Orphanet Report Series

Rare Diseases collection

2017

Expert reviewers for Orphanet in 2017

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METHODOLOGY

This document provides the list of Orphanet expert reviewers having contributed to the update of scientific information contained in the Orphanet database of Rare Diseases in 2017.

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 a form containing the scientific information related to a given disease and produced based on peer-reviewed publications:

- Its 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. 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 Orphanumber of the disease/ group of diseases.

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

List of expert reviewers for Orphanet in 2017

Expert Reviewer	Disease name	ORPHA number
Ms Yonit. A ADDISSIE	Muenke syndrome	ORPHA:53271
Pr Marie-Christine ALESSI	Syndromic constitutional thrombocytopenia	ORPHA:477794
	Isolated constitutional thrombocytopenia	ORPHA:477797
	Rare hemorrhagic disorder due to a constitutional platelet anomaly	ORPHA:71202
Dr Eric J. ARNAUD	Pfeiffer syndrome type 2	ORPHA:93259
	Pfeiffer syndrome type 1	ORPHA:93258
	Pfeiffer syndrome type 3	ORPHA:93260
Pr Anne-Catherine BACHOUD-LEVI	Huntington disease	ORPHA:399
	Juvenile Huntington disease	ORPHA:248111
Pr Timothy BARRETT	Alström syndrome	ORPHA:64
M Bart BARTELS	Osteogenesis imperfecta type 1	ORPHA:216796
	Osteogenesis imperfecta type 4	ORPHA:216820
Dr Anne BASSETT	22q11.2 deletion syndrome	ORPHA:567
Dr Isabelle BOCCON-GIBOD	Hereditary angioedema	ORPHA:91378
	Acquired angioedema	ORPHA:91385
Pr Laurence BOUILLET	Acquired angioedema	ORPHA:91385
	Hereditary angioedema	ORPHA:91378
Pr Dominique BREMOND-GIGNAC	Isolated aniridia	ORPHA:250923
Dr Peter BROWN	Primary orthostatic tremor	ORPHA:238606
Pr Katrin BÜRK	Friedreich ataxia	ORPHA:95
Dr Anette BYGUM	Acquired angioedema	ORPHA:91385
	Hereditary angioedema	ORPHA:91378
Dr Teresa CABALLERO	Acquired angioedema	ORPHA:91385
	Hereditary angioedema	ORPHA:91378
Dr Marco CARIFI	Recurrent respiratory papillomatosis	ORPHA:60032
Dr Catherine CORMIER	Osteogenesis imperfecta type 1	ORPHA:216796
	Osteogenesis imperfecta type 4	ORPHA:216820
Pr Helen CROSS	Early infantile epileptic encephalopathy without suppression burst	ORPHA:369894
Dr Danilo DALL'OLIO	Recurrent respiratory papillomatosis	ORPHA:60032
Dr Bénédicte DE FREMINVILLE	Down syndrome	ORPHA:870
Dr Sabine DEFOORT-DHELLEMMES	Leber hereditary optic neuropathy	ORPHA:104
Dr Kumaran DEIVA	Pediatric multiple sclerosis	ORPHA:477738
Dr Rick DERSCH	Lyme disease	ORPHA:91546

Pr Vincent DES PORTES	Fragile X syndrome	ORPHA:908
	Symptomatic form of fragile X syndrome in female carrier	ORPHA:449291
Dr Gérard DUPEYRON	Retinitis pigmentosa	ORPHA:791
Pr Jos I.M. EGGER	Koolen-De Vries syndrome	ORPHA:96169
Dr Salima EL CHEHADEH	VACTERL/VATER association	ORPHA:887
Pr Gareth EVANS	Neurofibromatosis type 2	ORPHA:637
Dr Polly FERGUSON	Majeed syndrome	ORPHA:77297
M Guillaume FERRON	Alström syndrome	ORPHA:64
Dr Trine FOLSERAAS	Primary sclerosing cholangitis	ORPHA:171
Dr Véronique FORIN	Osteogenesis imperfecta type 1	ORPHA:216796
	Osteogenesis imperfecta type 4	ORPHA:216820
Dr Willi GERSCHLAGER	Primary orthostatic tremor	ORPHA:238606
Pr Brigitte GILBERT-DUSSARDIER	Williams syndrome	ORPHA:904
Pr Olivier GOULET	Microvillus inclusion disease	ORPHA:2290
	Congenital tufting enteropathy	ORPHA:92050
	Enteric anendocrinosis	ORPHA:83620
Dr Aina GREIG	Pfeiffer syndrome type 1	ORPHA:93258
	Pfeiffer syndrome type 3	ORPHA:93260
Pr Randi HAGERMAN	Fragile X syndrome	ORPHA:908
Dr Austin HAMM	Pfeiffer syndrome type 1	ORPHA:93258
	Pfeiffer syndrome type 3	ORPHA:93260
Dr Laurence HEIDET	Renal cysts and diabetes syndrome	ORPHA:93111
	BOR syndrome	ORPHA:107
Dr Dominique HERVE	HTRA1-related autosomal dominant cerebral small vessel disease	ORPHA:482077
	Pontine autosomal dominant microangiopathy with leukoencephalopathy	ORPHA:477749
	COL4A1 or COL4A2-related cerebral small vessel disease	ORPHA:477759
	Moyomoya angiopathy	ORPHA:477768
	Familial schizencephaly	ORPHA:481986
	COL4A1 or COL4A2-related cerebral small vessel disease with hemorrhagic tendancy	ORPHA:477765
	HTRA1-related cerebral small vessel disease	ORPHA:482072
	Rare idiopathic macular telangiectasia	ORPHA:482092
	HERNS syndrome	ORPHA:63261
	Binswanger disease	ORPHA:1249
	Not NOTCH3-related small vessel disease of the brain	ORPHA:77304
	Rare central nervous system and retinal vascular disease	ORPHA:71281
Dr Melanie HINGORANI	Isolated aniridia	ORPHA:250923

