Orphanet Report Series

Rare Diseases collection

2015

Expert reviewers for Orphanet in 2015

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 updating the scientific information contained in the Orphanet database of rare diseases in 2015.
Experts were identified through their publications, and their activity related with 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 or group of diseases:

- Its nomenclature: preferred term and synonyms;
- Related genes and the type of relationship between the gene and the diseases, namely: causative genes, 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 can 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 – Prognosis

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 (on nomenclature, on genetics or on epidemiological data) in order to update the Orphanet content.

Data presentation

Experts are listed by alphabetical order of their name and with the expertised disease name and Orphanumber of the disease.

List of expert reviewers for Orphanet in 2015

Expert reviewer	Disease name	Orphanumber
Dr Michael ABRUZZO	Abruzzo-Erickson syndrome	921
Pr Alberto ALBANESE	Adult-onset cervical dystonia, DYT23 type	420492
	Autosomal dominant focal dystonia, DYT7 type	93963
	Cervical dystonia	93962
	Cranio-cervical dystonia with laryngeal and upper-limb involvement	420485
	Dystonia-aphonia syndrome	412217
	Laryngeal dyskinesia	93961
	Limb dystonia	93957
	Truncal dystonia	93956
Dr Mathieu ANHEIM	Spinocerebellar ataxia with axonal neuropathy type 2	64753
Pr Tania ATTIE-BITACH	Meckel syndrome	564
Pr TONY ATTWOOD	Asperger syndrome	1162
Dr Laurent BECQUEMONT	Antidepressant or antipsychotics toxicity or dose selection	413667
	Azathioprine or 6-mercatopurine toxicity or dose selection	413687
	Curariform drugs toxicity	413693
	Methotrexate toxicity or dose selection	413690
	Oral antidiabetic drugs toxicity or dose selection	413681
	Phenytoin or carbamazepine toxicity	414750
	Resistance to vitamin K antagonists	413684
	Statin toxicity	413696
	Vitamin K antagonists toxicity or dose selection	413674
Pr Soumeya BEKRI	X-linked sideroblastic anemia and ataxia	2802
Pr Peter BERLIT	Sneddon syndrome	820
Pr Regina BETZ	Woolly hair	170
Dr Kim BLAKE	CHARGE syndrome	138
Dr Catherine BLANCHET	Usher syndrome	886
Pr Jean-Yves BLAY	Pleomorphic undifferentiated sarcoma	293190
Pr Ulrike BLUME-PEYTAVI	Woolly hair	170
Mr Francesco BONELLA	Hereditary pulmonary alveolar proteinosis	264675
	Secondary pulmonary alveolar proteinosis	420259
Dr Didier BOUCCARA	Menière disease	45360
Pr Maria Luisa BRANDI	Multiple endocrine neoplasia type 1	652
	Multiple endocrine neoplasia type 2	653
	Multiple endocrine neoplasia type 2A	247698
	Multiple endocrine neoplasia type 2B	247709
Dr Jeffrey BROSCO	Histidinemia	2157

