Orphanet Report Series

Rare Diseases collection

2023

Expert reviewers for Orphanet in 2023

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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 name of the disease for which they have contributed 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 2023

Name of expert	Name of disease	ORPHA code
Dr David R. ADAMS	Free sialic acid storage disease	ORPHA:834
Dr Grazyna ADAMUS	Cancer-associated retinopathy	ORPHA:71505
Dr Laura A. ADANG	Multiple sulfatase deficiency	ORPHA:585
Petra AGEN	Segmental spinal dysgenesis	ORPHA:656126
Dr Rebecca C. AHRENS-NICKLAS	Multiple sulfatase deficiency	ORPHA:585
Dr Kohlschuetter Alfried	Neuronal ceroid lipofuscinosis	ORPHA:216
Pr Laurence AMAR	Pheochromocytoma-paraganglioma	ORPHA:573163
Pr Marc ANDRÉ	Corticosteroid-sensitive aseptic abscess syndrome	ORPHA:54251
Abiri ARASH	Silent sinus syndrome	ORPHA:71276
Pr Stefan ARETZ	Lynch syndrome	ORPHA:144
Dr Jean-Baptiste ARNOUX	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Pr Olivier AUMAITRE	Corticosteroid-sensitive aseptic abscess syndrome	ORPHA:54251
Dr Gilles BAGOU	Cryoglobulinemic vasculitis	ORPHA:91138
	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
	Pheochromocytoma-paraganglioma	ORPHA:573163
Dr Karthik BALAKRISHNAN	Tracheal agenesis	ORPHA:3346
Pr Bettina BALINT	Stiff person spectrum disorder	ORPHA:3198
Dr Judith BALMAÑA	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Dr Eulalia BASELGA TORRES	Autosomal dominant epidermolytic ichthyosis	ORPHA:312
Dr Nicole BAUMANN	Tay-Sachs disease	ORPHA:845
Dr Christine BELLANNE-CHANTELOT	MODY	ORPHA:552
Dr Jean-François BENOIST	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Pr Jérôme BERTHERAT	OBSOLETE: Cushing syndrome	ORPHA:553
	OBSOLETE: ACTH-independent Cushing syndrome	ORPHA:99893
	Adrenal/paraganglial tumor	ORPHA:100091
	Cushing syndrome due to bilateral macronodular adrenocortical disease	ORPHA:189427
	OBSOLETE: Primary pigmented nodular adrenocortical disease	ORPHA:189439
	Rare disease with adrenal Cushing syndrome as a major feature	ORPHA:314749
	NON RARE IN EUROPE: Cortisol-producing adrenal tumor	ORPHA:423668
	OBSOLETE: ACTH-independent Cushing syndrome due to rare cortisol-producing adrenal tumor	ORPHA:443287
	Endogenous Cushing syndrome	ORPHA:641613
	OBSOLETE: Adrenal Cushing syndrome due to isolated adrenocortical benign tumor	ORPHA:642013
	Cushing syndrome due to cortisol-producing adrenocortical adenoma	ORPHA:642788
	Adrenal Cushing syndrome	ORPHA:647758

	Rare adrenocortical nodular disease with Cushing syndrome as a major feature	ORPHA:647768
	Isolated primary pigmented nodular adrenocortical disease	ORPHA:647772
	Isolated micronodular adrenocortical disease	ORPHA:647782
	Rare adrenocortical nodular disease	ORPHA:649017
Pr Enrico S. BERTINI	Congenital muscular dystrophy, Fukuyama type	ORPHA:272
	Walker-Warburg syndrome	ORPHA:899
	Muscle-eye-brain disease	ORPHA:588
	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	ORPHA:370997
Pr Regina C. BETZ	Carvajal syndrome	ORPHA:65282
Dr Romain J. BLAIZOT	Furuncular myiasis	ORPHA:591
Pr Agnès BLOCH-ZUPAN	Segmental odontomaxillary dysplasia	ORPHA:67039
	Regional odontodysplasia	ORPHA:83450
Pr Daniel BOEHRINGER	Rare scleritis	ORPHA:648559
	Infectious scleritis	ORPHA:648665
	Idiopathic scleritis	ORPHA:648675
	Immune-mediated scleritis	ORPHA:648681
Dr Petra BOGOVIC	Tick-borne encephalitis	ORPHA:297
Dr Carsten G. BONNEMANN	Bethlem muscular dystrophy	ORPHA:610
	Ullrich congenital muscular dystrophy	ORPHA:75840
Dr Juliette BOUCHEREAU	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
	Homocystinuria due to cystathionine beta-synthase deficiency	ORPHA:394
Dr Jean-Baptiste BOUILLON-MINOIS	Cryoglobulinemic vasculitis	ORPHA:91138
Dr Simon BOUSSION	Thrombocytopenia-absent radius syndrome	ORPHA:3320
Pr Dominique BREMOND-GIGNAC	WAGR syndrome	ORPHA:893
Pr Patrice CACOUB	Cryoglobulinemic vasculitis	ORPHA:91138
Pr Bert CALLEWAERT	Congenital contractural arachnodactyly	ORPHA:115
Dr Alessandra CAMERINI	Classic glucose transporter type 1 deficiency syndrome	ORPHA:71277
Dr Dídac CASAS-ALBA	Marinesco-Sjögren syndrome	ORPHA:559
Pr Marco CASTAGNETTI	Congenital bilateral absence of vas deferens	ORPHA:48
	Congenital primary megaureter	ORPHA:617
Pr Teodora CHAMOVA	Congenital cataracts-facial dysmorphism-neuropathy syndrome	ORPHA:48431
Pr Philippe CHANSON	Non-functioning pituitary adenoma	ORPHA:91349
Dr Anthony CHAUVIN	Cryoglobulinemic vasculitis	ORPHA:91138
	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
	Pheochromocytoma-paraganglioma	ORPHA:573163
Dr Elzbieta CIARA	Fucosidosis	ORPHA:349
Dr Benjamin T. COCANOUGHER	Glycogen storage disease due to phosphoglycerate mutase deficiency	ORPHA:97234
Dr John COLEMAN	Currarino syndrome	ORPHA:1552
Pr Vincent COTTIN	Hypersensitivity pneumonitis	ORPHA:31740
	Acute interstitial pneumonia	ORPHA:79126

