Orphanet Report Series

Rare Diseases collection

2019

Expert reviewers for Orphanet in 2019

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MINISTÈRE DES AFFAIRES SOCIALES ET DE LA SANTÉ

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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 2019.

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 2019

Expert's name	Disease name	ORPHA code
Dr ABRUZZO Michael	Abruzzo-Erickson syndrome	921
Pr AHMED Faisal	46,XX testicular disorder of sex development	393
	46,XX ovotesticular disorder of sex development	2138
Dr ANGLES-CANO Eduardo	Congenital plasminogen activator inhibitor type 1 deficiency	465
Dr ARNAUD Eric J.	Pfeiffer syndrome type 3	93260
Pr ARNULF Isabelle	Kleine-Levin syndrome	33543
Pr AROCK Michel	Indolent systemic mastocytosis	98848
	Smoldering systemic mastocytosis	158775
	Systemic mastocytosis with associated hematologic neoplasm	98849
	Systemic mastocytosis	2467
Pr AUDO Isabelle	Cone rod dystrophy	1872
Pr BADENS Catherine	Syndromic diarrhea	84064
Pr BAHI-BUISSON Nadia	FOXG1 syndrome	561854
Dr BANNEAU Guillaume	Hereditary spastic paraplegia	685
Pr BARGY Frédéric	Annular pancreas	675
Dr BARRAGÁN GONZÁLEZ Eva	Acute promyelocytic leukemia	520
Dr BASSON Craig	Holt-Oram syndrome	392
Pr BECKERS Albert	X-linked acrogigantism	300373
	Familial isolated pituitary adenoma	314777
Dr BELLAÏCHE Marc	Chronic intestinal pseudoobstruction	2978
Dr BEN YAOU Rabah	Emery-Dreifuss muscular dystrophy	261
Dr BENLIAN Pascale	Dysbetalipoproteinemia	412
Pr BERTHERAT Jérôme	Acute adrenal insufficiency	95409
Pr BIEN Ewa	Pancreatoblastoma	677
Dr BÍKENKAMP Arend	Oculocerebrorenal syndrome of Lowe	534
Dr BLOISE Raffaella	Catecholaminergic polymorphic ventricular tachycardia	3286
	Brugada syndrome	130
Dr BOBER Michael	Microcephalic osteodysplastic primordial dwarfism type II	2637
Dr BOCCHERINI Chiara	Pfeiffer syndrome	710

Pr BOCKENHAUER Detlef	Gitelman syndrome	358
	Pseudohypoaldosteronism type 2	757
Pr BODEMER Christine	Infantile myofibromatosis	2591
Dr BOLLEE Guillaume	Adenine phosphoribosyltransferase deficiency	976
Pr BOLTSHAUSER Eugen	Hoyeraal-Hreidarsson syndrome	3322
Dr BONNE Gisþle	Emery-Dreifuss muscular dystrophy	261
Dr BOTTANI Armand	Cornelia de Lange syndrome	199
Dr BOUVATTIER Claire	Acute adrenal insufficiency	95409
Pr BRANDI Maria Luisa	Acrodysostosis	950
Pr BRENNAN Bernadette	Pancreatoblastoma	677
Pr BRON Alain	Nanophthalmos	35612
Dr BROSCO Jeffrey	Histidinemia	2157
Dr BUCELLI Robert	CANOMAD syndrome	71279
Ms BUCHANAN Catherine	48,XXYY syndrome	10
Dr BURTON Barbara	Mucopolysaccharidosis type 2	580
Pr CADRANEL Jacques	Primary pulmonary lymphoma	2420
Ms CAMMANN Victoria Lucia	Tako-Tsubo cardiomyopathy	66529
Dr CAMPEAU Philippe	DOORS syndrome	79500
Pr CAROFF STANLEY	Neuroleptic malignant syndrome	94093
Dr CASTORI Marco	Hypermobile Ehlers-Danlos syndrome	285
	Cooks syndrome	1487
Dr CATANEO Daniele	Congenital lobar emphysema	1928
Dr CESCA Laura	Primary membranous glomerulonephritis	97560
	HANAC syndrome	73229
	Oncogenic osteomalacia	352540
	Granulomatosis with polyangiitis	900
Dr CEYNOWA Dylan	Morning glory disc anomaly	35737
Pr CHABRIAT Hugues	CADASIL	136
Pr CHANDRAN Suresh	Syndromic diaphragmatic or abdominal wall malformation	108979
Pr CHANSON Philippe	Acromegaly	963
Pr CHAPURLAT Roland	Fibrous dysplasia of bone	249
Pr CHARRON Philippe	Arrhythmogenic right ventricular cardiomyopathy	247
Pr CIMAZ Rolando	Cogan syndrome	1467
Pr COLOMBI Marina	Arterial tortuosity syndrome	3342
Pr COTTIN Vincent	Mounier-Kühn syndrome	3347
	Idiopathic pulmonary fibrosis	2032
	Idiopathic chronic eosinophilic pneumonia	2902
	Lymphangioleiomyomatosis	538
Ms CROFT Joanne	Systemic primary carnitine deficiency	158
Pr CROSS Helen	CDKL5-related epileptic encephalopathy	505652

