Orphanet Report Series

Rare Diseases collection

2016

Expert reviewers for Orphanet in 2016

Instituts thématiques

Inserm

Institut national de la santé et de la recherche médicale

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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 2016. 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 a form containing the scientific information related to a given disease:

- 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.

The form is pre-filled with the data available in the Orphanet database or coming from a literature search.
The expert is invited to validate, to correct or to complete the information submitted to him/her.

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.

List of expert reviewers for Orphanet in 2016

Expert reviewer	Disease name	ORPHA number
Pr Véronique ABADIE	CHARGE syndrome	138
	Isolated Pierre Robin syndrome	718
	Stickler syndrome	828
Dr Olivier ABBO	Van der Woude syndrome	888
Ms Delphine ALIGON	Craniopharyngioma	54595
	Medulloblastoma	616
Dr Yves ALLENBACH	Autoimmune necrotizing myopathy	206569
	Dermatomyositis	221
	Juvenile polymyositis	93568
	Juvenile dermatomyositis	93672
	Polymyositis	732
Dr Jean-Bernard ANDRIEU	Juvenile neuronal ceroid lipofuscinosis	79264
Dr Arleen AUERBACH	Fanconi anemia	84
Pr Marie-Françoise AVRIL	Familial melanoma	618
Dr Carmen AYUSO GARCÍA	Choroideremia	180
Pr Elfride DE BAERE	Blepharophimosis-epicanthus inversus-ptosis due to 3q23 rearrangement syndrome	261559
Dr Gilles BAGOU	Exercise-induced malignant hyperthermia	466650
	Rare disease with malignant hyperthermia	466658
Dr Nadia BAHI-BUISSON	Rett syndrome	778
Dr Wilma BARCELLINI	Hereditary spherocytosis	822
Pr Timothy G. BARRETT	Bardet-Biedl syndrome	110
Dr Geneviève BAUJAT	Ellis Van Creveld syndrome	289
	Jeune syndrome	474
Dr Daniel BAYART	Bardet-Biedl syndrome	110
	Juvenile neuronal ceroid lipofuscinosis	79264
Dr Gareth BAYNAM	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	457485
Pr Philip L. BEALES	Bardet-Biedl syndrome	110
Ms Ouarda BENKHALED	Craniopharyngioma	54595
	Medulloblastoma	616
Dr Pascale BENLIAN	Familial chylomicronemia syndrome	444490
Dr Yasmine BENNIS	Isolated trigonocephaly	3366

Dr Sonia BERRIH-AKNIN	Immune-mediated acquired neuromuscular junction disease	464764
Pr Jérôme BERTHERAT	Cushing syndrome	553
Dr Alberto BETTINELLI	Gitelman syndrome	358
Dr Mario G. BIANCHETTI	Gitelman syndrome	358
Dr Marieke BIEGSTRAATEN	Gaucher disease type 1	77259
Pr Thierry BILLETTE DE VILLEMEUR	Ataxia-telangiectasia	100
Dr Guillaume BOLLEE	Adenine phosphoribosyltransferase deficiency	976
Ms Anne-Claire BOURGEOIS	Craniopharyngioma	54595
	Medulloblastoma	616
Dr Audrey BOUTRON	Lipoyl transferase 2 deficiency	447795
Pr Kym BOYCOTT	Dysequilibrium syndrome	1766
Pr Maria Luisa BRANDI	Multiple endocrine neoplasia type 2	653
Pr Gérard BREART	Fetal alcohol syndrome	1915
Pr Pierre BRISSOT	Hemochromatosis type 5	447792
Dr Jacinta BUSTAMANTE	Susceptibility to infection due to TYK2 deficiency	331226
Ms Céline CANIZARÈS	Craniopharyngioma	54595
	Medulloblastoma	616
Pr Mamede DE CARVALHO	Primary lateral sclerosis	35689
Dr Suzanne B. CASSIDY	Prader-Willi syndrome	739
Pr Stéphane CHABRIER	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome	280679
Dr Suresh CHANDRAN	Ectopia cordis	448270
Dr Gulraiz CHAUDRY	Diffuse lymphatic malformation	141209
Pr Jean-Alain CHAYVIALLE	Cap polyposis	160148
	Cronkhite-Canada syndrome	2930
Dr Mathilde CHEVIGNARD	Craniopharyngioma	54595
	Medulloblastoma	616
Dr Maria Roberta CILIO	KCNQ2-related epileptic encephalopathy	439218
Pr Angus CLARKE	Rett syndrome	778
Pr Robert J. COFFEY	Ménétrier disease	2494
Dr Martine COHEN-SOLAL	Familial hypocalciuric hypercalcemia	405
Ms Kim COLAPINTO	Hirschsprung disease	388
Dr Thomas COLSON	Supratip dysplasia	466695
Dr Joseph CONNORS	Hodgkin lymphoma	98293
Dr Catherine CORMIER	Achondroplasia	15
Pr Helen CROSS	KCNQ2-related epileptic encephalopathy	439218
Dr Ivana DABAJ	West syndrome	3451