Dr Célia CRETOLLE	NON RARE IN EUROPE: Horseshoe kidney	ORPHA:3029
	Isolated persistent urogenital sinus	ORPHA:647794
	Non-syndromic supernumerary kidneys	ORPHA:652528
Pr Julie DE BACKER	Marfan syndrome	ORPHA:558
	Loeys-Dietz syndrome	ORPHA:60030
	Marfan syndrome and Marfan-related disorders	ORPHA:284993
Dr Valentina DE GIORGIS GIORGIS	Classic glucose transporter type 1 deficiency syndrome	ORPHA:71277
Dr Corinne DE LAET	Tyrosinemia type 3	ORPHA:69723
	Tyrosinemia type 1	ORPHA:882
	Tyrosinemia type 2	ORPHA:28378
	Transient tyrosinemia of the newborn	ORPHA:3402
Pr Pascale DE LONLAY	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Dr Elisa DEFLORENNE	Pheochromocytoma-paraganglioma	ORPHA:573163
Pr F.G. [Frederik] DIKKERS	Recurrent respiratory papillomatosis	ORPHA:60032
Dr Jean DONADIEU	Langerhans cell histiocytosis	ORPHA:389
Dr Bruno DONADILLE	NON RARE IN EUROPE: Central precocious puberty	ORPHA:759
Pr Douglas LEVINSON	OBSOLETE: Early-onset schizophrenia	ORPHA:96369
	Childhood-onset schizophrenia	ORPHA:641496
Dr Danièle DUBOIS-LAFORGUE	MODY	ORPHA:552
Pr Jean-Michel DUPONT	Partial autosomal duplication/triplication	ORPHA:98132
Pr Ala EL-GHONEIMI	Renal or urinary tract malformation	ORPHA:93545
	Isolated persistent urogenital sinus	ORPHA:647794
	Non-syndromic supernumerary kidneys	ORPHA:652528
	NON RARE IN EUROPE: Horseshoe kidney	ORPHA:3029
Dr Heather C. ETCHEVERS	Large congenital melanocytic nevus	ORPHA:626
Dr Florence FELLMANN	Chromosome Y microdeletion	ORPHA:1646
	Isochromosomy Yq	ORPHA:98798
	Isochromosomy Yp	ORPHA:98797
	Ring chromosome Y syndrome	ORPHA:261529
Pr Pierre FENAUX	Chronic myelomonocytic leukemia	ORPHA:98823
	Myelodysplastic syndrome	ORPHA:52688
Dr Claire FIESCHI	Hyper-IgE syndrome	ORPHA:331223
	Autosomal recessive hyper-IgE syndrome due to ZNF341 deficiency	ORPHA:641368
Pr Judith FISCHER	Mal de Meleda	ORPHA:87503
	Neutral lipid storage disease with ichthyosis	ORPHA:98907
	Congenital ichthyosiform erythroderma	ORPHA:79394
Pr Jürgen FLOEGE	Immunoglobulin A nephropathy	ORPHA:34145
	Familial isolated hypoparathyroidism	ORPHA:2238
	Autosomal dominant hypocalcemia	ORPHA:428
	Familial isolated hypoparathyroidism due to impaired PTH secretion	ORPHA:189466

