D3.1 Mid-term Report on Genetic Annotations, ORPHAcodes alignments with other terminologies, and newly produced or updated texts Report

June 2024

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Content summary

Introduction

The main scope of OD4RD2 Work Package 3 (WP3) is to improve the Orphanet scientific information collected around the Nomenclature of rare diseases, either as a continuous internal updating activity or in collaboration with European Reference Networks, to reflect the evolution of knowledge.

This summary report will provide an overview of the main activities from April 2023 to April 2024 and it will contain 3 sections:

1) New discoveries around genes related to RD and their representation in the Orphanet database, with links to where these relationships are available for reuse;

2) RD coverage in target terminologies and the relationships between ORPHAcodes to each of the target terminologies, with links to where these alignments are available for reuse;

3) A list of newly produced definitions and of newly produced or updated RD summary information, with information on whether the text was realized in collaboration with ERNs.

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1. New discoveries around genes related to RD

Methodology

Updates of the gene-diseases relationships database are regularly performed by the Orphanet gene project manager according to a methodology published here (link). Updates in the gene-disease database can also result in a revision of the Orphanet nomenclature (addition or modification of an ORPHAcode) and follows the methodology for updating the Orphanet nomenclature. If these updates impact the revision done in collaboration with ERNs, the appropriate connections are done to avoid parallel revisions.

Data report

The following indicators were calculated for the period going from April the 1st 2023 to April the 30th 2024.

Cumulative number of genes linked to rare diseases with a disease-causing relationship	**4,606 (+144)**
Newly created disease-causing genes (i.e. genes were absent from Orphanet and were created with a disease-causing relationship)	**66 (+5)**
New disease-causing relationships attributed to unknowingly disease-causing genes (i.e. genes were present in Orphanet as parts of diagnostic panels and were subsequently identified as disease-causing)	**No more pertinent**
Newly added disease-causing relationships attributed to knowingly disease-causing genes (i.e. genes already present in Orphanet linked to other diseases as disease-causing)	**225 (+95)**
Modification of gene-disease relationships (e.g. disease-causing germline mutation was updated to disease-causing germline mutation loss of function)	**8 (+1)**
Suppressed gene-disease relationships	**218 (+99)**

Table 1: List of genetic indicators together with their value for the period considered. The increase in value compared to the previous period (April 2022-March 2023) is reported in parentheses. Of note, the indicator "New disease-causing relationships attributed to unknowingly disease-causing genes" is no longer pertinent due to a change in the diagnostic tests data model.

The complete Orphanet dataset of genes associated with rare diseases and their relationships is generated every 6 months (July and November) and available for download and exploitation in the Orphadata website, OrphadataScience (Orphanet Scientific Knowledge Base section, recognized Global Core Biodata Resource) (link).

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2. RD coverage in target terminologies

Methodology

Alignment of ORPHAcodes with other terminologies in use in health systems and in registries is regularly performed by the Orphanet alignments project manager under the supervision of the Orphanet Scientific Director. The objectives of the OD4RD project were axed on four different terminologies, with different advancement statuses:

- ✓ Complete and maintain the alignment between ORPHAcodes and ICD-10 codes according to the rules already established and published here (link). This process implies the identification of the best suitable ICD-10 code and the definition of a semantic relationship between both codes;

- ✓ Continue and deliver for the first time the alignments between ORPHAcodes and ICD-11, in the frame of the participation of Inserm, US14-Orphanet to the French WHO collaborating centre. Two complementary approaches are followed to complete the alignments and to identify the gaps in ICD-11 in terms of rare disease coverage: a) a syntactic approach, using language recognition algorithms performed by the Agence Numérique en Santé (ANS, lead of the WHO collaborating centre). Results of the application of these algorithms are curated by Orphanet and combined with b) a semantic approach, derived from the same rules used for ICD-10 alignments and needed to assess the proximity (exact or inexact) of both Orphanet and ICD-11 concepts.

- ✓ Continue the mapping exercise between ORPHAcodes and SNOMED-CT in the frame of the collaboration agreement in place between Orphanet and SNOMED International, in order to have a common release in October each year and containing new terms. This collaboration implies monthly follow-up meetings and exchange of information, in order to reach agreement on the concepts to be included in SNOMED-CT and mapped to ORPHAcodes. Orphanet releases a human-readable excel file with the updated mappings every year, and for the duration of the collaboration agreement.

- ✓ Continue the alignments between ORPHAcodes and OMIM concepts by capturing OMIM updates by running a script on a monthly basis, then manually curating the concepts in order to produce semantic relationships between concepts in both terminologies or update existing ones. This activity can also result in initiating a demand for updating the Orphanet nomenclature, and follows the methodology for updating the Orphanet nomenclature. Should these updates impact the revision done in collaboration with ERNs, the appropriate connections are done to avoid parallel revision.

Of note, outside the OD4RD project, a continuous effort is being made to align ORPHAcodes to other terminologies such as MeSH, UMLS, MedDRA, GARD and MONDO.