Dr Federica DAGRADI	Jervell and Lange-Nielsen syndrome	90647
	Romano-Ward syndrome	101016
Mr Jesper DAMMEYER	Usher syndrome type 1	231169
	Usher syndrome type 2	231178
Pr Mehul DATTANI	Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome	370006
Pr Olivier DEVUYST	Gitelman syndrome	358
Dr Anastasia DIMITROPOULOS	Prader-Willi syndrome	739
Dr Roni DODIUK-GAD	Darier disease	218
Pr Lex DOYLE	Bronchopulmonary dysplasia	70589
Pr Françis DOZ	Medulloblastoma	616
Dr Gérard DUPEYRON	Usher syndrome type 1	231169
	Usher syndrome type 2	231178
Dr Elizabeth DYKENS	Prader-Willi syndrome	739
Dr Sarah ELSEA	Smith-Magenis syndrome	819
Dr Elizabeth ENGLE	Duane retraction syndrome	233
Dr Odile ENJOLRAS	Tufted angioma	1063
Pr Bruno FAUSTREL	Adult-onset Still disease	829
Dr Salah FERKAL	Neurofibromatosis type 1	636
Pr Josef FINSTERER	Kennedy disease	481
Dr Véronique FORIN	Osteogenesis imperfecta type 2	216804
Ms Sonia FRAISSE	Rubinstein-Taybi syndrome	783
Pr Brenda L. GALLIE	Uveal melanoma	39044
Pr Noël GARABEDIAN	Isolated tracheo-esophageal fistula	454750
Pr David GENEVIEVE	Goldenhar syndrome	374
Ms Celia GIACHETI	Opitz G/BBB syndrome	2745
Dr Janine-Sophie GIRAUDET-LE QUINTREC	Marfan syndrome type 1	284963
	Marfan syndrome type 2	284973
Pr Olivier GOULET	Primary bile acid malabsorption	449262
Dr Therese GRANT	Fetal alcohol syndrome	1915
Dr Benjamin GREENBERG	Neuromyelitis optica	71211
Dr Aina GREIG	Pfeiffer syndrome type 1	93258
Pr Bodo GRIMBACHER	Autosomal dominant hyper-IgE syndrome	2314
Dr Pierre GUERRESCHI	Isolated trigonocephaly	3366
Dr Daniel GUERRIER	Mayer-Rokitansky-Küster-Hauser syndrome type 1	247775
	Mayer-Rokitansky-Küster-Hauser syndrome type 2	2578
Pr Pierre-Jean GUILLAUSSEAU	Maternally-inherited diabetes and deafness	225

Pr Loïc P. GUILLEVIN	Eosinophilic granulomatosis with polyangiitis	183
	Granulomatosis with polyangiitis	900
	Microscopic polyangiitis	727
	Polyarteritis nodosa	767
Pr Christian HAMEL	Usher syndrome type 1	231169
	Usher syndrome type 2	231178
Dr Austin HAMM	Pfeiffer syndrome type 1	93258
Dr Jean-Pierre HARDELIN	Kallmann syndrome	478
Dr Timothy HARTSHORNE	CHARGE syndrome	138
Dr Laurence HEIDET	Alport syndrome	63
Dr Björn HOFFMANN	Classic galactosemia	79239
Dr Roderick HOUWEN	Benign recurrent intrahepatic cholestasis type 1	99960
	Benign recurrent intrahepatic cholestasis type 2	99961
Dr Derralynn HUGHES	Gaucher disease type 1	77259
Dr Won Jae HUH	Ménétrier disease	2494
Pr Kimiyoshi ICHIDA	Hereditary xanthinuria	3467
Dr Tracy JIRIKOWIC	Fetal alcohol syndrome	1915
Pr Guillaume JONDEAU	Familial thoracic aortic aneurysm and aortic dissection	91387
Pr Cheryl JORCYK	Stüve-Wiedemann syndrome	3206
Dr Monique KAMINSKI	Fetal alcohol syndrome	1915
Ms Virginie KIEFFER	Craniopharyngioma	54595
	Medulloblastoma	616
Pr Alfried KOHLSCHÜTTER	Juvenile neuronal ceroid lipofuscinosis	79264
Pr Renée KRIVOSIC	Rare disease with malignant hyperthermia	466658
	Exercise-induced malignant hyperthermia	466650
Pr Philippe LABRUNE	Glycogen storage disease due to liver phosphorylase kinase deficiency	264580
	Glycogen storage disease due to phosphorylase kinase deficiency	370
Pr Didier LACOMBE	Bardet-Biedl syndrome	110
	Rubinstein-Taybi syndrome	783
Dr François LACOUR-GAYET	Double outlet right ventricle	3426
Dr Miriam LADSOUS	Pendred syndrome	705
Dr Thierry LEBLANC	Dyskeratosis congenita	1775
Pr Véronique LEBLOND	Waldenström macroglobulinemia	33226
Pr Nicolas LEBOULANGER	Choanal atresia	137914
Dr Hélène DE LEERSNYDER	Smith-Magenis syndrome	819

