA:3267
ERN CRANIO	Isolated plagiocephaly	ORPHA:35098
ERN CRANIO	Isolated oxycephaly	ORPHA:63440
ERN CRANIO	Isolated craniosynostosis	ORPHA:139390
ERN CRANIO	Isolated trigonocephaly	ORPHA:3366
ERN CRANIO	Isolated scaphocephaly	ORPHA:35093
ERN CRANIO	Coffin-Siris syndrome	ORPHA:1465
ERN CRANIO	Craniofrontonasal syndrome	ORPHA:1520
ERN CRANIO	KBG syndrome	ORPHA:2332
ERN CRANIO	Dubowitz syndrome	ORPHA:235
ERN CRANIO	Prenatal benign hypophosphatasia	ORPHA:247638
ERN CRANIO	Infantile hypophosphatasia	ORPHA:247651
ERN CRANIO	Childhood-onset hypophosphatasia	ORPHA:247667
ERN CRANIO	Osteoglosphonic dysplasia	ORPHA:2645
ERN CRANIO	3MC syndrome	ORPHA:293843
ERN CRANIO	Radioulnar synostosis-developmental delay-hypotonia syndrome	ORPHA:3270
ERN CRANIO	Acrocallosal syndrome	ORPHA:36
ERN CRANIO	Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	ORPHA:369837
ERN CRANIO	Noonan syndrome	ORPHA:648
ERN CRANIO	Hyaluronidase deficiency	ORPHA:67041
ERN CRANIO	Pycnodysostosis	ORPHA:763
ERN CRANIO	Williams syndrome	ORPHA:904
ERN CRANIO	Koolen-De Vries syndrome	ORPHA:96169
ERN CRANIO	Bohring-Opitz syndrome	ORPHA:97297
ERN CRANIO	Fetal valproate spectrum disorder	ORPHA:1906
ERN CRANIO	Thanatophoric dysplasia	ORPHA:2655
ERN CRANIO	Thanatophoric dysplasia type 1	ORPHA:1860
ERN CRANIO	Thanatophoric dysplasia type 2	ORPHA:93274
ERN CRANIO	Mucopolysaccharidosis	ORPHA:79213
ERN CRANIO	Mucopolysaccharidosis type 1	ORPHA:579
ERN CRANIO	Hurler syndrome	ORPHA:93473
ERN CRANIO	Scheie syndrome	ORPHA:93474
ERN CRANIO	Hurler-Scheie syndrome	ORPHA:93476
ERN CRANIO	Mucopolysaccharidosis type 2	ORPHA:580
ERN CRANIO	Mucopolysaccharidosis type 2, severe form	ORPHA:217085
ERN CRANIO	Mucopolysaccharidosis type 2, attenuated form	ORPHA:217093
ERN CRANIO	Mucopolysaccharidosis type 3	ORPHA:581
ERN CRANIO	Sanfilippo syndrome type A	ORPHA:79269

ERN CRANIO	Sanfilippo syndrome type B	ORPHA:79270
ERN CRANIO	Sanfilippo syndrome type C	ORPHA:79271
ERN CRANIO	Sanfilippo syndrome type D	ORPHA:79272
ERN CRANIO	Mucopolysaccharidosis type 4	ORPHA:582
ERN CRANIO	Mucopolysaccharidosis type 4A	ORPHA:309297
ERN CRANIO	Mucopolysaccharidosis type 4B	ORPHA:309310
ERN CRANIO	Mucopolysaccharidosis type 6	ORPHA:583
ERN CRANIO	Mucopolysaccharidosis type 6, rapidly progressing	ORPHA:276212
ERN CRANIO	Mucopolysaccharidosis type 6, slowly progressing	ORPHA:276223
ERN CRANIO	Mucopolysaccharidosis type 7	ORPHA:584
ERN CRANIO	Crouzon disease	ORPHA:207
ERN CRANIO	Hunter-McAlpine craniosynostosis	ORPHA:97340
ERN CRANIO	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	ORPHA:457193
ERN CRANIO	Pseudoaminopterin syndrome	ORPHA:221120
ERN CRANIO	Metopic ridging-ptosis-facial dysmorphism syndrome	ORPHA:502430
ERN CRANIO	Craniosynostosis-fibular aplasia syndrome	ORPHA:1533
ERN CRANIO	Acrocephalosyndactyly	ORPHA:946
ERN CRANIO	Craniofacial dysplasia-osteopenia syndrome	ORPHA:314555
ERN CRANIO	Aplasia cutis congenita	ORPHA:1114