	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	ORPHA:2239
Dr A. Reghan FOLEY	Bethlem muscular dystrophy	ORPHA:610
	Ullrich congenital muscular dystrophy	ORPHA:75840
Dr Massimo FUSCONI	Kikuchi-Fujimoto disease	ORPHA:50918
Dr Robert GARNIER	Fetal methylmercury syndrome	ORPHA:1917
Pr David GENEVIEVE	CDK13-related congenital heart defects-intellectual disability-facial dysmorphism syndrome	ORPHA:646278
	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	ORPHA:329224
Dr Christos GEORGIN-LAVIALLE	Periodic fever-immunodeficiency-thrombocytopenia syndrome	ORPHA:652522
Dr Cédric GIL-JARDINÉ	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Dr Alice GOLDENBERG	MYT1L-related developmental delay-intellectual disability-obesity syndrome	ORPHA:647800
Pr Frédéric GOTTRAND	Esophageal atresia	ORPHA:1199
	Congenital esophageal stenosis	ORPHA:645749
Pr Frédéric GRENOUILLET	Cystic echinococcosis	ORPHA:400
Pr Sarah C. GRÜNERT	Pearson syndrome	ORPHA:699
Dr Jérémy GUENEZAN	Pheochromocytoma-paraganglioma	ORPHA:573163
Dr Anne-Marie GUERROT	MYT1L-related developmental delay-intellectual disability-obesity syndrome	ORPHA:647799
Dr Maria K. HAANPÄÄ	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Dr S.E. [Sabine] HANNEMA	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:752
	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:752
Dr Robert A. HEGELE	CCDC115-CDG	ORPHA:468684
	Homozygous familial hypercholesterolemia	ORPHA:391665
	Familial chylomicronemia syndrome	ORPHA:444490
	Hyperlipidemia due to hepatic triacylglycerol lipase deficiency	ORPHA:140905
	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	ORPHA:209902
	TMEM199-CDG	ORPHA:466703
Pr Raoul C. HENNEKAM	Trichorhinophalangeal syndrome type 2	ORPHA:502
	Trichorhinophalangeal syndrome type 1	ORPHA:77258
Dr Bénédicte HERON	Tay-Sachs disease	ORPHA:845
	Sandhoff disease	ORPHA:796
	Tay-Sachs disease	ORPHA:845
Pr Olaf HIORT	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:752
	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:752
	46,XY difference of sex development due to isolated 17,20-lyase deficiency	ORPHA:90796
Dr Marjan HUIZING	Free sialic acid storage disease	ORPHA:834

Dr Tamara HUSSONG MILAGRE	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Pr Arvids IRMEJS	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Pr Aleksandra JEZELA-STANEK	Fucosidosis	ORPHA:349
Dr Nathalie JONCA	Peeling skin syndrome type B	ORPHA:263553
	Acral peeling skin syndrome	ORPHA:263534
	Peeling skin syndrome type A	ORPHA:263548
Pr Guillaume JONDEAU	Marfan syndrome	ORPHA:558
	Loeys-Dietz syndrome	ORPHA:60031
	Marfan syndrome and Marfan-related disorders	ORPHA:284994
	Marfanoid habitus-facial dysmorphism-skeletal abnormality-heart defect syndrome	ORPHA:643503
Manasadevi KARTHIKEYAN	Proteus-like syndrome	ORPHA:2969
	Lhermitte-Duclos disease	ORPHA:65285
	Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome	ORPHA:137608
Pr François KERBAUL	Cryoglobulinemic vasculitis	ORPHA:91138
	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
	Pheochromocytoma-paraganglioma	ORPHA:573163
Dr Priya S. KISHNANI	Glycogen storage disease due to phosphoglycerate mutase deficiency	ORPHA:97234
Dr Steven KNAFO	Syringomyelia	ORPHA:3280
Dr Rebecca L. KOCH	Glycogen storage disease due to phosphoglycerate mutase deficiency	ORPHA:97234
Pr Michihiro KONO	Dyschromatosis symmetrica hereditaria	ORPHA:41
Dr David KOOLEN	Jansen-de Vries syndrome	ORPHA:653767
Dr Mateja KRAJC	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Dr Edward C. KUAN	Silent sinus syndrome	ORPHA:71276
Dr Satoshi KUWABARA	Miller Fisher syndrome	ORPHA:98919
	Bickerstaff brainstem encephalitis	ORPHA:79138
Dr Magali LABADIE	Fetal methylmercury syndrome	ORPHA:1917
Pr Philippe LABRUNE	Glycogen storage disease due to glucose-6-phosphatase deficiency	ORPHA:364
Dr Svetlana Bajalica LAGERCRANTZ	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Dr Minna J. LAINE	Aspartylglucosaminuria	ORPHA:93
Dr Marc LAMBERT	Buerger disease	ORPHA:36258
Dr Claudine LAURENT-LEVINSON	Childhood-onset schizophrenia	ORPHA:641496
	OBSOLETE: Early-onset schizophrenia	ORPHA:96369
Dr Jessica LE GALL	Ataxia-telangiectasia	ORPHA:100
Pr Nicolas LEBOULANGER	Congenital laryngeal web	ORPHA:2374
Dr Guillaume LEFEVRE	Immunodeficiency due to selective anti-polysaccharide antibody deficiency	ORPHA:70593
Pr Juliane LEGER	Genetic precocious puberty	ORPHA:435554
	Central precocious puberty in male	ORPHA:649929