Dr CUISSET Jean-Marie	Proximal spinal muscular atrophy type 1	83330
	Proximal spinal muscular atrophy type 2	83418
	Proximal spinal muscular atrophy type 3	83419
	Proximal spinal muscular atrophy type 4	83420
Dr CUNNIFF Christopher	Bloom syndrome	125
Pr CZAUDERNA Piotr	Pediatric hepatocellular carcinoma	33402
Pr DALY Adrian	X-linked acrogigantism	300373
	Familial isolated pituitary adenoma	314777
	X-linked acrogigantism	300373
Dr D'AMBROSIO Valentina	Pfeiffer syndrome	710
Dr DAVIS Shanlee	48,XXYY syndrome	10
Dr DE SANDRE-GIOVANNOLI Annachiara Aurora	Restrictive dermopathy	1662
Pr DE LA PEÐA	Congenital plasminogen activator inhibitor type 1 deficiency	465
Dr DE PROST Nicolas	Toxic epidermal necrolysis	537
Dr de VRIES Annelou	Testicular regression syndrome	983
Dr DELLO STROLOGO Luca	Cystinuria	214
Dr DENJOY Isabelle	Familial short QT syndrome	51083
Dr DIENE GwenaÙlle	Prader-Willi syndrome	739
Pr DONADIEU Jean	Langerhans cell histiocytosis	389
Dr DONADILLE Bruno	Rare hyperlipidemia	181422
	Familial hypoaldosteronism	427
Pr DOZ Franþois	Retinoblastoma	790
Ms DUKER Angela	Microcephalic osteodysplastic primordial dwarfism type II	2637
Dr DUMAN Ozgur	Spontaneous periodic hypothermia	29822
Pr DUPIN Nicolas	Kaposi sarcoma	33276
Dr DUPUIS-GIROD Sophie	Hereditary hemorrhagic telangiectasia	774
Dr DUREAU Pascal	Juvenile glaucoma	98977
Dr DYMENT David	SHORT syndrome	3163
Dr EL-HATTAB Ayman	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency	279934
Dr ENGELEN M. [Marc]	Refsum disease	773
Dr ERICKSON Robert	Abruzzo-Erickson syndrome	921
Dr ESPINËS Carmen	Mitochondrial membrane protein-associated neurodegeneration	289560
Dr EWENCZYK Claire	Hereditary spastic paraplegia	685
Dr FABRE Alexandre	Syndromic diarrhea	84064
Pr FAUTREL Bruno	Adult-onset Still disease	829
Pr FERNANDEZ-ALVAREZ Emilio	Benign paroxysmal torticollis of infancy	71518