ERN CRANIO	Adams-Oliver syndrome	ORPHA:974
ERN CRANIO	Craniometaphyseal dysplasia	ORPHA:1522
ERN CRANIO	Frontometaphyseal dysplasia	ORPHA:1826
ERN CRANIO	Osteopathia striata-cranial sclerosis syndrome	ORPHA:2780
ERN CRANIO	Osteocraniostenosis	ORPHA:2763
ERN CRANIO	Cole-Carpenter syndrome	ORPHA:2050
ERN BOND	Acromegaloid facial appearance syndrome	ORPHA:965
ERN BOND	Hypertrichosis-acromegaloid facial appearance syndrome	ORPHA:966
ERN BOND	Frontonasal dysplasia-bifid nose-upper limb anomalies syndrome	ORPHA:521308
ERN BOND	Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome	ORPHA:444077
ERN BOND	Cantú syndrome	ORPHA:1517
ERN BOND	9q33.3q34.11 microdeletion syndrome	ORPHA:495818
ERN BOND	X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome	ORPHA:482606
ERN BOND	Retinitis pigmentosa-hearing loss-premature aging-short stature-facial dysmorphism syndrome	ORPHA:494439
ERN BOND	COG1-CDG	ORPHA:263508
ERN BOND	Camptodactyly-fibrous tissue hyperplasia-skeletal dysplasia syndrome	ORPHA:1321
ERN BOND	Heart-hand syndrome type 3	ORPHA:1342
ERN BOND	Heart-hand syndrome type 2	ORPHA:1350
ERN BOND	Dobrow syndrome	ORPHA:3262
ERN BOND	Imperforate oropharynx-costovertebral anomalies syndrome [Historic]	ORPHA:2759
ERN BOND	Craniorhiny [Historic]	ORPHA:157832

ERN BOND	Camptobrachydactyly [Historic]	ORPHA:1319
ERN BOND	Mononen-Karnes-Senac syndrome [Historic]	ORPHA:2565
ERN BOND	Brachydactyly type A7 [Historic]	ORPHA:93397
ERN BOND	Sugarman brachydactyly [Historic]	ORPHA:498602
ERN BOND	Brachytelephalangy-dysmorphism-Kallmann syndrome [Historic]	ORPHA:1295
ERN BOND	Brachydactyly-long thumb syndrome [Historic]	ORPHA:2946
ERN BOND	Acrodysplasia scoliosis [Historic]	ORPHA:2956
ERN BOND	Banki syndrome [Historic]	ORPHA:1228
ERN BOND	Symphalangism with multiple anomalies of hands and feet [Historic]	ORPHA:3246
ERN BOND	Sillence syndrome [Historic]	ORPHA:3168
ERN BOND	WT limb-blood syndrome [Historic]	ORPHA:3466
ERN BOND	Tetramelic monodactyly [Historic]	ORPHA:2564
ERN BOND	Absent radius-anogenital anomalies syndrome [Historic]	ORPHA:3016
ERN BOND	Absent tibia-polydactyly-arachnoid cyst syndrome [Historic]	ORPHA:3328
ERN BOND	Humerus trochlea aplasia [Historic]	ORPHA:3383
ERN BOND	Radial deficiency-tibial hypoplasia syndrome [Historic]	ORPHA:1121
ERN BOND	Ulnar hypoplasia-split foot syndrome [Historic]	ORPHA:1122
ERN BOND	Charlie M syndrome [Historic]	ORPHA:1406
ERN BOND	Ectrodactyly-polydactyly syndrome [Historic]	ORPHA:1892
ERN BOND	Polydactyly-myopia syndrome [Historic]	ORPHA:2917
ERN BOND	Eyebrow duplication-syndactyly syndrome [Historic]	ORPHA:3172
ERN BOND	Syndactyly-polydactyly-ear lobe syndrome [Historic]	ORPHA:3259
ERN BOND	Camptodactyly-joint contractures-facial skeletal defects syndrome	ORPHA:1323
ERN BOND	Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome	ORPHA:94095
ERN BOND	Spondylocostal dysostosis-hypospadias-intellectual disability syndrome	ORPHA:329252