Data report

The following indicators were calculated for the period going from April the 1st 2023 to April the 30th 2024.

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ORPHAcodes aligned to ICD-10:	**Total:**	**7478**	
	Disorders:	6361	(99,7% aligned)
	Subtypes of disorders:	995	(96,6% aligned)
	Groups of disorders:	122	(5,6% aligned) *
ORPHAcodes aligned to ICD-11:	**Total:**	**5550**	
	Disorders:	4251	(66,6% aligned)
	Subtypes of disorders:	884	(85,8% aligned)
	Groups of disorders:	415	(19,2% aligned) *
ORPHAcodes aligned to SNOMED-CT:	**Total:**	**6552**	
	Disorders:	6016	(94,3% aligned)
	Subtypes of disorders:	305	(29,6% aligned)
	Groups of disorders:	231	(10,7% aligned) *
ORPHAcodes (genetic entities only) aligned to OMIM:	**Total:**	**4742**	
	Disorders:	4061	(88,2% aligned)
	Subtypes of disorders:	630	(80,5% aligned)
	Groups of disorders:	51	(3,8% aligned) *

*Please note that for group of disorders, Orphanet only aligns the exact concepts in all terminologies

Table 2: ORPHAcodes aligned to target terminologies. Percentages are calculated on the total of Orphanet active clinical entities, by classification level, for ICD-10, ICD-11 and SNOMED-CT, while they are calculated on the total of Orphanet active genetic clinical entities for OMIM.

The complete Orphanet dataset of RD coverage in target terminologies and the relationships between ORPHAcodes to each of the target terminologies is generated every 6 months (July and November, except for SNOMED-CT, that is released annually in October) and is available for download and exploitation in the Orphadata website, OrphadataScience (Orphanet Scientific Knowledge Base section) (link).

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3. Newly produced definitions and newly produced or updated RD summary information

Methodology

Production or update of textual information related to rare disorders either as a continuous internal updating activity or in collaboration with European Reference Networks. This activity follows a general methodology published here (link).
Collaborations can be established with ERNs in order to prioritize the common work to maintain up-to-date summary information on rare diseases in Orphanet, as well as the definitions that accompany every rare disorder and that are delivered together with the nomenclature to help assigning the right ORPHAcode to a patient.

The collaboration with ERNs follows these steps:

- ✓ Initiation of collaboration: there are three points of entry 1) following the finalization of a collaboration on the Orphanet nomenclature; 2) The ERN establishes contact for a large collaboration independently of a nomenclature collaboration; 3) an ERN is approached directly by the Orphanet Editorial team to update a selection of diseases;

- ✓ Identification of training required: for large collaborations an initial introduction to Orphanet along with Editorial training is useful. For smaller collaborations, experts are directly issued guidelines on the task and text content;

- ✓ Establishing a list of diseases and text type (definition quality control or production/update of a disease abstract) according to specialty and predefined set of priorities;

- ✓ Identification of suitable experts by the ERN coordinating team or working group leads. Diseases involving multiple specialties may require cross-ERN collaboration;

- ✓ Cycle of revision and curation of text(s) between expert(s) and Editorial team, resulting in the validation and publication of text(s)

- ✓ Maintenance of disease summary information (ideally every 3 years)

- ✓ Review and feedback of collaboration: given punctually throughout collaboration and at the end of a collaboration cycle.

This methodology has been established based on the previous ERN collaboration experience and is adaptable to the needs and resources of each network.

Data report

The following indicators were calculated for the period going from April the 1st 2023 to April the 30th 2024.

A total of 450 texts were newly produced, or updated (95 more than targetted). Among these texts, 128 were published abstracts reviewed by an expert and 224 were internally validated and published definitions.

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Among the 224 published definitions, 153 were newly produced, and 71 were updated/quality controlled. 39 definitions are currently under revision (Table 3).

Among the 128 published abstracts, 79 were published in collaboration with 18 different ERNs. In total 107 abstracts were updated and 21 were newly produced. Currently 59 more abstracts are under revision, 32 of which are part of a collaborative work with ERNs (Table 4).

The complete list of newly produced or updated RD summary information (with a focus on those realized in collaboration with ERNs) and of newly produced definitions is available in Annex 2.

Indicators (01.04.2023-30.04.2024)		**Total**		
Total number of produced texts				**450**
Published Abstracts	New abstracts	21 (19 ERN)	128	
	Updated abstracts	107 (60 ERN)		
Ongoing abstracts	New abstracts	7 (6 ERN)	59	
	Updated abstracts	52 (26 ERN)		
Total number of abstracts produced				**187**
Published definitions	New definitions	153 (1 ERN)	224	
	Updated/Quality controlled definitions	71 (6 ERN)		
Ongoing definitions	New definitions	22 (6 ERN)	39	
	Updated/Quality controlled definitions	17 (2 ERN)		
Total number of definitions produced				**263**

Table 3: Summary of produced texts including published or ongoing abstracts and definitions either newly created or updated/QC controlled).