	Rare central precocious puberty	ORPHA:650063
	Rare central precocious puberty in female	ORPHA:650070
	Genetic central precocious puberty in female	ORPHA:650077
	Secondary central precocious puberty in female	ORPHA:650082
	Primary central precocious puberty in male	ORPHA:650087
	Secondary central precocious puberty in male	ORPHA:650092
	Genetic central precocious puberty in male	ORPHA:650097
	Non-genetic central precocious puberty in male	ORPHA:650102
	Genetic central precocious puberty	ORPHA:650182
	Rare peripheral precocious puberty in female	ORPHA:650187
	Rare precocious puberty	ORPHA:95708
Dr Christophe LEROY	Cryoglobulinemic vasculitis	ORPHA:91138
	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
	Pheochromocytoma-paraganglioma	ORPHA:573163
Pr Douglas LEVINSON	OBSOLETE: Early-onset schizophrenia	ORPHA:96369
	Childhood-onset schizophrenia	ORPHA:641496
Dr James LESPINASSE	Chromosome Y microdeletion	ORPHA:1646
	Isochromosomy Yp	ORPHA:98797
	Isochromosomy Yq	ORPHA:98798
	Ring chromosome Y syndrome	ORPHA:261529
Dr Max LIEBAU	PBX1-related congenital anomalies of kidney and urinary tract syndrome	ORPHA:656130
Dr Roman LUSCAN	Congenital laryngeal web	ORPHA:2374
Pr Sally Ann LYNCH	Currarino syndrome	ORPHA:1552
Dr S.M. [Saskia] MAAS	Trichorhinophalangeal syndrome type 2	ORPHA:502
	Trichorhinophalangeal syndrome type 1	ORPHA:77258
Pr Eamonn R. MAHER	Von Hippel-Lindau disease	ORPHA:892
Dr Nizar MAHLAOUI	Ataxia-telangiectasia	ORPHA:100
Dr Hélène MAILLARD	NLRP3-associated autoinflammatory disease	ORPHA:208650
Dr Coralie MALLEBRANCHE	X-linked combined immunodeficiency due to SASH3 deficiency	ORPHA:653751
Pr Michelangelo MANCUSO	MERRF	ORPHA:551
Pr Martin G. MARTIN	Glucose-galactose malabsorption	ORPHA:35710
Pr Anibh MARTIN DAS	Congenital sucrase-isomaltase deficiency	ORPHA:35122
Pr Irene MATHIJSSEN	Craniosynostosis-facial dysmorphism-Chiari-1 malformation-developmental and language delay syndrome	ORPHA:647681
Pr Juliette MAZEREEUW-HAUTIER	Peeling skin syndrome type A	ORPHA:263548
	Peeling skin syndrome type B	ORPHA:263553
	Acral peeling skin syndrome	ORPHA:263534
	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	ORPHA:444138
Dr Francine MECKERT	Cryoglobulinemic vasculitis	ORPHA:91138
	Pheochromocytoma-paraganglioma	ORPHA:573163
	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Pr Bruno MÉGARBANE	Acute ackee fruit intoxication	ORPHA:73423

Dr Marie-France MERCIER	Cryoglobulinemic vasculitis	ORPHA:91138
	Disorder of urea cycle metabolism and ammonia detoxification	ORPHA:79167
Pr Giampaolo MERLINI	Amyloidosis	ORPHA:69
	AApoAIV amyloidosis	ORPHA:439232
Pr Konstantin MILLER	Partial autosomal duplication/triplication	ORPHA:98132
Dr Charline MIOT	X-linked combined immunodeficiency due to SASH3 deficiency	ORPHA:653751
Dr Giovanni MONTINI	PBX1-related congenital anomalies of kidney and urinary tract syndrome	ORPHA:656130
Dr Silvia MORAR	Syringomyelia	ORPHA:3280
Dr Nicole MORICHON-DELVALLEZ	2q32q33 microdeletion syndrome	ORPHA:251019
Dr Francesca MORO	X-linked lissencephaly with abnormal genitalia	ORPHA:452
Dr Giovanni MOSIELLO	Congenital bilateral absence of vas deferens	ORPHA:48
	Congenital primary megaureter	ORPHA:617
Pr Hermann L. MÜLLER	Craniopharyngioma	ORPHA:54595
Dr Yann NADJAR	Tay-Sachs disease	ORPHA:845
	Sandhoff disease	ORPHA:796
	Tay-Sachs disease	ORPHA:845
Pr Nadia NATHAN	Non-syndromic bridging bronchus	ORPHA:648992
	Non-syndromic congenital bronchial atresia	ORPHA:649010
	Bronchial malformation	ORPHA:649014
	Isolated left bronchial isomerism	ORPHA:649029
Dr Joanne NGEOW	Proteus-like syndrome	ORPHA:2969
	Lhermitte-Duclos disease	ORPHA:65285
	Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome	ORPHA:137608
Dr Eithne M. NIC AN RÍOGH	Drug-induced vasculitis	ORPHA:251325
Pr Marc NICOLINO	MODY	ORPHA:552
Dr Laura OBICI	ATTRV122I amyloidosis	ORPHA:85451
	Hereditary ATTR amyloidosis	ORPHA:271861
	ATTRV30M amyloidosis	ORPHA:85447
Pr Sylvie ODENT	Rhombencephalosynapsis	ORPHA:59315
Pr Francesc PALAU	Marinesco-Sjögren syndrome	ORPHA:559
Dr Elena PEDRINI	Autosomal recessive malignant osteopetrosis	ORPHA:667
Pr Isabelle PELLIER	X-linked combined immunodeficiency due to SASH3 deficiency	ORPHA:653751
Dr Naveen PEMMARAJU	Blastic plasmacytoid dendritic cell neoplasm	ORPHA:86870
Pr Luca PERSANI	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:752
Pr Florence PETIT	Thrombocytopenia-absent radius syndrome	ORPHA:3320
Dr Alex PETZOLD	Single isolated optic neuritis	ORPHA:659626
	Relapsing isolated optic neuritis	ORPHA:659634
	Chronic relapsing inflammatory optic neuritis	ORPHA:499085
	Isolated optic neuritis	ORPHA:499096
Pr Capucine PICARD	ICF syndrome	ORPHA:2268