Expert reviewer	Disease name	Orphanumber
Dr Alfredo BRUSCO	Adult-onset autosomal dominant leukodystrophy	99027
Ms Teresa BULGER	Malignant hyperthermia	423
Dr James BURRELL	Corticobasal degeneration	278
Pr Jean CABANE	Rare autonomic nervous system disorder	423662
Pr Jacques CADRANEL	Primary pulmonary lymphoma	2420
Dr Almuth CALIEBE	Catel-Manzke syndrome	1388
Dr Michael CARR	Anterior urethral valve	435372
	Fetal lower urinary tract obstruction	435365
Dr Suresh CHANDRAN	Isolated agenesis of gallbladder	440987
Dr Perrine CHARLES-IGNATIEW	Ataxia - oculomotor apraxia type 1	1168
	Spinocerebellar ataxia with axonal neuropathy type 2	64753
Dr Maria Roberta CILIO	Benign familial neonatal epilepsy	1949
Pr Rolando CIMAZ	Kawasaki disease	2331
Pr Adrian CLARK	Familial glucocorticoid deficiency	361
Dr Laurie COHEN	Diencephalic syndrome	1672
Pr Bernard COMBE	Psoriatic arthritis	40050
Pr Paul COPPO	Acquired thrombotic thrombocytopenic purpura	93585
	Congenital thrombotic thrombocytopenic purpura	93583
	Thrombotic thrombocytopenic purpura	54057
Pr Valérie CORMIER-DAIRE	Desbuquois syndrome	1425
Pr Vincent COTTIN	Cryptogenic organizing pneumonia	1302
Dr Helen CROSS	Undetermined early-onset epileptic encephalopathy	442835
Pr Piotr CZAUDERNA	Hepatoblastoma	449
Dr Annachiara DE SANDRE-GIOVANNOLI	Restrictive dermopathy	1662
Dr Daniele DE SETA	Idiopathic facial palsy	2810
Dr Marc D'HOOGHE	X-linked sideroblastic anemia and ataxia	2802
Dr Federico DI ROCCO	Familial esophageal achalasia	99723
	Idiopathic achalasia	930
	Sporadic achalasia	99722
Pr Angela DISPENZIERI	POEMS syndrome	2905
Pr François DOZ	Hereditary retinoblastoma	357027
	Non-hereditary retinoblastoma	357034
	Retinoblastoma	790
Dr Nicolas DUPIN	Kaposi sarcoma	33276
Dr Hans DUVEKOT	Maternal phenylketonuria	2209
Dr David DYMENT	SHORT syndrome	3163
Pr Malcolm FERGUSON-SMITH	Multiple self-healing squamous epithelioma	65748
Dr Evelyne FERRARY	Menière disease	45360
Dr Roseline FROISSART	Sialidosis type 1	812
Dr Toshiyuki FUKAO	Beta-ketothiolase deficiency	134

Expert reviewer	Disease name	Orphanumber
Dr Sixto GARCÍA-MIÑAÚR	Simpson-Golabi-Behmel syndrome	373
Pr Loïc GARÇON	Dehydrated hereditary stomatocytosis	3202
Dr Sidney GOSPE	Pyridoxine-dependent epilepsy	3006
Dr David GOUDIE	Multiple self-healing squamous epithelioma	65748
Dr Philippe GOUPILLE	Psoriatic arthritis	40050
Pr Gilles GRATEAU	AA amyloidosis	85445
	AApoAI amyloidosis	93560
	AApoAII amyloidosis	238269
	ABeta2M amyloidosis	439246
	AFib amyloidosis	93562
	ATTRV122I amyloidosis	85451
	ATTRV30M amyloidosis	85447
	Hereditary amyloidosis with primary renal involement	85450
	Hereditary ATTR amyloidosis	271861
	Hereditary cerebral hemorrhage with amyloidosis	85458
	Primary cutaneous amyloidosis	137807
Pr Gilles GRATEAU	Wild type ATTR amyloidosis	330001
Pr Eyal GRUNEBAUM	Purine nucleoside phosphorylase deficiency	760
Dr Nathalie GUFFON	Sialidosis type 1	812
Pr Loïc GUILLEVIN	Cutaneous polyarteritis nodosa	439729
	Primary polyarteritis nodosa	439737
	Secondary polyarteritis nodosa	439746
	Single-organ polyarteritis nodosa	439755
	Systemic polyarteritis nodosa	439762
Mr Claude-Alexandre GUSTAVE	Bacterial toxic-shock syndrome	36234
	Staphylococcal toxic-shock syndrome	99919
	Streptococcal toxic-shock syndrome	99918
Pr Johannes HÄBERLE	Argininosuccinic aciduria	23
	Carbamoyl-phosphate synthetase 1 deficiency	147
	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	415
	Ornithine transcarbamylase deficiency	664
Pr Christian HAMEL	Usher syndrome	886
Pr R.C. [Raoul] HENNEKAM	Temple-Baraitser syndrome	420561
Dr A.Micheil INNES	SHORT syndrome	3163
Pr Harald JÜPPNER	Caffey disease	1310
Dr Agnieszka JURECKA	Adenylosuccinate lyase deficiency	46
Pr Jean KANITAKIS	Disseminated superficial actinic porokeratosis	79152
	Porokeratosis of Mibelli	735
	Porokeratosis plantaris palmaris et disseminata	737
Pr Alexander KATOULIS	Syringocystadenoma papilliferum	840