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ERN	Number of published new & updated abstracts in collaboration with ERNs	Ongoing collaborations with ERNs (in revision)
Total number if abstracts	**79**	**32**
ERN-Endo	9	4
EpiCARE	1	7
ERKNet	1	
ERN CRANIO	2	
GENTURIS	3	1
PaedCan	1	
PaedCan & MetabERN	1	
RITA	5	1
ERN-BOND	3	
ERN-Eye	2	
ERN-Skin	4	
EuroBloodNet	7	
eUROGEN	3	4
EURO-NMD	3	2
ITHACA	10	3
ITHACA & ERN-CRANIO	2	
MetabERN	20	2
VASCERN	1	5
ERN-Lung		1
ERN-RND		1
EURACAN		1

Table 4: Summary of the texts reviewed by an ERN-affiliated expert or part of an ongoing collaboration, divided by ERN

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Annex list of Abstracts and Definition made available during the project

Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Abstract	Published	163634	Maffucci syndrome	BOND	New
Abstract	Published	296	Ollier disease	BOND	Update
Abstract	Published	642788	Cushing syndrome due to cortisol-producing adrenocortical adenoma	ENDO-ERN	New
Abstract	Published	667	Autosomal recessive malignant osteopetrosis	BOND	Update
Abstract	Published	71277	Classic glucose transporter type 1 deficiency syndrome	EpiCare	Update
Abstract	Ongoing	64280	Childhood absence epilepsy	EpiCare	Update
Abstract	Ongoing	150	Nasopharyngeal carcinoma	EURACAN	Update
Abstract	Ongoing	1941	Juvenile absence epilepsy	EpiCare	Update
Abstract	Ongoing	1501	Adrenocortical carcinoma	ERN-Endo	Update
Abstract	Ongoing	86909	Myoclonic epilepsy of infancy	EpiCare	New
Abstract	Published	110	Bardet-Biedl syndrome	ERKNet, ERN Eye	Update
Abstract	Published	2377	Laurence-Moon syndrome	ERN EYE	Update
Abstract	Published	145	Hereditary breast and ovarian cancer syndrome	ERN GENTURIS	Update
Abstract	Published	144	Lynch syndrome	ERN GENTURIS	New
Abstract	Published	54595	Craniopharyngioma	ERN PaedCan	Update
Abstract	Published	2337	Diffuse palmoplantar keratoderma, Bothnian type	ERN SKIN	Update
Abstract	Ongoing	2841	Hailey-Hailey disease	_	Update
Abstract	Published	98907	Neutral lipid storage disease with ichythyosis	ERN SKIN	New
Abstract	Ongoing	79134	DEND syndrome	ERN-Endo	Update
Abstract	Published	96253	Cushing disease	ERN-Endo	Update
Abstract	Published	99889	Cushing syndrome due to ectopic ACTH secretion	ERN-Endo	Update
Abstract	Published	189427	Cushing syndrome due to bilateral macronodular adrenocortical disease	ERN-Endo	Update
Abstract	Published	641613	Endogenous Cushing syndrome	ERN-Endo	New
Abstract	Published	752	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	ERN-Endo	Update
Abstract	Published	1359	Carney complex	ERN-Endo	Update
Abstract	Ongoing	422	Idiopathic/heritable pulmonary arterial hypertension	ERN-Lung	New
Abstract	Published	69	Amyloidosis	ERN-RITA	Update
Abstract	Published	85451	ATTRV122I amyloidosis	ERN-RITA	Update
Abstract	Published	85447	ATTRV30M amyloidosis	ERN-RITA	Update
Abstract	Published	271861	Hereditary ATTR amyloidosis	ERN-RITA	New
Abstract	Published	100	Ataxia-telangiectasia	ERN-RITA	Update
Abstract	Published	98908	Neutral lipid storage myopathy	ERN-RND	New
Abstract	Ongoing	135	CACH syndrome	ERN-RND	Update