Dr Christine LIANG	Dermatofibrosarcoma protuberans	31112
Dr Tobias LODDENKEMPER	Undetermined early-onset epileptic encephalopathy	442835
Pr Dietmar R. LOHMANN	Uveal melanoma	39044
Dr Anne LOMBES	Kearns-Sayre syndrome	480
Dr Juan Carlos LOPEZ GUTIERREZ	Vascular anomaly or angioma	68419
Pr Birgit LORENZ	Usher syndrome type 1	231169
	Usher syndrome type 2	231178
Dr Alfredo LUCENDO	Eosinophilic esophagitis	73247
	Eosinophilic gastroenteritis	2070
Dr Caroline LUND	Aicardi syndrome	50
Dr Kerstin LUNDBERG LARSEN	Marfan syndrome type 1	284963
Dr Franciska MALFAIT	Ehlers-Danlos syndrome, classic type	287
Dr Romain MARIGNIER	Neuromyelitis optica	71211
Dr Sandrine MARLIN	Neurologic Waardenburg-Shah syndrome	163746
	Waardenburg-Shah syndrome	897
	Waardenburg syndrome type 1	894
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Pr Irene M.J. MATHIJSSEN	Muenke syndrome	53271
Dr Eve MAUBEC	Familial melanoma	618
Ms Luciana MAXIMINO	Opitz G/BBB syndrome	2745
Dr Sandra MERCIER	Alobar holoprosencephaly	93925
	Lobar holoprosencephaly	93924
	Semilobar holoprosencephaly	220386
Dr Paolo MERELLA	Anisakiasis	1070
Dr Michele MEYER	Idiopathic dropped head syndrome	447881
Dr Dawn MIKELONIS	Stüve-Wiedemann syndrome	3206
Dr José María MILLÁN SALVADOR	Choroideremia	180
Dr Saddek MOHAND-SAID	Usher syndrome type 1	231169
	Usher syndrome type 2	231178
Pr Anne MONCLA	Angelman syndrome	72
Dr Karine MORCEL	Mayer-Rokitansky-Küster-Hauser syndrome type 1	247775
	Mayer-Rokitansky-Küster-Hauser syndrome type 2	2578
Dr Hermann L. MÜLLER	Craniopharyngioma	54595
Pr Rima NABBOUT	West syndrome	3451
Pr Irene NETCHINE	Silver-Russell syndrome	813
Pr Maria NIJHUIS-VAN DER SANDEN	Prader-Willi syndrome	739
Dr Gen NISHIMURA	Autosomal recessive brachyolmia	448242

Dr Laure Hélène NOEL	Immunotactoid glomerulopathy	97567
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Pr Sylvie ODENT	Alobar holoprosencephaly	93925
	Lobar holoprosencephaly	93924
	Semilobar holoprosencephaly	220386
Ms Marianne ØKSNES	Addison disease	85138
Pr Arnold ORANJE	Maculopapular cutaneous mastocytosis	79457
Pr Julia OXFORD	Stüve-Wiedemann syndrome	3206
Pr Tonya PALERMO	Sickle cell anemia	232
Dr Davide PAREYSON	Charcot-Marie-Tooth disease type 1	65753
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Dr Mahsa PARVIZ	Gamma-aminobutyric acid transaminase deficiency	2066
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Dr Gregory PASTORES	Gaucher disease type 1	77259
Pr Jean-François PAYEN	Exercise-induced malignant hyperthermia	466650
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Dr Phillip PEARL	Gamma-aminobutyric acid transaminase deficiency	2066
	Succinic semialdehyde dehydrogenase deficiency	22
Dr Alessandro PECCI	MYH9-related disease	182050
Dr Antonio PERCESEPE	VACTERL/VATER association	887
Dr Alan K. PERCY	Rett syndrome	778
Dr Petros PERROS	Euthyroid Graves orbitopathy	466682
Dr Chiara PISCIOTTA	Charcot-Marie-Tooth disease type 1	65753
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	Charcot-Marie-Tooth disease type 1D	101084
	Charcot-Marie-Tooth disease type 1E	90658