Pr Aldona PIETRZAK	CHILD syndrome	ORPHA:139
	Diffuse palmoplantar keratoderma, Bothnian type	ORPHA:2337
Dr Marta PINEDA RÍU	Lynch syndrome	ORPHA:144
Pr Alberto PIPERNO	Hereditary hyperferritinemia-cataract syndrome	ORPHA:163
Pr Roser PONS	Classic glucose transporter type 1 deficiency syndrome	ORPHA:71277
Pr Graça PORTO	TFR2-related hemochromatosis	ORPHA:225123
	Ferroportin disease	ORPHA:648562
	Non-HFE-related hemochromatosis	ORPHA:648569
	SLC40A1-related hemochromatosis	ORPHA:647834
	HJV or HAMP-related hemochromatosis	ORPHA:79230
	Symptomatic form of HFE-related hemochromatosis	ORPHA:465508
Dr Blandine PREVOST	Non-syndromic bridging bronchus	ORPHA:648992
	Non-syndromic congenital bronchial atresia	ORPHA:649010
	Bronchial malformation	ORPHA:649014
	Isolated left bronchial isomerism	ORPHA:649029
Pr Ana PRIEGO	Immunotherapy induced hypophysitis	ORPHA:641350
Dr Alexandros PROTONOTARIOS	Naxos disease	ORPHA:34217
Dr Hervé PUY	Autosomal erythropoietic protoporphyria	ORPHA:79278
Pr Susana QUIJANO-ROY	Walker-Warburg syndrome	ORPHA:899
	Muscle-eye-brain disease	ORPHA:588
	Congenital muscular dystrophy, Fukuyama type	ORPHA:272
	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	ORPHA:370997
Dr Marie RAFFIN	Idiopathic catatonia	ORPHA:648919
Pr Shamima RAHMAN	Leigh syndrome	ORPHA:506
Dr Dabor RÉSIÈRE	Acute ackee fruit intoxication	ORPHA:73423
Dr Frédéric RIEUX LACAUT	Early-onset immune dysregulation with autoimmunity due to DOCK11 partial deficiency	ORPHA:658946
	Early-onset immune dysregulation due to DOCK11 complete deficiency	ORPHA:658951
Dr Mayka SÁNCHEZ FERNÁNDEZ	Congenital dyserythropoietic anemia type III	ORPHA:98870
Dr Luca SANGIORGI	Ollier disease	ORPHA:296
	Maffucci syndrome	ORPHA:163634
	46,XY difference of sex development due to isolated 17,20-lyase deficiency	ORPHA:90796
Dr Elke SATTLER	Birt-Hogg-Dubé syndrome	ORPHA:122
NA J. [Jelmer] SAVELKOEL	Melioidosis	ORPHA:31202
Pr. Franz SCHAEFER	Immunoglobulin A nephropathy	ORPHA:34145
Pr Manuel SCHIFF	Homocystinuria due to cystathionine beta-synthase deficiency	ORPHA:394
Dr Lars SCHLOTAWA	Multiple sulfatase deficiency	ORPHA:585
Pr Matthias SCHMUTH	CHILD syndrome	ORPHA:139
	Diffuse palmoplantar keratoderma, Bothnian type	ORPHA:2337