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Abstract	Published	893	WAGR syndrome	ERN EYE	New
Abstract	Published	79230	HJV or HAMP-related hemochromatosis	EuroBloodNet	Update
Abstract	Published	225123	TFR2-related hemochromatosis	EuroBloodNet	Update
Abstract	Published	52688	Myelodysplastic syndrome	EuroBloodNet	New
Abstract	Published	648569	Non-HFE-related hemochromatosis	EuroBloodNet	New
Abstract	Ongoing	93930	Bladder exstrophy	eUROGEN	Update
Abstract	Ongoing	93929	Cloacal exstrophy	eUROGEN	Update
Abstract	Published	48	Congenital bilateral absence of vas deferens	eUROGEN	Update
Abstract	Published	617	Congenital primary megaureter	eUROGEN	Update
Abstract	Ongoing	93928	Epispadias	eUROGEN	Update
Abstract	Ongoing	322	Exstrophy-epispadias complex	eUROGEN	Update
Abstract	Ongoing	64747	X-linked Charcot-Marie-Tooth disease	EURO-NMD	Update
Abstract	Published	610	Bethlem myopathy	_	Update
Abstract	Published	75840	Congenital muscular dystrophy, Ullrich type	_	Update
Abstract	Published	48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome	EURO-NMD	Update
Abstract	Published	54251	Corticosteroid-sensitive aseptic abscess syndrome	FAI2R	Update
Abstract	Ongoing	99886	Transient neonatal diabetes mellitus	ERN-Endo	Update
Abstract	Ongoing	36387	Generalized epilepsy with febrile seizures-plus	EpiCare	Update
Abstract	Ongoing	501	Lafora disease	EpiCare	New
Abstract	Ongoing	307	Juvenile myoclonic epilepsy	EpiCare	New
Abstract	Published	2836	PEHO syndrome	ITHACA	Update
Abstract	Published	1520	Craniofrontonasal dysplasia	ITHACA	Update
Abstract	Ongoing	293642	Blepharophimosis-intellectual disability syndrome	ITHACA	New
Abstract	Published	974	Adams-Oliver syndrome	ITHACA	Update
Abstract	Published	3378	Trisomy 13	ITHACA	Update
Abstract	Ongoing	3380	Trisomy 18	ITHACA	Update
Abstract	Published	3320	Thrombocytopenia-absent radius syndrome	ITHACA	Update
Abstract	Published	93	Aspartylglucosaminuria	MetabERN	Update
Abstract	Published	13	6-pyruvoyl-tetrahydropterin synthase deficiency	MetabERN	Update
Abstract	Published	238583	Hyperphenylalaninemia [due to tetrahydrobiopterin deficiency]	MetabERN	New
Abstract	Ongoing	2102	GTP cyclohydrolase I deficiency	MetabERN	Update
Abstract	Published	364	Glycogen storage disease due to glucose-6-phosphatase deficiency	MetabERN	Update
Abstract	Published	226	Dihydropteridine reductase deficiency	MetabERN	Update
Abstract	Published	882	Tyrosinemia type 1	MetabERN	Update
Abstract	Published	738	Porphyria	MetabERN	Update
Abstract	Published	95157	Acute hepatic porphyria	MetabERN	Update
Abstract	Published	79276	Acute intermittent porphyria	MetabERN	Update
Abstract	Published	79278	Autosomal erythropoietic protoporphyria	MetabERN	Update
Abstract	Published	79277	Congenital erythropoietic porphyria	MetabERN	Update
Abstract	Published	95159	Hepatoerythropoietic porphyria	MetabERN	Update

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Abstract	Published	79273	Hereditary coproporphyria	MetabERN	Update
Abstract	Published	101330	Porphyria cutanea tarda	MetabERN	Update
Abstract	Published	100924	Porphyria due to ALA dehydratase deficiency	MetabERN	Update
Abstract	Published	79473	Porphyria variegata	MetabERN	Update
Abstract	Published	443197	X-linked erythropoietic protoporphyria	MetabERN	New
Abstract	Published	394	Classic homocystinuria	MetabERN	Update
Abstract	Ongoing	141189	Cerebrofacial arteriovenous metameric syndrome	VASCERN	New
Abstract	Ongoing	53721	Spinal arteriovenous metameric syndrome	VASCERN	Update
Abstract	Ongoing	42775	PHACE syndrome	VASCERN	Update
Abstract	Ongoing	1053	Vein of Galen aneurysmal malformation	VASCERN	Update
Abstract	Published	115	Congenital contractural arachnodactyly	VASCERN	Update
Abstract	Published	34145	Immunoglobulin A nephropathy	ERKNet	New
Abstract	Ongoing	33572	5-oxoprolinase deficiency	_	Update
Abstract	Published	90289	Localized scleroderma	_	Update
Abstract	Published	166113	Bazex syndrome	_	Update
Abstract	Published	79138	Bickerstaff brainstem encephalitis	_	Update
Abstract	Published	98919	Miller Fisher syndrome	_	Update
Abstract	Published	122	Birt-Hogg-Dubé syndrome	_	Update
Abstract	Published	79140	Cutaneous neuroendocrine carcinoma	_	Update
Abstract	Ongoing	581	Mucopolysaccaridosis type 3	MetabERN	Update
Abstract	Ongoing	744	Proteus syndrome	_	Update
Abstract	Ongoing	1509	Coxopodopatellar syndrome	ITHACA	Update
Abstract	Published	47044	Hereditary papillary renal cell carcinoma	_	Update
Abstract	Ongoing	2929	Juvenile Polyposis Syndrome	Genturis	Update
Abstract	Published	65285	Lhermitte-Duclos disease	_	Update
Abstract	Published	2969	Proteus-like syndrome	_	Update
Abstract	Ongoing	59135	Laing early-onset distal myopathy	_	Update
Abstract	Ongoing	994	Fetal akinesia deformation sequence	_	Update
Abstract	Ongoing	2345	Isolated Klippel-Feil syndrome	_	Update
Abstract	Ongoing	33573	Gamma-glutamyl transpeptidase deficiency	_	Update
Abstract	Ongoing	33574	Gamma-glutamylcysteine synthetase deficiency	_	Update
Abstract	Ongoing	32	Glutathione synthetase deficiency	_	Update
Abstract	Published	97234	Glycogen storage disease due to phosphoglycerate mutase deficiency	_	Update
Abstract	Published	85202	Keutel syndrome		Update
Abstract	Published	647834	SLC40A1-related hemochromatosis	EuroBloodNet	New
Abstract	Published	98	Spastic ataxia charlevoix-saguenay type	_	Update
Abstract	Published	796	Sandhoff disease	MetabERN	Update
Abstract	Published	238606	Primary orthostatic tremor	_	Update
Abstract	Published	3198	Stiff person spectrum disorder	_	Update
Abstract	Published	845	Tay-Sachs disease	MetabERN	Update
Abstract	Ongoing	48652	Phelan-McDermid syndrome	_	Update
Abstract	Ongoing	498474	Hyaline fibromatosis syndrome	_	New