	Charcot-Marie-Tooth disease type 1F	101085
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Dr Manuel POSADA DE LA PAZ	Toxic oil syndrome	227972
Dr Nicolas POURSAC	Idiopathic dropped head syndrome	447881
Dr Markus PREISING	Usher syndrome type 1	231169
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Dr Anne PUEL	Predisposition to invasive fungal disease due to CARD9 deficiency	457088
Pr Hervé PUY	Familial porphyria cutanea tarda	443062
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Pr Pierre QUARTIER DIT MAIRE	Systemic-onset juvenile idiopathic arthritis	85414
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RARECARE	Squamous cell carcinoma of rectum	424002
Dr Linda REUS	Prader-Willi syndrome	739
Dr Antonio RICHIERI-COSTA	Opitz G/BBB syndrome	2745
Pr Stephen P ROBERTSON	Otopalatodigital syndrome	669
Ms Elizabeth ROOF	Prader-Willi syndrome	739
Dr M.E. [Estela] RUBIO-GOZALBO	Classic galactosemia	79239
Pr Monique RYAN	Autosomal dominant Charcot-Marie-Tooth disease type 2E	99939
Dr Iván SÁNCHEZ FERNÁNDEZ	Undetermined early-onset epileptic encephalopathy	442835
Dr Filippo SANTORELLI	NARP syndrome	644
Dr Fernando SANTOS	Gitelman syndrome	358
Pr Jean-Christophe SAURIN	Cap polyposis	160148
	Cronkhite-Canada syndrome	2930
Dr Sharon SAVAGE	Dyskeratosis congenita	1775
Dr Chrysalyne D. SCHMULTS	Dermatofibrosarcoma protuberans	31112
Dr Michael SECKELER	Rheumatic fever	3099
Dr Sherin SHAABAN	Duane retraction syndrome	233
Dr Geir SIEM	Jervell and Lange-Nielsen syndrome	90647
Pr Katherine SIMS	Norrie disease	649
Dr Blanka STIBURKOVA	Hereditary xanthinuria	3467
Dr Katrina TATTON BROWN	Weaver syndrome	3447
Pr Maithé TAUBER	Genetic non-syndromic obesity	98267
	Prader-Willi syndrome	739
Ms Emmanuelle TAUPIAC	Rubinstein-Taybi syndrome	783
Mr Ken TAWARA	Stüve-Wiedemann syndrome	3206
Dr Denise THUILLEAUX	Prader-Willi syndrome	739

Ms Eva TOUSSAINT	Rubinstein-Taybi syndrome	783
Dr Stéphanie TOUTAIN	Fetal alcohol syndrome	1915
Dr Cameron TRENOR	Diffuse lymphatic malformation	141209
Dr Catherine TURLEAU	Cat-eye syndrome	195
Pr Dominique-Charles VALLA	Caroli disease	53035
Dr Nens VAN ALFEN	Neuralgic amyotrophy	2901
Dr Janielle VAN ALFEN-VAN DER VELDEN	Prader-Willi syndrome	739
Dr Andreas VAN BAALEN	Febrile infection-related epilepsy syndrome	163703
Dr Lideke VAN DER STEEG	Hirschsprung disease	388
Dr Wendy VAN DER WOERD	Benign recurrent intrahepatic cholestasis type 1	99960
	Benign recurrent intrahepatic cholestasis type 2	99961
Dr Leo VAN VLIMMEREN	Prader-Willi syndrome	739
Dr Christelle VAULOUP-FELLOUS	Congenital rubella syndrome	290
Dr Gry VELVIN	Marfan syndrome type 1	284963
Ms Valentine VERDIER	Craniopharyngioma	54595
	Medulloblastoma	616
Dr Anja VIEHMANN	Uveal melanoma	39044
Pr John VISSING	Oculopharyngeal muscular dystrophy	270
Dr Emma WAKELING	Silver-Russell syndrome	813
Dr Heng WANG	Prolidase deficiency	742
Dr Emma WEBB	Hypomyelinating leukodystrophy with or without oligondontia and/or hypogonadism	289494
	Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome	370006
Pr Udo WENDEL	Hartnup disease	2116
Dr Mark WILKINSON	Leber congenital amaurosis	65
	Stargardt disease	827
Dr Paul W.WIRTZ	Lambert-Eaton myasthenic syndrome	43393
Dr Marcin WLODARSKI	Monocytopenia with susceptibility to infections	228423
Dr Cristina WOELLNER	Autosomal dominant hyper-IgE syndrome	2314
Pr Pierre WOLKENSTEIN	Neurofibromatosis type 1	636
Pr Jacques YOUNG	Kallmann syndrome	478
Dr Alberto ZANELLA	Hemolytic anemia due to red cell pyruvate kinase deficiency	766
	Hereditary spherocytosis	822
Pr Martin ZENKER	Johanson-Blizzard syndrome	2315

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

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