Dr FERREIRA Carlos R.	Triglyceride deposit cardiomyovasculopathy	565612
	NAD(P)HX dehydratase deficiency	555402
Pr FEURLE Gerhard	Whipple disease	3452
Pr FILLA Alessandro	Early-onset cerebellar ataxia with retained tendon reflexes	1177
Pr FINSTERER Josef	Kearns-Sayre syndrome	480
Ms FLANAGAN Maeve	Bloom syndrome	125
Dr FOÏS Eléna	Sickle cell anemia	232
Dr FORZANO Francesca	Gollop-Wolfgang complex	1986
Pr FOSTER Paul	Nanophthalmos	35612
Dr FOUDRINOY Sylvie	Hereditary hemorrhagic telangiectasia	774
Dr FUJIOKA Shinsuke	Spinocerebellar ataxia type 11	98767
	Spinocerebellar ataxia type 26	101112
Pr G WAXMAN Stephen	Paroxysmal extreme pain disorder	46348
Pr GALACTEROS Frédéric	Sickle cell anemia	232
Dr GANDJBAKHCH Estelle	Arrhythmogenic right ventricular cardiomyopathy	247
Dr GARCIA-MERIC Patricia	Congenital toxoplasmosis	858
Dr GARCIA-SANTIBANEZ ROCIO	CANOMAD syndrome	71279
Dr GELB Bruce	Char syndrome	46627
Ms GIANCOTTI Antonella	Pfeiffer syndrome	710
Dr GIANI Teresa	Cogan syndrome	1467
Dr GIANSILY-BLAIZOT Muriel	Congenital factor VII deficiency	327
Dr GIRERD Barbara	Pulmonary veno-occlusive disease and/or pulmonary capillary haemangiomatosis	431353
Pr GIRSCHICK Hermann	SAPHO syndrome	793
Dr GIURGEA Irina	Goldberg-Shprintzen megacolon syndrome	66629
Pr GOADSBY Peter	Paroxysmal hemicrania	157835
Pr GOIZET Cyril	Hereditary spastic paraplegia	685
Dr GOLDENBERG Alice	Cornelia de Lange syndrome	199
Pr GONZÁLEZ SARMIENTO Rogelio	X-linked dominant chondrodysplasia punctata	35173
Dr GOSPE Sidney	Pyridoxine-dependent epilepsy	3006
Pr GOTTRAND Frédéric	Esophageal atresia	1199
Pr GOUDEMAND Jenny	Von Willebrand disease	903
Pr GOZES Illana	ADNP syndrome	404448
Dr GREIG Aina	Pfeiffer syndrome type 3	93260
Dr GRIPP Karen	Costello syndrome	3071
Dr GUERRIER Daniel	Mayer-Rokitansky-K¸ster-Hauser syndrome type 2	2578
Dr GUIGNAT Laurence	Acute adrenal insufficiency	95409
Dr GUPTA Sangeeta	Umbilical cord ulceration-intestinal atresia syndrome	3405