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Abstract	Published	400	Cystic echinococcosis	_	Update
Abstract	Ongoing	1926	Diabetic embryopathy	_	Update
Abstract	Published	1917	Fetal methylmercury syndrome	_	Update
Abstract	Published	591	Furuncular myiasis	_	Update
Abstract	Published	70593	Immunodeficiency due to selective anti-polysaccharide antibody deficiency		Update
Abstract	Ongoing	99885	Isolated permanent neonatal diabetes mellitus	ERN-Endo	Update
Abstract	Ongoing	2089	Glycogen storage disease due to hepatic glycogen synthase deficiency	_	Update
Abstract	Published	647815	Keratoendotheliitis fugax hereditaria	ERN-Eye	New
Abstract	Published	140944	CLOVES syndrome	ITHACA/CRANIO	Update
Abstract	Published	1646	Partial chromosome Y deletion	_	Update
Abstract	Published	699	Pearson syndrome	MetabERN/Paedcan-ERN	Update
Abstract	Published	90308	Klippel-Trénaunay syndrome	ITHACA/CRANIO	New
Abstract	Published	297	Tick-borne encephalitis	_	Update
Abstract	Published	28378	Tyrosinemia type 2	MetabERN	Update
Abstract	Published	43116	Serotonin syndrome	_	Update
Abstract	Published	470	Lysinuric protein intolerance	MetabERN	Update
Abstract	Published	69723	Tyrosinemia type 3	MetabERN	Update
Abstract	Published	626	Large congenital melanocytic nevus	_	Update
Abstract	Published	86884	Subcutaneous panniculitis-like T-cell lymphoma	_	Update
Abstract	Published	163	Hereditary hyperferritinemia-cataract syndrome	EuroBloodNet	Update
Abstract	Published	83450	Regional odontodysplasia	ERN CRANIO	Update
Abstract	Published	67039	Segmental odontomaxillary dysplasia	ERN CRANIO	Update
Abstract	Ongoing	369	Glycogen storage disease due to liver glycogen phosphorylase deficiency	_	Update
Abstract	Published	2374	Congenital laryngeal web	CRANIO	Update
Abstract	Ongoing	51	Aicardi-Goutières syndrome	RITA	Update
Abstract	Published	73423	Acute ackee fruit intoxication	_	Update
Abstract	Published	41	Dyschromatosis symmetrica hereditaria	_	Update
Abstract	Ongoing	450	Heterotaxia	_	Update
Abstract	Ongoing	33001	Lymphedema-distichiasis syndrome	VASCERN	Update
Abstract	Published	559	Marinesco-Sjögren syndrome	ITHACA	Update
Abstract	Published	647799	MYT1L-related developmental delay-intellectual disability-obesity syndrome		New
Abstract	Ongoing	48377	Subcorneal pustular dermatosis	_	Update
Abstract	Ongoing	91412	Marcus-Gunn syndrome	_	Update
Abstract	Published	34587	Glycogen storage disease due to LAMP-2 deficiency	_	Update
Abstract	Ongoing	3352	Tricho-dento-osseous syndrome	_	Update
Abstract	Published	329224	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	ITHACA	New
Abstract	Published	36258	Buerger disease	_	Update
Abstract	Published	98823	Chronic myelomonocytic leukemia	EuroBloodNet	New