ERN BOND	Familial chondromalacia patellae [Historic]	ORPHA:1428
ERN BOND	Say-Field-Coldwell syndrome [Historic]	ORPHA:3133
ERN BOND	Eye defects-arachnodactyly-cardiopathy syndrome	ORPHA:2725
ERN BOND	Frontonasal dysplasia-bifid nose-upper limb anomalies syndrome	ORPHA:521308
ERN BOND	Cerebrofaciothoracic dysplasia	ORPHA:1394
ERN BOND	Heart-hand syndrome, Slovenian type	ORPHA:168796
ERN BOND	Fibular dimelia-diplopodia syndrome	ORPHA:1757
ERN BOND	Split hand-split foot-deafness syndrome	ORPHA:71271
ERN BOND	Orofaciodigital syndrome type 6	ORPHA:2754
ERN BOND	Orofaciodigital syndrome type 5	ORPHA:2919
ERN BOND	Orofaciodigital syndrome type 9	ORPHA:141007
ERN BOND	Oral-facial-digital syndrome with short stature and brachymesophalangy	ORPHA:508501
ERN BOND	Endocrine-cerebro-osteodysplasia syndrome	ORPHA:199332
ERN BOND	Proximal femoral focal deficiency	ORPHA:633228
ERN BOND	Acrodysostosis with multiple hormone ressitance	ORPHA:280651
ERN BOND	Peripheral dysostosis [Historic]	ORPHA:1795
ERN BOND	Hyaluronidase deficiency	ORPHA:67041
ERN BOND	Leukoencephalopathy-spondylometaphyseal dysplasia syndrome	ORPHA:83629

ERN BOND	Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome	ORPHA:369837
ERN BOND	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	ORPHA:52430
ERN BOND	Spastic paraplegia-Paget disease of bone syndrome	ORPHA:329475
ERN BOND	Dominant hypophosphatemia with nephrolithiasis or osteoporosis	ORPHA:244305
ERN BOND	Pancreatic insufficiency-anemia-hyperostosis syndrome	ORPHA:199337
ERN BOND	Classical-like Ehlers-Danlos syndrome type 2	ORPHA:536532
ERN BOND	MIR140-related spondyloepiphyseal dysplasia	ORPHA:623695
ERN BOND	Early-onset calcifying leukoencephalopathy-skeletal dysplasia	ORPHA:556985
ERN BOND	SBDS-related severe neonatal spondylometaphyseal dysplasia	ORPHA:622934
ERN BOND	Spondylometaphyseal dysplasia-corneal dystrophy syndrome	ORPHA:589435
ERN BOND	Short stature-skeletal dysplasia-retinal degeneration-intellectual disability-sensorineural hearing loss syndrome	ORPHA:589442
ERN BOND	Luscan-Lumish syndrome	ORPHA:597738
ERN BOND	X-linked severe syndromic thoracic aortic aneurysm and dissection	ORPHA:622925
ERN BOND	Oligosyndactyly, radio-ulnar synostosis, hearing loss and renal defects syndrome	_
ERN BOND	Tall stature with long halluces, NPR3 type	_
ERN BOND	X-linked osteogenesis imperfecta	_
ERN BOND	Preaxial brachydactyly, PAX3 type	ORPHA:633211
ERN BOND	Cole-Carpenter syndrome	ORPHA:2050
ERN BOND	Dysplastic cortical hyperostosis	ORPHA:2204
ERN BOND	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467
ERN BOND	Spondyloepiphyseal dysplasia, Reardon type [Historic]	ORPHA:163662
ERN BOND	Rhizomelic syndrome, Urbach type [Historic]	ORPHA:3098
ERN BOND	Autosomal recessive distal osteolysis syndrome [Historic]	ORPHA:2776
ERN BOND	Thoracomelic dysplasia [Historic]	ORPHA:1803