Pr HÄBERLE Johannes	Argininosuccinic aciduria	23
	Carbamoyl-phosphate synthetase 1 deficiency	147
	Ornithine transcarbamylase deficiency	664
	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	415
Pr HAMM Henning	Aplasia cutis congenita	1114
Dr HAMM Austin	Pfeiffer syndrome type 3	93260
Ms HARDING Philippa	Isolated microphthalmia-anophthalmia-coloboma	2542
Pr HARTNETT Mary Elizabeth	Retinopathy of prematurity	90050
Dr HARVENGT Julie	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	293987
Dr HAYWARD Catherine	Quebec platelet disorder	220436
Dr HERVE Dominique	CADASIL	136
Dr HIDDEN-LUCET Françoise	Arrhythmogenic right ventricular cardiomyopathy	247
Dr HINSON Vanessa	Psychogenic movement disorders	71519
Dr HIRANO Ken-ichi	Triglyceride deposit cardiomyovasculopathy	565612
Pr HIROSE Shinichi	Autosomal dominant nocturnal frontal lobe epilepsy	98784
Pr HOLLAK C.E.M. [Carla]	Refsum disease	773
Dr HOTZ Claire	Neurofibromatosis type 1	636
Pr HOUYEL Lucile	Coronary artery congenital malformation	1081
Dr HOWELL Susan	48,XXYY syndrome	10
Pr HUGOT Jean-Pierre	Chronic intestinal pseudoobstruction	2978
Dr INJETI Gnanavelu	Marden-Walker syndrome	2461
Dr INNES A.Micheil	SHORT syndrome	3163
Pr JEUNEMAITRE Xavier	Apparent mineralocorticoid excess	320
Pr JOUNEAU Stéphane	Idiopathic pulmonary fibrosis	2032
Dr KÄLVIÄINEN Reetta	CDKL5-related epileptic encephalopathy	505652
Dr KAMM Christoph	Early-onset generalized limb-onset dystonia	256
Pr KAUFMANN Horacio	Familial dysautonomia	1764
	Hereditary sensory and autonomic neuropathy type 4	642
	Hereditary sensory and autonomic neuropathy type 2	970
Dr KERJOUAN Mallorie	Idiopathic pulmonary fibrosis	2032
Dr KIEFFER François	Congenital toxoplasmosis	858
Dr KIRITSI Dimitra	CHILD syndrome	139
Pr KNOWLES Michael	Primary ciliary dyskinesia	244
Dr KOOLEN D.A. [David]	Koolen-De Vries syndrome	96169
Pr KOOY Frank	ADNP syndrome	404448

Dr KURAHASHI Hirokazu	Autosomal dominant nocturnal frontal lobe epilepsy	98784
Pr LACOMBE Didier	Floating-Harbor syndrome	2044
Pr LAMBERT Marc	Takayasu arteritis	3287
	Takayasu arteritis	3287
Dr LANGEVELD M. [Mirjam]	Refsum disease	773
Pr LAUGEL Vincent	Proximal spinal muscular atrophy type 1	83330
	Proximal spinal muscular atrophy type 2	83418
	Proximal spinal muscular atrophy type 3	83419
	Proximal spinal muscular atrophy type 4	83420
Dr LAURENT Lucie	Hereditary chronic pancreatitis	676
Pr LAZARUS John	Bamforth-Lazarus syndrome	1226
Dr LEDERER Damien	Pai syndrome	1993
Pr LEGER Juliane	Acute adrenal insufficiency	95409
Pr LEGIUS Eric	Legius syndrome	137605
Dr LEJEUNE Stéphanie	Esophageal atresia	1199
Dr LESLIE Elizabeth	Autosomal dominant popliteal pterygium syndrome	1300
Dr LETURCQ France	Emery-Dreifuss muscular dystrophy	261
Pr LEVADE Thierry	Farber disease	333
Pr LEVY Philippe	Hereditary chronic pancreatitis	676
Dr LIEBAU Max	Autosomal recessive polycystic kidney disease	731
	Renal nutcracker syndrome	71273
Dr LIPSANEN-NYMAN Marita	Mulibrey nanism	2576
Pr LIPSKA-ZIETKIEWICZ Beata	Denys-Drash syndrome	220
	Frasier syndrome	347
Pr LO MUZIO Lorenzo	Gorlin syndrome	377
Dr LOMBARDI Maria Paola	Focal dermal hypoplasia	2092
Dr LONGONI Mauro Miguel	Donnai-Barrow syndrome	2143
Dr LOPEZ DE HEREDIA	Wolfram syndrome	3463
Dr MAFFEI Pietro	Alstr÷m syndrome	64
Dr MAGGI Lorenzo	Thomsen and Becker disease	614
Pr MAIER Lisa	Chronic beryllium disease	133
Dr MALFAIT Fransiska	Hypermobile Ehlers-Danlos syndrome	285
	Vascular Ehlers-Danlos syndrome	286
	Ehlers-Danlos syndrome	98249
Dr MANTEGAZZA Renato	Thomsen and Becker disease	614
Dr MARCIANO Emmanuel	Juvenile glaucoma	98977
Pr MARINO Bruno	Abnormal origin of right or left pulmonary artery	99050