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Abstract	Published	684	Paramyotonia congenita of Von Eulenburg	EURO-NMD	Update
Abstract	Ongoing	813	Silver-Russell syndrome	_	Update
Abstract	Published	2268	ICF syndrome	_	Update
Abstract	Ongoing	2978	Chronic intestinal pseudoobstruction	_	Update
Abstract	Ongoing	33314	Jessner lymphocytic infiltration of the skin	_	Update
Abstract	Published	35710	Glucose-galactose malabsorption	_	Update
Abstract	Published	50918	Kikuchi-Fujimoto disease	_	Update
Abstract	Published	389	Langerhans cell histiocytosis	_	Update
Abstract	Ongoing	2176	Infantile systemic hyalinosis	_	Update
Abstract	Ongoing	2806	Subacute sclerosing leukoencephalitis	_	Update
Abstract	Published	71276	Silent sinus syndrome	_	Update
Abstract	Published	3005	Pyle disease	_	Update
Abstract	Published	31202	Melioidosis	_	Update
Abstract	Published	71505	Cancer-associated retinopathy	_	Update
Abstract	Ongoing	48104	Pyoderma gangrenosum	_	Update
Abstract	Published	2406	Locked-in syndrome	_	Update
Abstract	Published	892	Von Hippel Lindau disease	GENTURIS	Update
Abstract	Ongoing	43115	Hereditary myopathy with lactic acidosis due to ISCU deficiency	_	Update
Abstract	Published	36386	Hereditary sensory and autonomic neuropathy type 1	_	Update
Abstract	Published	70472	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type	_	Update
Abstract	Ongoing	98818	Landau-Kleffner syndrome	EpiCare	Update
Abstract	Ongoing	562	McCune-Albright syndrome	_	Update
Abstract	Published	1248	Maxillonasal dysplasia	_	Update
Abstract	Published	3280	Syringomyelia	_	Update
Abstract	Published	646113	Intermediate collagen VI-related dystrophy	_	New
Abstract	Ongoing	34217	Naxos disease	_	Update
Abstract	Published	2788	Osteoporosis-pseudoglioma syndrome		Update
Abstract	Published	59315	Rhombencephalosynapsis	ITHACA	Update
Abstract	Published	3346	Tracheal agenesis	_	Update
Abstract	Published	887	VACTERL/VATER association	_	Update
Abstract	Published	646278	CDK13-related congenital heart defects-intellectual disability-facial dysmorphism syndrome	ITHACA	New
Abstract	Published	33445	Neuroectodermal melanolysosomal disease	_	Update
Abstract	Published	98870	Congenital dyserythropoietic anemia type III	_	Update
Abstract	Ongoing	51188	Ethylmalonic encephalopathy	EURO-NMD	Update
Definition QC	Published	3402	Transient tyrosinemia of the newborn	MetabERN	Update
Definition QC	Published	465508	Symptomatic form of HFE-related hemochromatosis	EuroBloodNet	Update
Definition QC	Published	439232	AApoAIV amyloidosis	ERN RITA	Update
Definition QC	Published	90796	46,XY difference of sex development due to isolated 17,20-lyase deficiency	Endo-ERN; ERN-BOND	Update