Dr Jacqueline SILLERAN CHASSANY	Pheochromocytoma-paraganglioma	ORPHA:573163
Dr Benjamin D. SOLOMON	VACTERL/VATER association	ORPHA:887
	VACTERL with hydrocephalus	ORPHA:3412
Dr Paolo SPINNATO	Autosomal recessive malignant osteopetrosis	ORPHA:667
Pr Ortrud K. STEINLEIN	Birt-Hogg-Dubé syndrome	ORPHA:122
Dr Karolina M. STEPIEN	Fucosidosis	ORPHA:349
Pr Dominique STOPPA-LYONNET	Ataxia-telangiectasia	ORPHA:100
Pr Volker STRAUB	Bethlem muscular dystrophy	ORPHA:610
	Autosomal dominant limb-girdle muscular dystrophy type 1A	ORPHA:266
	TOR1AIP1-related limb-girdle muscular dystrophy	ORPHA:424261
	BVES-related limb-girdle muscular dystrophy	ORPHA:476084
	Autosomal recessive limb-girdle muscular dystrophy, type 28	ORPHA:653725
Dr Elizabeth STREETEN	Osteoporosis-pseudoglioma syndrome	ORPHA:2788
Pr Franc STRLE	Tick-borne encephalitis	ORPHA:297
Dr Salvatore TESTA	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Dr Briac THIERRY	Congenital laryngeal web	ORPHA:2374
Pr Ritva TIKKANEN	Aspartylglucosaminuria	ORPHA:93
Dr Marc TISCHKOWITZ	Hereditary breast and/or ovarian cancer syndrome	ORPHA:145
Pr Ivailo TOURNEV	Congenital cataracts-facial dysmorphism-neuropathy syndrome	ORPHA:48431
Dr Christine TOURNOUD	Fetal methylmercury syndrome	ORPHA:1917
Dr Ludovic TREFOND	Corticosteroid-sensitive aseptic abscess syndrome	ORPHA:54251
Dr Adalena TSATSOPOULOU	Naxos disease	ORPHA:34217
Dr J. C. TURPIN	Tay-Sachs disease	ORPHA:845
Dr Santiago UCROS	Non-transplant-related bronchiolitis obliterans	ORPHA:658612
Dr Laura VALLE	Lynch syndrome	ORPHA:144
Dr Guillaume VELASCO	ICF syndrome	ORPHA:2268
Pr Alain VERLOES	Trisomy 13	ORPHA:3378
Dr Savine VICART	Paramyotonia congenita of Von Eulenburg	ORPHA:684
Dr Nur VILLAR	Paramyotonia congenita of Von Eulenburg	ORPHA:684
Dr Catherine S. VINCENT-DELORME	Perrault syndrome	ORPHA:2855
Dr Melissa P. WASSERSTEIN	Free sialic acid storage disease	ORPHA:834
Pr W.J. [Joost] WIERSINGA	Melioidosis	ORPHA:31202
Pr Enrico S. WRIGHT	Glucose-galactose malabsorption	ORPHA:35710
Dr Ayami YOSHIMI	Pearson syndrome	ORPHA:699
Dr Fabiana ZACCARELLI	Kikuchi-Fujimoto disease	ORPHA:50918
Pr Christos ZOUBOULIS	Malignant atrophic papulosis	ORPHA:679
	PAPASH syndrome	ORPHA:641380
	PASS syndrome	ORPHA:641385
	PsAPASH syndrome	ORPHA:641390
	Genetic autoinflammatory syndrome with acne and/or hidradenitis suppurativa	ORPHA:652510

	Autoinflammatory syndrome with acne and/or hidradenitis suppurativa	ORPHA:653434
	Atrophic papulosis	ORPHA:656071
	Benign atrophic papulosis	ORPHA:656085

List of expert networks reviewing the Orphanet nomenclature in 2023

Name of Expert Network	Disease Name	ORPHA code
ERN BOND	OBSOLETE: Spondyloepimetaphyseal dysplasia with joint laxity	ORPHA:93359
	EXOC6B-related spondyloepimetaphyseal dysplasia with joint laxity	ORPHA:642085
	Spondyloepimetaphyseal dysplasia with joint laxity, Beighton type	ORPHA:642099
	Dysplastic cortical hyperostosis, Al-Gazali type	ORPHA:646136
	Dysplastic cortical hyperostosis	ORPHA:646139
	Hip dysplasia, Beukes type	ORPHA:2114
	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467
ERN BOND, ISDS	Multiple epiphyseal dysplasia, Lowry type	ORPHA:166016
	Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome	ORPHA:166024
	Multiple epiphyseal dysplasia-severe proximal femoral dysplasia syndrome	ORPHA:166029
	Multiple epiphyseal dysplasia-miniepiphyses syndrome	ORPHA:166032
	Spondylometaphyseal dysplasia, Czarny-Ratajczak type	ORPHA:370019
	Hip dysplasia, Beukes type	ORPHA:2114
	OBSOLETE: Spondyloepimetaphyseal dysplasia with joint laxity	ORPHA:93359
	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467
	EXOC6B-related spondyloepimetaphyseal dysplasia with joint laxity	ORPHA:642085
	Spondyloepimetaphyseal dysplasia with joint laxity, Beighton type	ORPHA:642099
	Dysplastic cortical hyperostosis, Al-Gazali type	ORPHA:646136
	Dysplastic cortical hyperostosis	ORPHA:646139
	Multiple epiphyseal dysplasia, Beighton type	ORPHA:166011
	OBSOLETE: Spondyloepimetaphyseal dysplasia with severe short stature	ORPHA:642737
	Mandibuloacral dysplasia associated to MTX2	ORPHA:647667
	Multiple epiphyseal dysplasia type 7	ORPHA:647676
	Dysplasia of head of femur, Meyer type	ORPHA:168621
	OBSOLETE: Peripheral dysostosis	ORPHA:1795
	Metaphyseal acroscyphodysplasia	ORPHA:1240
	Eye defects-arachnodactyly-cardiopathy syndrome	ORPHA:2725