	from the aorta	
Dr MARLIN Sandrine	Non-syndromic genetic deafness	87884
	Auditory neuropathy-optic atrophy syndrome	542585
	Optic atrophy-ataxia-peripheral neuropathy-global developmental delay syndrome	543470
Dr MARUANI Anna	Monosomy 22q13.3	48652
Pr MATSUMOTO Naomichi	Coffin-Siris syndrome	1465
Dr MATUSIK Paweł T.	Familial sick sinus syndrome	166282
Pr MAZEREEUW-HAUTIER Juliette	Superficial epidermolytic ichthyosis	455
Pr MCELREAVEY Kenneth	46,XX testicular disorder of sex development	393
	46,XX ovotesticular disorder of sex development	2138
Dr MCGOWAN Ruth	46,XX testicular disorder of sex development	393
	46,XX ovotesticular disorder of sex development	2138
Dr MENKE L.A. [Leonie]	Rubinstein-Taybi syndrome	783
Dr MENSAH Sandrine	Sickle cell anemia	232
Dr MERLINI Luciano	Congenital fiber-type disproportion myopathy	2020
Dr MITROVIC Stéphane	Adult-onset Still disease	829
Pr MONTANI David	Pulmonary veno-occlusive disease and/or pulmonary capillary haemangiomatosis	431353
Pr MOOG Ute	Oculocerebrocutaneous syndrome	1647
Pr MOORE Tony	Oligocone trichromacy	75378
Dr MOOS Verena	Whipple disease	3452
Dr MOOSAJEE Mariya	Isolated microphthalmia-anophthalmia-coloboma	2542
Dr MORCEL Karine	Mayer-Rokitansky-K¸ster-Hauser syndrome type 2	2578
Dr MURAWSKI Maciej	Pediatric hepatocellular carcinoma	33402
Pr NABBOUT-TARANTINO Rima	CDKL5-related epileptic encephalopathy	505652
	SYNGAP1-related developmental and epileptic encephalopathy	544254
	FOXG1 syndrome	561854
Dr NASSER Mouhamad	Mounier-Kühn syndrome	3347
Dr NIJENHUIS Tom	Oncogenic osteomalacia	352540
Dr NITSCHKE Yvonne	Generalized arterial calcification of infancy	51608
Dr NORIS Marina	Fibronectin glomerulopathy	84090
Dr NUNES MARTÍNEZ Virginia	Wolfram syndrome	3463
Dr OKAMOTO Nobuhiko	Coffin-Siris syndrome	1465
Dr OLPIN Simon	Systemic primary carnitine deficiency	158
Dr ORBACH Daniel	Infantile myofibromatosis	2591
Dr ORO Saskia	Toxic epidermal necrolysis	537
Pr OTTMAN Ruth	Autosomal dominant epilepsy with auditory features	101046
Dr OWEN Katharine	MODY	552