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition QC	Published	648562	Ferroportin disease	EuroBloodNet	Update
Definition QC	Ongoing	352675	X-linked Charcot-Marie-Tooth disease type 6	EURO-NMD; EURO-NMD	New
Definition QC	Ongoing	65288	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	Endo-ERN	Update
Definition QC	Ongoing	99014	X-linked Charcot-Marie-Tooth disease type 5	EURO-NMD; EURO-NMD	New
Definition QC	Ongoing	101077	X-linked Charcot-Marie-Tooth disease type 3	EURO-NMD	New
Definition QC	Ongoing	101078	X-linked Charcot-Marie-Tooth disease type 4	EURO-NMD	New
Definition QC	Ongoing	101075	X-linked Charcot-Marie-Tooth disease type 1	EURO-NMD	New
Definition QC	Ongoing	101076	X-linked Charcot-Marie-Tooth disease type 2	EURO-NMD	New
Definition QC	Ongoing	99989	Intermediate DEND syndrome	Endo-ERN	Update
Definition QC	Published	99892	ACTH-dependent Cushing syndrome	Endo-ERN	Update
Definition QC	Published	647758	Adrenal Cushing syndrome	Endo-ERN	New
Definition	Published	1247	Schistosomiasis	Orphanet	
Definition	Published	168598	Methionine adenosyltransferase I/III deficiency	Orphanet	
Definition	Published	446	Neonatal hemochromatosis	Orphanet	
Definition	Published	2118	Hawkinsinuria	Orphanet	
Definition	Published	2334	Autosomal dominant keratitis	Orphanet	
Definition	Published	945	Acalvaria	Orphanet	
Definition	Published	1120	Lung agenesis-heart defect-thumb anomalies syndrome	Orphanet	
Definition	Published	989	Hypoglossia-hypodactyly syndrome	Orphanet	
Definition	Published	137608	Segmental outgrowth-lipomatosis-arteriovenous malformation-epidermal nevus syndrome	Orphanet	
Definition	Published	1170	Autosomal recessive cerebelloparenchymal disorder type 3	Orphanet	
Definition	Published	1208	Pulmonary atresia-intact ventricular septum syndrome	Orphanet	
Definition	Published	141239	Median cleft of the upper lip and maxilla	Orphanet	
Definition	Published	155878	Submucosal cleft palate	Orphanet	
Definition	Published	141288	Midline cervical cleft	Orphanet	
Definition	Published	139480	Autosomal recessive spastic paraplegia type 39	Orphanet	
Definition	Published	139578	Mutilating hereditary sensory neuropathy with spastic paraplegia	Orphanet	
Definition	Published	1671	Split cord malformation type I	Orphanet	
Definition	Published	140949	Low-flow priapism	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Published	1952	Epiphyseal stippling-osteoclastic hyperplasia syndrome	Orphanet	
Definition	Published	1791	Frontofacionasal dysplasia	Orphanet	
Definition	Published	2022	Endocardial fibroelastosis	Orphanet	
Definition	Published	2006	Median cleft lip/mandible	Orphanet	
Definition	Published	247815	Autosomal recessive ataxia due to PEX10 deficiency	Orphanet	
Definition	Published	247691	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations	Orphanet	
Definition	Published	2470	Matthew-Wood syndrome	Orphanet	
Definition	Published	251019	2q32q33 microdeletion syndrome	Orphanet	
Definition	Published	2674	Cyprus facial-neuromusculoskeletal syndrome	Orphanet	
Definition	Published	2699	Median nodule of the upper lip	Orphanet	
Definition	Published	254875	Mitochondrial DNA depletion syndrome, myopathic form	Orphanet	
Definition	Published	261529	Ring chromosome Y syndrome	Orphanet	
Definition	Published	263708	Complex chromosomal rearrangement	Orphanet	
Definition	Published	263297	Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency	Orphanet	
Definition	Published	3132	Say-Barber-Miller syndrome	Orphanet	
Definition	Published	268744	Spinal dysraphism with a posterior meningocele	Orphanet	
Definition	Published	268813	Myelocystocele	Orphanet	
Definition	Published	2959	Progeria-short stature-pigmented nevi syndrome	Orphanet	
Definition	Published	3374	Unilateral ocular duplication	Orphanet	
Definition	Published	3412	VACTERL with hydrocephalus	Orphanet	
Definition	Published	3471	Young syndrome	Orphanet	
Definition	Published	3168	Sillence syndrome	Orphanet	
Definition	Published	3180	Spondylocamptodactyly syndrome	Orphanet	
Definition	Published	276608	Non-insulinoma pancreatogenous hypoglycemia syndrome	Orphanet	
Definition	Published	209004	Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy	Orphanet	
Definition	Published	3240	Early-onset progressive leukoencephalopathy-central nervous system calcification-deafness-visual impairment syndrome	Orphanet	
Definition	Published	2330	Kasabach-Merritt phenomenon	Orphanet	
Definition	Published	217067	Pouchitis	Orphanet	