Dr Nathalie ITZHAR-BAIKIAN	Von Willebrand disease type 1	ORPHA:166078
	Von Willebrand disease type 2	ORPHA:166081
	Von Willebrand disease type 3	ORPHA:166096
Pr Emmanuel JACQUEMIN	Progressive familial intrahepatic cholestasis type 4	ORPHA:480483
	Progressive familial intrahepatic cholestasis type 5	ORPHA:480476
	MYO5B-related progressive familial intrahepatic cholestasis	ORPHA:480491
Ms Brigitte JARRET	22q11.2 deletion syndrome	ORPHA:567
Pr Guillaume JONDEAU	EMILIN-1-related connective tissue disease	ORPHA:485418
Dr Tom Hemming KARLSEN	Primary sclerosing cholangitis	ORPHA:171
Dr Bunyamin KISACIK	Familial Mediterranean fever	ORPHA:342
Dr Michael KNOWLES	Primary ciliary dyskinesia	ORPHA:244
Dr David KOOLEN	Koolen-De Vries syndrome	ORPHA:96169
Dr Paul KRUSZKA	Muenke syndrome	ORPHA:53271
Pr Philippe LABRUNE	Crigler-Najjar syndrome type 1	ORPHA:79234
	Crigler-Najjar syndrome type 2	ORPHA:79235
Pr Didier LACOMBE	Koolen-De Vries syndrome	ORPHA:96169
Pr Nicolas LEBOULANGER	Recurrent respiratory papillomatosis	ORPHA:60032
Dr Irene LITVAN	Multiple system atrophy	ORPHA:102
Pr Avi LIVNEH	Familial Mediterranean fever	ORPHA:342
Pr Birgit LORENZ	Severe early-childhood-onset retinal dystrophy	ORPHA:364055
Dr Angela LUCAS-HERALD	46,XX gonadal dysgenesis	ORPHA:243
	46,XX ovotesticular disorder of sex development	ORPHA:2138
	46,XX testicular disorder of sex development	ORPHA:393
Dr Raffaele MANNA	Familial Mediterranean fever	ORPHA:342
Dr Donna M. MCDONALD-MCGINN	22q11.2 deletion syndrome	ORPHA:567
Pr Wassilios G. MEISSNER	Multiple system atrophy	ORPHA:102
Dr Fanny MOCHEL	Cerebrotendinous xanthomatosis	ORPHA:909
	Refsum disease	ORPHA:773
Dr Fanny MORICE-PICARD	Trichothiodystrophy	ORPHA:33364
Dr Maximilian MUENKE	Muenke syndrome	ORPHA:53271
Dr Yann NADJAR	Niemann-Pick disease type C, adult neurologic onset	ORPHA:216986
Pr Malgorzata NOWACZYK	Smith-Lemli-Opitz syndrome	ORPHA:818
Dr Christophe ORSSAUD	Leber hereditary optic neuropathy	ORPHA:104