Pr PALMA Jose-Alberto	Familial dysautonomia	1764
	Hereditary sensory and autonomic neuropathy type 4	642
	Hereditary sensory and autonomic neuropathy type 2	970
Dr PALMIO Johanna	Miyoshi myopathy	45448
Dr PEREIRA Elaine	Achondroplasia	15
	Thanatophoric dysplasia	2655
Pr PEYRON François	Congenital toxoplasmosis	858
Pr PIRACCINI Bianca Maria	Yellow nail syndrome	662
Dr PIZZUTI Antonio	Pfeiffer syndrome	710
Pr PLAISIER Emmanuelle	HANAC syndrome	73229
Dr POBER Barbara	Donnai-Barrow syndrome	2143
Pr PORFIRIO Berardino	Alkaptonuria	56
Pr POUGET Jean	Proximal spinal muscular atrophy type 1	83330
	Proximal spinal muscular atrophy type 2	83418
	Proximal spinal muscular atrophy type 3	83419
Dr PRADAT Pierre-François	Proximal spinal muscular atrophy type 4	83420
Pr PROBST Vincent	Familial progressive cardiac conduction defect	871
Dr R SCHULMAN Betsy	Paroxysmal extreme pain disorder	46348
Pr REBOURS Vinciane	Hereditary chronic pancreatitis	676
Dr RIPPERGER Tim	Familial platelet disorder with associated myeloid malignancy	71290
Dr RISHI Pukhraj	Eales disease	40923
Dr RITTER Alaina	Cholera	173
Dr RIZZO William	Sj÷gren-Larsson syndrome	816
Pr ROBERTSON Stephen	Larsen syndrome	503
	Atelosteogenesis type I	1190
	Spondylocarpotarsal synostosis	3275
	Atelosteogenesis type III	56305
Pr RODOLICO Carmelo	Scapuloperoneal spinal muscular atrophy	431255
Pr RONCO Pierre	Primary membranous glomerulonephritis	97560
Dr ROSENBERG Henry	Neuroleptic malignant syndrome	94093
Pr ROSENDAHL Jonas	Hereditary chronic pancreatitis	676
Pr RUMI Elisa	Primary myelofibrosis	824
Dr RUSHING Elisabeth	Focal myositis	48918
Pr RUTSCH Frank	Generalized arterial calcification of infancy	51608
Pr SAADOUN David	Behçet disease	117
Pr SAILLER Laurent Angel	Relapsing polychondritis	728
Dr SANZ ALONSO Miguel	Acute promyelocytic leukemia	520
Pr SASS Jörn Oliver	Propionic acidemia	35

Dr SAVAGE Sharon	Dyskeratosis congenita	1775
Pr SCHAEFER Franz	Congenital nephrotic syndrome, Finnish type	839
	Granulomatosis with polyangiitis	900
Dr SCHEUERLE Angela	Incontinentia pigmenti	464
Pr SCHLEGELBERGER Brigitte	Familial platelet disorder with associated myeloid malignancy	71290
Pr SCHUSTER Volker	Ligneous conjunctivitis	97231
Dr SCHUTTE Brian	Autosomal dominant popliteal pterygium syndrome	1300
Dr SCHYNS-LIHARSKA Tsveta	Alternating hemiplegia of childhood	2131
Dr SHARMA Aman Flore	Relapsing polychondritis	728
Dr SICRE DE FONTBRUNE	Paroxysmal nocturnal hemoglobinuria	447
Dr SLY William	Mucopolysaccharidosis type 7	584
Pr SMITH Richard	Pendred syndrome	705
Dr SNAPE Katie	Mosaic variegated aneuploidy syndrome	1052
Dr SPECCHIO Nicola	CDKL5-related epileptic encephalopathy	505652
Pr SUSEN Sophie	Von Willebrand disease	903
Pr SUTTON V. Reid	Aicardi syndrome	50
	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	1071
Dr TABET Anne-Claude	Monosomy 22q13.3	48652
Dr TAKSANDE Amar	Marden-Walker syndrome	2461
Pr TARNOPOLSKY Mark	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	713
Dr TARTAGLIA Nicole	48,XXYY syndrome	10
Pr TAUBER Maithé	Prader-Willi syndrome	739
	MAGEL2-related Prader-Willi-like syndrome	398069
Dr TEÄR FAHNEHJELM Kristina	Morning glory disc anomaly	35737
Pr TEMPLIN Christian	Tako-Tsubo cardiomyopathy	66529
Dr TEMPLIN-GHADRI Jelena-Rima	Tako-Tsubo cardiomyopathy	66529
Dr THIAGARAJAN Ravi	Hypoplastic left heart syndrome	2248
Pr TOBIAS Edward	46,XX testicular disorder of sex development	393
	46,XX ovotesticular disorder of sex development	2138
Dr TOURAINE Renaud	Down syndrome	870
Pr TOURNIER-LASSERVE Elisabeth	Familial cerebral cavernous malformation	221061
Pr TRANEBJAERG Lisbeth	Mohr-Tranebjaerg syndrome	52368
Dr TSUBOI Yoshio	Spinocerebellar ataxia type 11	98767
	Spinocerebellar ataxia type 26	101112
Pr UDD Bjarne	Distal myopathy, Welander type	603