Expert reviewer	Disease name	Orphanumber
Dr Maria Ines KAVAMURA	Cardiofaciocutaneous syndrome	1340
Dr Usha KINI	Ulnar-mammary syndrome	3138
Dr Karin KLIJN	Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism	280679
	Moyamoya disease	2573
Pr Isabelle KONE-PAUT	Familial cold urticaria	47045
	Genetic urticaria	182734
	Muckle-Wells syndrome	575
Pr Philippe LABRUNE	Fructose-1,6-bisphosphatase deficiency	348
	Hereditary fructose intolerance	469
Dr Francois LACOUR-GAYET	Double outlet right ventricle	3426
	Double outlet right ventricle with atrioventricular septal defect, pulmonary stenosis, heterotaxy	423712
	Double outlet right ventricle with doubly committed ventricular septal defect	99047
	Double outlet right ventricle with subaortic or doubly committed ventricular septal defect	423693
	Double outlet right ventricle with subaortic ventricular septal defect	99044
	Taussig-Bing syndrome	101042
Dr Pablo LAPUNZINA BADIA	Simpson-Golabi-Behmel syndrome	373
Dr Véronique LEBLOND	Waldenström macroglobulinemia	33226
Dr Jean-Christophe LEGA	Kawasaki disease	2331
Dr Jules LEROY	Mucolipidosis type III alpha/beta	423461
	Mucolipidosis type III gamma	423470
Pr Thierry LEVADE	Farber disease	333
Dr Aikaterini LIAKOU	Syringocystadenoma papilliferum	840
Pr Gerard LINA	Bacterial toxic-shock syndrome	36234
	Staphylococcal toxic-shock syndrome	99919
	Streptococcal toxic-shock syndrome	99918
Dr Helen LINDEN	Ulnar-mammary syndrome	3138
Pr Lorenzo LO MUZIO	Gorlin syndrome	377
Dr Miguel LOPEZ	Wolfram-like syndrome	411590
Pr Birgit LORENZ	Leber congenital amaurosis	65
Dr Silvia LOURENÇO	Cheilitis glandularis	1221
Dr Alfredo LUCENDO	Proton-pump inhibitor-responsive esophageal eosinophilia	411696
Dr Henry LYNCH	Familial atypical multiple mole melanoma syndrome	404560
	Melanoma-pancreatic cancer syndrome	51013
Dr Carl MALCHOFF	Differentiated thyroid carcinoma	146
Dr Víctor MARTÍNEZ-GLEZ	Simpson-Golabi-Behmel syndrome	373
Pr Juliette MAZEREEUW-HAUTIER	Erythrokeratoderma variabilis progressiva	308166

