Orphanet Report Series

Rare Diseases collection

2018

Expert reviewers for Orphanet in 2018

Inserm
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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 2018.

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 2018

Expert's name	Disease name	ORPHA number
Dr Margaret ADAM	Kabuki syndrome	2322
	Mowat-Wilson syndrome	2152
	ATTRV30M amyloidosis	85447
Pr Isabelle ARNULF	Kleine-Levin syndrome	33543
Dr Amin BARAKAT	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	2237
Dr Genevieve BAUJAT	Brachytelephalangic chondrodysplasia punctata	79345
Dr Rabah BEN YAOU	Emery-Dreifuss muscular dystrophy	261
Pr Dominique BREMOND-GIGNAC	Oculocerebrorenal syndrome of Lowe	534
Pr Brigitte CHABROL	MELAS	550
Dr Geneviève DE SAINT-BASILE - CHAZELAS	Griscelli syndrome	381
Dr Sylvie DELANIAN	Radiation-induced disorder	521132
Dr Chantal FARRA	Dubowitz syndrome	235
Dr Florence FELLMANN	Partial chromosome Y deletion	1646
Dr Joost FRENKEL	Mevalonate kinase deficiency	309025
Dr Toshiyuki FUKAO	Succinyl-CoA:3-ketoacid CoA transferase deficiency	832
Dr Alice GOLDENBERG	Cornelia de Lange syndrome	199
Pr Eric HACHULLA	Limited cutaneous systemic sclerosis	220402
	Diffuse cutaneous systemic sclerosis	220393
	Systemic sclerosis	90291
	CREST syndrome	90290
Dr Dominique HERVE	Moyamoya disease	2573
Dr Robert KLAASSEN	Immune thrombocytopenic purpura	3002
	Ehlers-Danlos syndrome, arthrochalasis type	1899
Dr Alfried KOHLSCHUTTER	Late infantile neuronal ceroid lipofuscinosis	168491
Dr Thierry LEBLANC	Blackfan-Diamond anemia	124
Pr Nicolas LEBOULANGER	Choanal atresia	137914
	Choanal atresia, bilateral	137920
	Choanal atresia, unilateral	137917
Dr Sylvie MANOUVRIER	Holt-Oram syndrome	392
Dr Juliette MAZEREEUW-HAUTIER	Peeling skin syndrome type B	263553
Pr Eneida MIOSHI	Amyotrophic lateral sclerosis	803
Dr Bénédicte NEVEN	CINCA syndrome	1451
Dr Johanna PALMIO	GNE myopathy	602

	Vocal cord and pharyngeal distal myopathy	600
	Distal nebulin myopathy	399103
	Distal anoctaminopathy	399096
Dr Juha PALONEVA	Nasu-Hakola disease	2770
Dr Véronique PINGAULT	Waardenburg-Shah syndrome	897
	Peripheral demyelinating neuropathy-central dysmyelinating leukodystrophy-Waardenburg syndrome-Hirschsprung disease	163746
Dr Aurelia POUJOIS	Wilson disease	905
Dr Rogelio Gonzales SARMIENTO	X-linked dominant chondrodysplasia punctata	35173
Dr Angela SCHEUERLE	Incontinentia pigmenti	464
Dr Connie SCHRANDER STUMPEL	Kabuki syndrome	2322
Pr Juan Antonio TOVAR LARRUCEA	Congenital diaphragmatic hernia	2140
Pr Bjarne UDD	GNE myopathy	602
	Vocal cord and pharyngeal distal myopathy	600
	Distal nebulin myopathy	399103
	Distal anoctaminopathy	399096
Dr Andoni URTIZBEREA	Facioscapulohumeral dystrophy	269
	Bethlem myopathy	610
Dr Stephane VIGNES	Primary lymphedema	77240
Dr France WOIMANT	Wilson disease	905
Dr Alberto ZANELLA	Hereditary elliptocytosis	288
Pr Johannes ZSCHOCKE	HSD10 disease	391417

List of expert networks reviewing Orphanet data in 2018

Expert group	Disease name	ORPHA number
European Reference Network on Rare Multisystemic Vascular Diseases - VASCERN	Hereditary hemorrhagic telangiectasia	774
MARIH - Filière de santé maladies rares : maladies rares immuno-hématologiques	Non-histaminic angioedema	658
European Reference Network on Transplantation in Children - TransplantChild	Rare disorder potentially indicated for transplant	506207
European Reference Network on Rare Eye Diseases - ERN-EYE	Rare ophthalmic disorder	97966
FIRENDO - Filière de santé maladies rares : maladies rares endocriniennes	Pediatric-onset Graves disease	525731
	Prepubertal anorexia nervosa	525738
European Rare Kidney Diseases Reference Network - ERKNet	Rare renal disease	93665
European Reference Network on Rare craniofacial anomalies and ENT disorders - ERN CRANIO	Cleft lip with or without cleft palate	1991
	Cleft palate	2014
BRAIN-TEAM - Filière de santé maladies rares : maladies rares à expression motrice ou cognitive du système nerveux central	Narcolepsy type 1	2073
	Narcolepsy type 2	83465
	Idiopathic hypersomnia	33208
FAI2R - Filière de santé maladies rares : maladies auto-immunes et auto-inflammatoires systémiques rares	Systemic lupus erythematosus	536
FIMARAD - Filière de santé maladies rares : maladies rares en dermatologie	Erythema multiforme major	502499
	Ichthyosis	79354

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