ERN BOND	Cleidorhizomelic syndrome [Historic]	ORPHA:1453
ERN BOND	X-linked calvarial hyperostosis [Historic]	ORPHA:391327
ERN BOND	Osteoporosis-oculocutaneous hypopigmentation syndrome	ORPHA:2786
ERN BOND	Rhizomelic dysplasia, Patterson-Lowry type	ORPHA:2831
ERN BOND	Otopalatodigital syndrome type 2	ORPHA:90652
ERN BOND	Nestor-Guillermo progeria syndrome	ORPHA:280576
ERN BOND	Osteosclerosis-ichthyosis-premature ovarian failure syndrome	ORPHA:75325
ERN BOND	Hyperostosis cranialis interna	ORPHA:443098
ERN BOND	Ehlers-Danlos/osteogenesis imperfecta syndrome	ORPHA:230857
ERN BOND	Regressive spondylometaphyseal dysplasia	ORPHA:448267
ERN BOND	Metaphyseal dysostosis-intellectual disability-conductive deafness syndrome [Historic]	ORPHA:2502
ERN BOND	Metaphyseal chondrodysplasia, Kaitila type [Historic]	ORPHA:166038
ERN BOND	Spondylometaphyseal dysplasia, Golden type [Historic]	ORPHA:168544
ERN BOND	12q14 microdeletion syndrome	ORPHA:94063
ERN BOND	Short stature-brachydactyly-obesity-global developmental delay syndrome	ORPHA:464288
ERN BOND	Thoracic dysplasia-hydrocephalus syndrome [Historic]	ORPHA:1861

ERN BOND	Leukoencephalopathy-spondylometaphyseal dysplasia syndrome	ORPHA:83629
ERN BOND	Spondyloepiphyseal dysplasia, Cantu type	ORPHA:163654
ERN BOND	Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome	ORPHA:477831
ERN BOND	Pacman dysplasia	ORPHA:1952
ERN BOND	Hypoplastic tibiae-postaxial polydactyly syndrome	ORPHA:3332
ERN BOND	Triphalangeal thumb-polysyndactyly syndrome	ORPHA:2950
ERN BOND	Thin ribs-tubular bones-dysmorphism syndrome	ORPHA:1506
ERN BOND	Congenital bowing of long bones	ORPHA:2292
ERN BOND	Mesomelic dysplasia-digital anomalies-intellectual disability syndrome	ORPHA:632603
ERN BOND	Severe lateral tibial bowing-short stature-mild winged scapula-mild facial dysmorphism syndrome	ORPHA:324307
ERN BOND	Buschke-Ollendorff syndrome	ORPHA:1306
ERN BOND	Spondyloepimetaphyseal dysplasia with joint laxity	ORPHA:93359
ERN BOND	Spondyloepimetaphyseal dysplasia with joint laxity, EXOC6B type	ORPHA:642085
ERN BOND	Spondyloepimetaphyseal dysplasia with joint laxity, Beighton type	ORPHA:642099
ERN BOND	Spondyloepimetaphyseal dysplasia, Isidor type	ORPHA:370015
ERN BOND	Atypical femur fracture	_
ERN BOND	Mandibuloacral dysplasia	ORPHA:2457
ERN BOND	Spondylometaphyseal dysplasia-bowed forearms-facial dysmorphism syndrome	ORPHA:168552
ERN BOND	Spondyloepimetaphyseal dysplasia, A4 type	ORPHA:168555
ERN BOND	Spondylometaphyseal dysplasia, Czarny-Ratajczak type	ORPHA:370019
ERN BOND	Mutliple epihyseal dysplasia, Al-Gazali type	ORPHA:166024
ERN BOND	Multiple epiphyseal dysplasia, Beighton type	ORPHA:166011
ERN BOND	Multiple epiphyseal dysplasia, Lowry type	ORPHA:166016
ERN BOND	Multiple epiphyseal dysplasia, Al-Gazali type	ORPHA:166024

List of expert networks reviewing Orphanet abstracts/ definitions in 2022

Name of expert network	Disease name	ORPHAcode