Expert reviewer	Disease name	Orphanumber
Pr Jeffrey MEDIN	Farber disease	333
Dr Maria J MERINO	Familial renal cell carcinoma	151
	Hereditary papillary renal cell carcinoma	47044
	Renal cell carcinoma	217071
Pr Dan MILEA	Autosomal dominant optic atrophy plus syndrome	1215
	Autosomal dominant optic atrophy, classic form	98673
Dr David MONTANI	Pulmonary veno-occlusive disease and/or pulmonary capillary haemangiomatosis	431353
Dr Karine MORCEL	Mayer-Rokitansky-Küster-Hauser syndrome type 1	247775
	Mayer-Rokitansky-Küster-Hauser syndrome type 2	2578
Pr Etienne MORNET	Adult hypophosphatasia	247676
	Childhood-onset hypophosphatasia	247667
	Hypophosphatasia	436
	Infantile hypophosphatasia	247651
	Odontohypophosphatasia	247685
	Perinatal lethal hypophosphatasia	247623
	Prenatal benign hypophosphatasia	247638
Pr Peter S MORTIMER	Congenital primary lymphedema	2416
	Late-onset primary lymphedema	289825
	Non hereditary congenital primary lymphedema	79450
	Non-hereditary late-onset primary lymphedema	90185
Pr Robert NAVIAUX	Alpers-Huttenlocher syndrome	726
Pr Giovanni NERI	Cardiofaciocutaneous syndrome	1340
Mr Julián NEVADO BLANCO	Simpson-Golabi-Behmel syndrome	373
Pr Patrick NIAUDET	Juvenile nephropathic cystinosis	411634
	Nephropathic infantile cystinosis	411629
	Ocular cystinosis	411641
Dr Marcello NICO	Cheilitis glandularis	1221
Dr Gen NISHIMURA	Autosomal dominant brachyolmia	93304
	Brachyolmia	1293
	Brachyolmia type 1, Hobaek type	93301
	Brachyolmia, Maroteaux type	93302
	Brachyolmia-amelogenesis imperfecta syndrome	2899
Dr Virginia NUNES	Wolfram-like syndrome	411590
Pr Sylvie ODENT	PDE4D haploinsufficiency syndrome	439822
Ms Viveca ODLIND	Placental insufficiency	439167
Pr Asher ORNOY	Fetal valproate syndrome	1906
Dr Coro PAISAN-RUIZ	Hereditary late-onset Parkinson disease	411602
Dr Johanna PALMIO	Distal myopathy	599
	Distal myopathy, Welander type	603
	Miyoshi myopathy	45448

Expert reviewer	Disease name	Orphanumber
Dr Nirav PATEL	Progressive hemifacial atrophy	1214
Dr Nikos PATSINAKIDIS	Cutaneous lupus erythematosus	535
Pr Giorgio PERILONGO	Hepatoblastoma	449
Dr Véronique PICARD	Dehydrated hereditary stomatocytosis	3202
Dr Véronique PINGAULT	Tietz syndrome	42665
	Waardenburg syndrome	3440
	Waardenburg syndrome type 1	894
	Waardenburg syndrome type 2	895
	Waardenburg syndrome type 3	896
Dr J.K. [Hans Kristian] PLOOS VAN AMSTEL	Wilson-Turner syndrome	3459
Dr Neil POLLOCK	Malignant hyperthermia	423
Pr B.T. [Bwee Tien] POLL-THE	Refsum disease	773
Dr Chitra PRASAD	CHARGE syndrome	138
Dr Markus PREISING	Leber congenital amaurosis	65
Pr Vincent PROCACCIO	Autosomal dominant optic atrophy plus syndrome	1215
	Autosomal dominant optic atrophy, classic form	98673
Dr Neil RAJAN	Brooke-Spiegler syndrome	79493
RareCareNet	Acinar cell carcinoma of pancreas	424046
	Adenocarcinoma of anal canal	424016
	Adenocarcinoma of gallbladder and extrahepatic biliary tract	424991
	Adenocarcinoma of liver and intrahepatic biliary tract	424943
	Biliary cystadenocarcinoma	424982
	Carcinoma of anal canal	424013
	Carcinoma of esophagus, salivary gland type	418945
	Carcinoma of liver and intrahepatic biliary tract	424936
	Carcinoma of stomach, salivary gland type	423781
	Epithelial tumor of anal canal	424010
	Hereditary gastric cancer	423776
	Inherited digestive cancer-predisposing syndrome	425003
	Intraductal papillary mucinous carcinoma of pancreas	424058
	Mucinous cystadenocarcinoma of pancreas	424053
	Neuroendocrine tumor of small intestine	423975
	Osteoclastic giant cell tumor of pancreas	424080
	Rare carcinoma of small intestine	423957
	Rare carcinoma of stomach	423771
	Rare epithelial tumor of colon	423991
	Rare epithelial tumor of liver and intrahepatic biliary tract	424933
	Rare epithelial tumor of pancreas	424033