	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome	ORPHA:536467
ERN ITHACA	OBSOLETE: Arnold-Chiari malformation type II	ORPHA:1136
	Split cord malformation type I	ORPHA:1671
	Spina bifida and other spinal dysraphisms	ORPHA:823
	Open spinal dysraphism with a myelomeningocele	ORPHA:93969
	Open spinal dysraphism	ORPHA:268369
	OBSOLETE: Total spina bifida aperta	ORPHA:268377
	OBSOLETE: Thoracolumbosacral spina bifida aperta	ORPHA:268384
	OBSOLETE: Lumbosacral spina bifida aperta	ORPHA:268388
	OBSOLETE: Cervical spina bifida aperta	ORPHA:268392
	OBSOLETE: Cervicothoracic spina bifida aperta	ORPHA:268397
	OBSOLETE: Upper thoracic spina bifida aperta	ORPHA:268740
	Spinal dysraphism with a posterior meningocele	ORPHA:268744
	OBSOLETE: Total spina bifida cystica	ORPHA:268748
	OBSOLETE: Thoracolumbosacral spina bifida cystica	ORPHA:268752
	OBSOLETE: Cervical spina bifida cystica	ORPHA:268762
	OBSOLETE: Cervicothoracic spina bifida cystica	ORPHA:268766
	OBSOLETE: Upper thoracic spina bifida cystica	ORPHA:268770
	Isolated posterior meningocele	ORPHA:268810
	Myelocystocele	ORPHA:268813
	OBSOLETE: Lipoma associated with neurospinal dysraphism	ORPHA:268832
	Lipomyelomeningocele	ORPHA:268835
	OBSOLETE: Leptomyelolipoma	ORPHA:268838
	Split cord malformation type II	ORPHA:573253
	Split cord malformation	ORPHA:573278
	Split cord malformation, composite type	ORPHA:633076
	Spinal dermal sinus	ORPHA:645188
	Dysraphism with stalk	ORPHA:645193
	Limited dorsal myeloschisis	ORPHA:645196
	Closed spinal dysraphism	ORPHA:645202
	Open spinal dysraphism with a posterior meningocele	ORPHA:645270
	Dysraphic spinal cord lipoma	ORPHA:645273
	Spinal cord lipoma	ORPHA:645276
	Fibrolipomatous filum anomaly	ORPHA:645279
	Chaotic conus spinal cord lipoma	ORPHA:645285
	Terminal extramedullary conus spinal cord lipoma	ORPHA:645288
	Transitional extramedullary conus spinal cord lipoma	ORPHA:645291
	Posterior extramedullary conus spinal cord lipoma	ORPHA:645294
	Extramedullary conus spinal cord lipoma	ORPHA:645297
	Lipomatous non-saccular limited dorsal myeloschisis	ORPHA:645300
	Fibroneural non-saccular limited dorsal myeloschisis	ORPHA:645310
	Saccular spinal dysraphism with a stalk to the dome	ORPHA:645319

	Isolated transitional filum lipoma	ORPHA:645322
	Isolated filum lipoma	ORPHA:645325
	Retained medullary cord	ORPHA:645334
	Terminal myelocystocele	ORPHA:645337
	Non-terminal myelocystocele	ORPHA:645340
	Non-saccular limited dorsal myeloschisis	ORPHA:645343
	Saccular limited dorsal myeloschisis	ORPHA:645354
	Intramedullary non-dysraphic spinal cord lipoma	ORPHA:645359
	Dorsal spinal cord lipoma	ORPHA:645362
	Conus spinal cord lipoma	ORPHA:645367
	Myelic limited dorsal malformation	ORPHA:645378
	True myelomeningocele	ORPHA:645383
	Hemi-myelomeningocele	ORPHA:645388
	Hemi-myeloschisis	ORPHA:645393
	Myeloschisis	ORPHA:645398
	True myeloschisis	ORPHA:645401
	OBSOLETE: Lumbosacral spina bifida cystica	ORPHA:268758
ERN VASCERN	Rare lymphatic system anomaly	ORPHA:211255
	Kasabach-Merritt phenomenon	ORPHA:2330
	Rare lymphatic system anomaly	ORPHA:211255
	PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	ORPHA:568062