Endo-ERN	45,X/46,XY mixed gonadal dysgenesis	ORPHA:1772
Endo-ERN	46,XX gonadal dysgenesis	ORPHA:243
Endo-ERN	46,XY complete gonadal dysgenesis	ORPHA:242
Endo-ERN	46,XY partial gonadal dysgenesis	ORPHA:251510
Endo-ERN	Autosomal dominant hypophosphatemic rickets	ORPHA:89937
Endo-ERN	Autosomal recessive hypophosphatemic rickets	ORPHA:289176
Endo-ERN	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	ORPHA:90794
Endo-ERN	Congenital adrenal hyperplasia	ORPHA:418

Endo-ERN	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	ORPHA:90795
Endo-ERN	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	ORPHA:90793
Endo-ERN	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	ORPHA:95699
Endo-ERN	Congenital lipoid adrenal hyperplasia due to STAR deficency	ORPHA:90790
Endo-ERN	Hereditary hypophosphatemic rickets with hypercalciuria	ORPHA:157215
Endo-ERN	Hypocalcemic vitamin D-dependent rickets	ORPHA:289157
Endo-ERN	Hypocalcemic vitamin D-resistant rickets	ORPHA:93160
Endo-ERN	X-linked hypophosphatemia	ORPHA:89936
EpiCARE	Infantile spasms syndrome	ORPHA:3451
ERKNet	Autosomal dominant distal renal tubular acidosis	ORPHA:93608
ERKNet	Autosomal dominant proximal renal tubular acidosis	ORPHA:314889
ERKNet	Autosomal recessive distal renal tubular acidosis	ORPHA:402041
ERKNet	Autosomal recessive proximal renal tubular acidosis	ORPHA:93607
ERKNet	Dent disease	ORPHA:1652
ERKNet	Distal renal tubular acidosis with anemia	ORPHA:93610
ERKNet	Idiopathic steroid-resistant nephrotic syndrome	ORPHA:567548
ERKNet	Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	ORPHA:567546
ERKNet	Pauci-immune glomerulonephritis with ANCA	ORPHA:97563
ERKNet	Renal dysplasia, unilateral	ORPHA:93172
ERN CRANIO	Recurrent respiratory papillomatosis	ORPHA:60032
ERN GENTURIS	Familial colorectal cancer Type X	ORPHA:440437
ERNICA	Congenital esophageal diverticulum	ORPHA:91358
ERNICA	Esophageal atresia	ORPHA:1199
ERNICA	Esophageal duplication cyst	ORPHA:100047
ERNICA	Tubular duplication of the esophagus	ORPHA:100048
ERN-LUNG	Cryptogenic organizing pneumonia	ORPHA:1302
ERN-LUNG	Lymphangioleiomyomatosis	ORPHA:538
ERN-Skin	Acral peeling skin syndrome	ORPHA:263534
ERN-Skin	Acute generalized exanthematous pustulosis	ORPHA:293173
ERN-Skin	Autosomal dominant epidermolytic ichthyosis	ORPHA:312
ERN-Skin	CHILD syndrome	ORPHA:139
ERN-Skin	Congenital ichthyosiform erythroderma	ORPHA:79394
ERN-Skin	Drug reaction with eosinophilia and systemic symptoms	ORPHA:139402
ERN-Skin	Erythema multiforme major	ORPHA:502499
ERN-Skin	Fixed drug eruption	ORPHA:293812
ERN-Skin	Generalized peeling skin syndrome	ORPHA:263543
ERN-Skin	Harlequin ichthyosis	ORPHA:457
ERN-Skin	Lamellar ichthyosis	ORPHA:313
ERN-Skin	Mal de Meleda	ORPHA:87503
ERN-Skin	Pachyonychia congenita	ORPHA:2309
ERN-Skin	Peeling skin syndrome	ORPHA:817
ERN-Skin	Peeling skin syndrome type A	ORPHA:263548

ERN-Skin	Peeling skin syndrome type B	ORPHA:263553