Expert reviewer	Disease name	Orphanumber
RareCareNet	Rare epithelial tumor of rectum	423998
	Rare epithelial tumor of small intestine	425368
	Rare tumor of small intestine	423793
	Serous cystadenocarcinoma of pancreas	424073
	Solid pseudopapillary carcinoma of pancreas	424065
	Squamous cell carcinoma of anal canal	424019
	Squamous cell carcinoma of colon	423994
	Squamous cell carcinoma of gallbladder and extrahepatic biliary tract	424996
	Squamous cell carcinoma of liver and intrahepatic biliary tract	424975
	Squamous cell carcinoma of pancreas	424039
	Squamous cell carcinoma of rectum	424002
	Squamous cell carcinoma of small intestine	423968
	Squamous cell carcinoma of stomach	418959
	Undifferentiated carcinoma of esophagus	418951
	Undifferentiated carcinoma of liver and intrahepatic biliary tract	424970
	Undifferentiated carcinoma of stomach	423786
Dr Agnes RENNER	Gyrate atrophy of choroid and retina	414
Pr Pascal RICHETTE	Mild phosphoribosylpyrophosphate synthetase superactivity	411536
	Severe phosphoribosylpyrophosphate synthetase superactivity	411543
Dr Marlène RIO	3C syndrome	7
Dr Henry ROSENBERG	Malignant hyperthermia	423
Dr R.M.H. [Rudi] ROUMEN	Anterior cutaneous nerve entrapment syndrome	51890
Dr Russell SANETO	Alpers-Huttenlocher syndrome	726
Dr Fernando SANTOS SIMARRO	Simpson-Golabi-Behmel syndrome	373
Ms Anja SCHIEMANN	Malignant hyperthermia	423
Dr Michiel SCHREUDER	Renal agenesis	411709
Dr Jeoffrey SEGURA	Herpes simplex encephalitis	1930
Dr Claire SHOVLIN	Pulmonary arteriovenous fistula	2038
Dr Caroline SILVE	Acrodysostosis	950
Pr Tom SOLOMON	Herpes simplex encephalitis	1930
Dr Nicola SPECCHIO	Rolandic epilepsy	1945
Dr Kathryn STOWELL	Malignant hyperthermia	423
Dr Jair Antonio TENORIO CASTAÑO	Simpson-Golabi-Behmel syndrome	373
Dr Stanislav TOLKACHJOV	Progressive hemifacial atrophy	1214
Dr Megha TOLLEFSON	Progressive hemifacial atrophy	1214
Dr Emilie TOURNIER	Erythrokeratoderma variabilis progressiva	308166
Dr Anne TRISTAN	Bacterial toxic-shock syndrome	36234

Expert reviewer	Disease name	Orphanumber
Dr Anne TRISTAN	Staphylococcal toxic-shock syndrome	99919
	Streptococcal toxic-shock syndrome	99918
Dr Marina TRIVISANO	Rolandic epilepsy	1945
Pr Anna TYLKI-SZYMANSKA	Adenylosuccinate lyase deficiency	46
Pr Bjarne UDD	Distal myopathy	599
	Distal myopathy, Welander type	603
	Miyoshi myopathy	45448
Dr Christelle VAULOUP-FELLOUS	Congenital enterovirus infection	292
	Congenital rubella syndrome	290
Dr Jaime VAZQUEZ-JIMENEZ	Cardiac diverticulum	1686
Pr Alain VERLOES	ARX-related encephalopathy-brain malformation spectrum	423655
Dr Corinne VIGOUROUX	Familial partial lipodystrophy	98306
	Familial partial lipodystrophy, Dunnigan type	2348
Pr Ronald WANDERS	Refsum disease	773
Dr Zbigniew WSZOLEK	Dentatorubral pallidoluysian atrophy	101
Dr Andreas ZANKL	Multicentric osteolysis-nodulosis-arthropathy spectrum	371428
Dr Abraham ZLOTOGORSKI	Faisalabad histiocytosis	254707
	Familial sinus histiocytosis with massive lymphadenopathy	254712
	H syndrome	168569
	Pigmented hypertrichosis with insulin dependent diabetes mellitus syndrome	254723

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

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