Dr URTIZBEREA Andoni	Proximal spinal muscular atrophy type 1	83330
	Proximal spinal muscular atrophy type 2	83418
	Proximal spinal muscular atrophy type 3	83419
	Proximal spinal muscular atrophy type 4	83420
Pr VALLA Dominique	Isolated polycystic liver disease	2924
Pr VAN BOGAERT Patrick	Porencephaly	2940
Pr van STEENSEL Maurice Marjo	Birt-Hogg-Dubé syndrome	122
Dr VAN DER KNAAP	ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement	457375
Pr VANTYGHEM Marie-Christine	Multiple symmetric lipomatosis	2398
	Acquired partial lipodystrophy	79087
	Localized lipodystrophy	79088
Dr VAZQUEZ-LOPEZ Esther	Benign paroxysmal torticollis of infancy	71518
Pr VERLOES Alain	Bardet-Biedl syndrome	110
Dr VERSCHUEREN Annie	Proximal spinal muscular atrophy type 4	83420
Dr VIGNES Stéphane	Primary intestinal lymphangiectasia	90362
Pr VIGOUROUX Corinne	Berardinelli-Seip congenital lipodystrophy	528
Dr VILLAGOMEZ Adrianne	48,XXYY syndrome	10
Dr VIVARELLI Marina	Primary membranous glomerulonephritis	97560
Pr VIVES-CORRONS Joan Luis	Congenital amegakaryocytic thrombocytopenia	3319
Dr VOGT Julie	Lethal multiple pterygium syndrome	33108
Dr WADE Emma	Larsen syndrome	503
	Atelosteogenesis type I	1190
	Spondylocarpotarsal synostosis	3275
Pr WALLON Martine	Congenital toxoplasmosis	858
Dr WALSH Stephen	Acquired monoclonal Ig light chain-associated Fanconi syndrome	91136
	Oncogenic osteomalacia	352540
Dr WEI Diana	Paroxysmal hemicrania	157835
Pr WENDEL Udo	Hartnup disease	2116
Pr WETZELS J.F.M. [Jack]	Primary membranous glomerulonephritis	97560
Pr WOLKENSTEIN Pierre	Neurofibromatosis type 1	636
Pr WOODLEY David	Epidermolysis bullosa acquisita	46487
Dr WSZOLEK Zbigniew	Spinocerebellar ataxia type 11	98767
	Spinocerebellar ataxia type 26	101112
Dr ZANKL Andreas	Multicentric osteolysis-nodulosis-arthropathy spectrum	371428
Pr ZARIWALA Maimoona	Primary ciliary dyskinesia	244
Pr ZLOTOGORSKI Abraham	H syndrome	168569

List of expert networks reviewing Orphanet data in 2019

Name of expert network	Disease name	ORPHA code
European Rare Kidney Diseases Reference Network - ERKNet	Denys-Drash syndrome	220
	Frasier syndrome	347
	Cystinuria	214
	Acquired monoclonal Ig light chain-associated Fanconi syndrome	91136
	Fibronectin glomerulopathy	84090
	Congenital nephrotic syndrome, Finnish type	839
	Primary membranous glomerulonephritis	97560
	Pseudohypoaldosteronism type 2	757
	HANAC syndrome	73229
	Oncogenic osteomalacia	352540
	Autosomal recessive polycystic kidney disease	731
	Renal nutcracker syndrome	71273
	Granulomatosis with polyangiitis	900
European Reference Network on Rare and Complex Epilepsies - EpiCARE	CDKL5-related epileptic encephalopathy	505652
European Reference Network on Rare Multisystemic Vascular Diseases - VASCERN	Hereditary hemorrhagic telangiectasia	774

Editor-in-chief: Ana Rath – Editor of the report: Charlotte Rodwell
Contributing editors: Valérie Lanneau,
Annie Olry & Dam-Thi Tsuvaltsidis
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