ERN-Skin	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	ORPHA:444138
ERN-Skin	Punctate palmoplantar keratoderma type 1	ORPHA:79501
ERN-Skin	Stevens-Johnson syndrome/toxic epidermal necrolysis overlap syndrome	ORPHA:506784
ERN-Skin	Stevens-Johnson syndrome/toxic epidermal necrolysis spectrum	ORPHA:95455
ERN-Skin	Toxic epidermal necrolysis	ORPHA:537
ERN-Skin	Trichothiodystrophy	ORPHA:33364
EURACAN	Complete hydatidiform mole	ORPHA:254688
EURACAN	Epithelioid trophoblastic tumor	ORPHA:254698
EURACAN	Gestational choriocarcinoma	ORPHA:99926
EURACAN	Gestational trophoblastic neoplasm	ORPHA:59305
EURACAN	Invasive mole	ORPHA:99925
EURACAN	Partial hydatidiform mole	ORPHA:254693
EURACAN	Placental site trophoblastic tumor	ORPHA:99928
EuroBloodNet	Bleeding disorder in hemophilia A carriers	ORPHA:177926
EuroBloodNet	Bleeding disorder in hemophilia B carriers	ORPHA:177929
EuroBloodNet	Hemophilia	ORPHA:448
EuroBloodNet	Hemophilia A	ORPHA:98878
EuroBloodNet	Hemophilia B	ORPHA:98879
EuroBloodNet	Mild hemophilia A	ORPHA:169808
EuroBloodNet	Mild hemophilia B	ORPHA:169799
EuroBloodNet	Moderate hemophilia B	ORPHA:169796
EuroBloodNet	Severe hemophilia A	ORPHA:169802
EuroBloodNet	Severe hemophilia B	ORPHA:169793
eUROGEN	Caudal regression syndrome	ORPHA:3027
eUROGEN	Caudal regression-sirenomelia spectrum	ORPHA:444941
eUROGEN	Familial caudal dysgenesis	ORPHA:1768
eUROGEN	Ochoa syndrome	ORPHA:2704
eUROGEN	Sirenomelia	ORPHA:3169
EURO-NMD	Congenital muscular dystrophy, Fukuyama type	ORPHA:272
EURO-NMD	MERRF	ORPHA:551
EURO-NMD	Muscle-eye-brain disease	ORPHA:588
EURO-NMD	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	ORPHA:370997
EURO-NMD	Walker-Warburg syndrome	ORPHA:899
ITHACA	BOR syndrome	ORPHA:107
ITHACA	Caudal regression-sirenomelia spectrum	ORPHA:444941
ITHACA	Familial caudal dysgenesis	ORPHA:1768
ITHACA	Isolated childhood apraxia of speech	ORPHA:209908
ITHACA	KLHL7-related Bohring-Opitz-like and Crisponi/Cold-induced sweating-like overlap syndrome	ORPHA:603684
ITHACA	KLHL7-related Bohring-Opitz-like syndrome	ORPHA:603689
ITHACA	KLHL7-related Crisponi/cold-induced sweating-like syndrome	ORPHA:603694
ITHACA	Ochoa syndrome	ORPHA:2704

ITHACA	Recessive KLHL7-related disorder	ORPHA:603699
ITHACA	Sirenomelia	ORPHA:3169
MetabERN	Autosomal dominant hyperinsulinism due to SUR1 deficiency	ORPHA:276575
MetabERN	Congenital isolated hyperinsulinism	ORPHA:657
MetabERN	Diazoxide-resistant focal hyperinsulinism	ORPHA:79298
MetabERN	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	ORPHA:276598
MetabERN	Diazoxide-resistant hyperinsulinism	ORPHA:276585
MetabERN	Exercise-induced hyperinsulinism	ORPHA:165991
MetabERN	Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency	ORPHA:71212
MetabERN	Hyperinsulinism due to UCP2 deficiency	ORPHA:276556

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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