Pr Willem OUWEHAND	Lethal hydranencephaly-diaphragmatic hernia syndrome	ORPHA:480528
	Cytosolic phospholipase-A2 alpha deficiency associated bleeding disorder	ORPHA:477787
	Rare hemorrhagic disorder due to a constitutional platelet anomaly	ORPHA:71202
Dr Davide PAREYSON	X-linked Charcot-Marie-Tooth disease type 2	ORPHA:101076
	X-linked Charcot-Marie-Tooth disease type 3	ORPHA:101077
	X-linked Charcot-Marie-Tooth disease type 4	ORPHA:101078
	X-linked Charcot-Marie-Tooth disease type 5	ORPHA:99014
	X-linked Charcot-Marie-Tooth disease type 6	ORPHA:352675
	X-linked Charcot-Marie-Tooth disease type 1	ORPHA:101075
Dr Anne PAVY-LE TRAON	Multiple system atrophy	ORPHA:102
Pr Régis PEFFAULT DE LA TOUR	Paroxysmal nocturnal hemoglobinuria	ORPHA:447
Pr Nicole PHILIP-SARLES	22q11.2 deletion syndrome	ORPHA:567
Dr Chiara PISCIOTTA	X-linked Charcot-Marie-Tooth disease type 1	ORPHA:101075
	X-linked Charcot-Marie-Tooth disease type 2	ORPHA:101076
	X-linked Charcot-Marie-Tooth disease type 3	ORPHA:101077
	X-linked Charcot-Marie-Tooth disease type 4	ORPHA:101078
	X-linked Charcot-Marie-Tooth disease type 5	ORPHA:99014
	X-linked Charcot-Marie-Tooth disease type 6	ORPHA:352675
Dr Christine POITOU-BERNERT	Obesity due to MC3R deficiency	ORPHA:217031
Dr Markus PREISING	Severe early-childhood-onset retinal dystrophy	ORPHA:364055
Ms Laura RALPH	Koolen-De Vries syndrome	ORPHA:96169
Dr Paolo RAMA	Neurotrophic keratopathy	ORPHA:137596
Dr Bernhard RINDLISBACHER	Secondary ciliary dyskinesia	ORPHA:91365
Dr R.M.H. [Rudi] ROUMEN	Anterior cutaneous nerve entrapment syndrome	ORPHA:51890
Dr Marta SACCHETTI	Neurotrophic keratopathy	ORPHA:137596
Dr Julie SALOMON	Microvillus inclusion disease	ORPHA:2290
	Congenital tufting enteropathy	ORPHA:92050
	Enteric anendocrinosis	ORPHA:83620
Dr Charles SCHWARTZ	Allan-Herndon-Dudley syndrome	ORPHA:59
Dr Flore SICRE DE FONTBRUNE	Paroxysmal nocturnal hemoglobinuria	ORPHA:447
Dr Marion SIMONETTA-MOREAU	Primary orthostatic tremor	ORPHA:238606
Pr Ola H. SKJELDAL	Refsum disease	ORPHA:773
Pr Gérard SOCIE	Paroxysmal nocturnal hemoglobinuria	ORPHA:447
Dr Jörg SPIEGEL	Primary orthostatic tremor	ORPHA:238606
Dr Douglas SPROULE	Giant axonal neuropathy	ORPHA:643
Dr Katia STANKOVIC-STOJANOVIC	Familial Mediterranean fever	ORPHA:342
Dr Robert STEINER	Osteogenesis imperfecta type 1	ORPHA:216796
	Osteogenesis imperfecta type 4	ORPHA:216820

Dr Rachel STRAUSSBERG	Familial infantile bilateral striatal necrosis	ORPHA:225154
Pr Jeffrey J. SWIGRIS	Idiopathic pulmonary fibrosis	ORPHA:2032
Pr Maithé TAUBER,	Obesity due to MC3R deficiency	ORPHA:217031
Dr. G.M. [Gisela] TERWINDT	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	ORPHA:247691
Dr Elaine TIERNEY	Smith-Lemli-Opitz syndrome	ORPHA:818
Dr Renaud TOURAINE	Down syndrome	ORPHA:870
Pr Elisabeth TOURNIER-LASSERVE	Rare central nervous system and retinal vascular disease	ORPHA:71281
Pr Annick TOUTAIN	Koolen-De Vries syndrome	ORPHA:96169
Dr A.J. [Anneke] VAN DER KOOI	Limb-girdle muscular dystrophy	ORPHA:263
Pr Hélène VERDOUX	Diethylstilbestrol syndrome	ORPHA:1916
Pr William M.A. VERHOEVEN	Koolen-De Vries syndrome	ORPHA:96169
Dr Catherine VIGNAL-CLERMONT	Leber hereditary optic neuropathy	ORPHA:104
Pr Corinne VIGOUROUX	Laminopathy type Decaudain-Vigouroux	ORPHA:137871
Pr Gregor K. WENNING	Multiple system atrophy	ORPHA:102
Pr Dagmar WIECZOREK	Mandibulofacial dysostosis-microcephaly syndrome	ORPHA:79113
Pr Anthony S. WIERZBICKI	Refsum disease	ORPHA:773
Dr Katia YOUSSOV	Juvenile Huntington disease	ORPHA:248111
	Huntington disease	ORPHA:399
Dr Patrick YU-WAI-MAN	Leber hereditary optic neuropathy	ORPHA:104

List of expert networks reviewing Orphanet data in 2017

Expert group	Classification name	ORPHA number
FAI2R (Filière de santé des maladies auto-immunes et auto-inflammatoires rares)	Rare systemic or rheumatologic disease	ORPHA:98023
Filfoie (Filière de santé maladies rares du foie de l'adulte et de l'enfant)	Rare hepatic disease	ORPHA:57146
OSCAR (Filière de santé maladies rares de l'os, du calcium et du cartilage)	Rare bone disease	ORPHA:93419

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

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