List of expert networks reviewing Orphanet abstracts/ definitions in 2023

Name of Expert Network	Disease name	ORPHA code
Endo-ERN	46,XY difference of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ORPHA:796
	46,XY difference of sex development due to isolated 17,20-lyase deficiency	ORPHA:551
	Autosomal dominant hypocalcemia	ORPHA:684
	Familial isolated hypoparathyroidism	ORPHA:892
	Familial isolated hypoparathyroidism due to agenesis of parathyroid gland	ORPHA:100
	Familial isolated hypoparathyroidism due to impaired PTH secretion	ORPHA:394
EpiCARE	Classic glucose transporter type 1 deficiency syndrome	ORPHA:699
ERKNet	Immunoglobulin A nephropathy	ORPHA:699
ERN CRANIO	Congenital laryngeal web	ORPHA:752
	Perrault syndrome	ORPHA:893
	Recurrent respiratory papillomatosis	ORPHA:3378

	Regional odontodysplasia	ORPHA:502
	Segmental odontomaxillary dysplasia	ORPHA:2374
ERN PaedCan	Craniopharyngioma	ORPHA:3320
	Pearson syndrome	ORPHA:69
ERN RITA	AApoAIV amyloidosis	ORPHA:364
	Amyloidosis	ORPHA:845
	Ataxia-telangiectasia	ORPHA:48
	ATTRV122I amyloidosis	ORPHA:1552
	ATTRV30M amyloidosis	ORPHA:69723
	Hereditary ATTR amyloidosis	ORPHA:115
ERN-BOND	46,XY difference of sex development due to isolated 17,20-lyase deficiency	ORPHA:163
	Autosomal recessive malignant osteopetrosis	ORPHA:312
	Maffucci syndrome	ORPHA:2238
	Ollier disease	ORPHA:139
ERN-EYE	WAGR syndrome	ORPHA:2337
ERN-Skin	Acral peeling skin syndrome	ORPHA:296
ERN-Skin	Autosomal dominant epidermolytic ichthyosis	ORPHA:559
	CHILD syndrome	ORPHA:667
	Congenital ichthyosiform erythroderma	ORPHA:2855
	Diffuse palmoplantar keratoderma, Bothnian type	ORPHA:145
	Mal de Meleda	ORPHA:882
	Neutral lipid storage disease with ichthyosis	ORPHA:617
	Peeling skin syndrome type A	ORPHA:272
	Peeling skin syndrome type B	ORPHA:899
	Peeling skin-leukonychia-acral punctate keratoses-cheilitis-knuckle pads syndrome	ORPHA:28378
EuroBloodNet	Chronic myelomonocytic leukemia	ORPHA:48431
	Ferroportin disease	ORPHA:54595
	Hereditary hyperferritinemia-cataract syndrome	ORPHA:71277
	HJV or HAMP-related hemochromatosis	ORPHA:77258
	Myelodysplastic syndrome	ORPHA:67039
	Non-HFE-related hemochromatosis	ORPHA:225123
	SLC40A1-related hemochromatosis	ORPHA:263548
	Symptomatic form of HFE-related hemochromatosis	ORPHA:263553
	TFR2-related hemochromatosis	ORPHA:85451
eUROGEN	Congenital bilateral absence of vas deferens	ORPHA:87503
	Congenital primary megaureter	ORPHA:439232
EURO-NMD	Congenital cataracts-facial dysmorphism-neuropathy syndrome	ORPHA:588
	Congenital muscular dystrophy, Fukuyama type	ORPHA:98823
	MERRF	ORPHA:144
	Muscle-eye-brain disease	ORPHA:271861
	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	ORPHA:648562
	Paramyotonia congenita of Von Eulenburg	ORPHA:648569

	Walker-Warburg syndrome	ORPHA:34145
GENTURIS	Hereditary breast and/or ovarian cancer syndrome	ORPHA:263534
	Lynch syndrome	ORPHA:98907
	Von Hippel-Lindau disease	ORPHA:163634
ITHACA	CDK13-related congenital heart defects-intellectual disability-facial dysmorphism syndrome	ORPHA:647834
	Currarino syndrome	ORPHA:83450
	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	ORPHA:79230
	Marinesco-Sjögren syndrome	ORPHA:646278
	Rhombencephalosynapsis	ORPHA:428
	Thrombocytopenia-absent radius syndrome	ORPHA:79394
	Trichorhinophalangeal syndrome type 1	ORPHA:59315
	Trichorhinophalangeal syndrome type 2	ORPHA:370997
	Trisomy 13	ORPHA:329224
MetabERN	Glycogen storage disease due to glucose-6-phosphatase deficiency	ORPHA:85447
	Homocystinuria due to cystathionine beta-synthase deficiency	ORPHA:90796
	Pearson syndrome	ORPHA:3402
	Sandhoff disease	ORPHA:60032
	Tay-Sachs disease	ORPHA:52688
	Transient tyrosinemia of the newborn	ORPHA:189466
	Tyrosinemia type 1	ORPHA:90796
	Tyrosinemia type 2	ORPHA:2239
	Tyrosinemia type 3	ORPHA:444138
VASCERN	Congenital contractural arachnodactyly	ORPHA:465508

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

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Editor-in-chief: Ana Rath – Editor of the report: Charlotte Rodwell
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The correct form when quoting this document is:
«Expert Reviewers for Orphanet in 2023 », Orphanet Report Series, December 2024
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