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Definition	Published	3385	African trypanosomiasis	Orphanet	
Definition	Published	341	Viral hemorrhagic fever	Orphanet	
Definition	Published	980	Absence of the pulmonary artery	Orphanet	
Definition	Published	3124	Saccharopinuria	Orphanet	
Definition	Published	2203	Hyperlysinemia	Orphanet	
Definition	Published	2195	Dicarboxylic aminoaciduria	Orphanet	
Definition	Published	221083	Hemifacial spasm	Orphanet	
Definition	Published	2126	Solitary fibrous tumor	Orphanet	
Definition	Published	228396	Ptosis-upper ocular movement limitation-absence of lacrimal punctum syndrome	Orphanet	
Definition	Published	452	X-linked lissencephaly with abnormal genitalia	Orphanet	
Definition	Published	329324	Inverse Klippel-Trénaunay syndrome	Orphanet	
Definition	Published	80	Antiphospholipid syndrome	Orphanet	
Definition	Published	464343	Catastrophic antiphospholipid syndrome	Orphanet	
Definition	Published	464458	Paracetamol poisoning	Orphanet	
Definition	Published	449285	Snakebite envenomation	Orphanet	
Definition	Published	466670	Cyanide poisoning	Orphanet	
Definition	Published	404443	Tatton-Brown-Rahman syndrome	Orphanet	
Definition	Published	398069	MAGEL2-related Prader-Willi-like syndrome	Orphanet	
Definition	Published	398073	Prader-Willi-like syndrome	Orphanet	
Definition	Published	398079	SIM1-related Prader-Willi-like syndrome	Orphanet	
Definition	Published	443291	HIV-associated cancer	Orphanet	
Definition	Published	443804	Focal stiff limb syndrome	Orphanet	
Definition	Published	420429	Glycogen storage disease due to acid maltase deficiency, late-onset	Orphanet	
Definition	Published	26137	Juvenile temporal arteritis	Orphanet	
Definition	Published	26348	Acquired prothrombin deficiency	Orphanet	
Definition	Published	38	Acrokeratoelastoidosis of Costa	Orphanet	
Definition	Published	66637	Diaphanospondylodysostosis	Orphanet	
Definition	Published	65282	Carvajal syndrome	Orphanet	
Definition	Published	64692	Bartonella bacilliformis infection	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Published	79154	2-aminoadipic 2-oxoadipic aciduria	Orphanet	
Definition	Published	79094	Grange syndrome	Orphanet	
Definition	Published	75374	Bradyopsia	Orphanet	
Definition	Published	566862	Left sided atrial isomerism	Orphanet	
Definition	Published	36235	Staphylococcal scarlet fever	Orphanet	
Definition	Published	53372	Hereditary geniospasm	Orphanet	
Definition	Published	53698	Myosin storage myopathy	Orphanet	
Definition	Published	50817	Duane anomaly-myopathy-scoliosis syndrome	Orphanet	
Definition	Published	90307	Parkes Weber syndrome	Orphanet	
Definition	Published	90078	Invasive infections due to vancomycin-resistant enterococci	Orphanet	
Definition	Published	88660	Hypertension due to gain-of-function mutations in the mineralocorticoid receptor	Orphanet	
Definition	Published	79323	MPDU1-CDG	Orphanet	
Definition	Published	79322	DPM1-CDG	Orphanet	
Definition	Published	85163	Hypomyelination-congenital cataract syndrome	Orphanet	
Definition	Published	84132	Desmin-related myopathy with Mallory body-like inclusions	Orphanet	
Definition	Published	603448	Cerebellar hypoplasia-intellectual disability-congenital microcephaly-dystonia-anemia-growth retardation syndrome	Orphanet	
Definition	Published	603494	Coloboma-osteopetrosis-microphthalmia-macrocephaly-albinism-deafness syndrome	Orphanet	
Definition	Published	604680	Symptomatic form of X-linked centronuclear myopathy in female carriers	Orphanet	
Definition	Published	611256	Pontocerebellar hypoplasia type 12	Orphanet	
Definition	Published	611247	Pontocerebellar hypoplasia type 11	Orphanet	
Definition	Published	611216	Aplastic anemia-intellectual disability-dwarfism syndrome	Orphanet	
Definition	Published	613267	Pontocerebellar hypoplasia type 13	Orphanet	
Definition	Published	613274	Pontocerebellar hypoplasia type 14	Orphanet	
Definition	Published	600668	CCNK-related neurodevelopmental disorder-severe intellectual disability-facial dysmorphism syndrome	Orphanet	
Definition	Published	600731	Clark-Baraitser syndrome	Orphanet	
Definition	Published	97337	Sinding-Larsen-Johansson disease	Orphanet	
Definition	Published	95494	Combined pituitary hormone deficiencies, genetic forms	Orphanet	
Definition	Published	94086	Blue diaper syndrome	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Published	597623	IRF2BPL-related regressive neurodevelopmental disorder-dystonia-seizures syndrome	Orphanet	
Definition	Published	597733	Oculocutaneous albinism type 8	Orphanet	
Definition	Published	597738	Luscan-Lumish syndrome	Orphanet	
Definition	Published	597887	ALPI-related inflammatory bowel disease	Orphanet	
Definition	Published	597939	Euthyroid dysprealbuminemic hyperthyroxinemia	Orphanet	
Definition	Published	597743	SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome	Orphanet	
Definition	Published	597746	Blepharophimosis-intellectual disability syndrome/genitopatellar overlap syndrome	Orphanet	
Definition	Published	597874	MTHFS-related developmental delay-microcephaly-short stature-epilepsy syndrome	Orphanet	
Definition	Published	598216	Upper tract urothelial carcinoma	Orphanet	
Definition	Published	598603	Facial dysmorphism-hypertrichosis-epilepsy-intellectual disability/developmental delay-gingival overgrowth syndrome	Orphanet	
Definition	Published	598363	Multisystem inflammatory syndrome in children and adults	Orphanet	
Definition	Published	597201	TRIM22-related inflammatory bowel disease	Orphanet	
Definition	Published	599376	Hypomyelination of early myelinating structures	Orphanet	
Definition	Published	599082	CHD3-related developmental delay-speech delay-intellectual disability-abnormalities of vision-facial dysmorphism syndrome	Orphanet	
Definition	Published	93969	Open spinal dysraphism with a myelomeningocele	Orphanet	
Definition	Published	596759	Combined immunodeficiency due to RELA haploinsufficiency	Orphanet	
Definition	Published	596753	VEXAS syndrome	Orphanet	
Definition	Published	99361	Familial medullary thyroid carcinoma	Orphanet	
Definition	Published	99704	Early-onset obesity-hyperphagia-severe developmental delay syndrome	Orphanet	
Definition	Published	583856	Isolated splenic vein thrombosis	Orphanet	
Definition	Published	583861	Isolated mesenteric vein thrombosis	Orphanet	
Definition	Published	98798	Isochromosomy Yq	Orphanet	
Definition	Published	98797	Isochromosomy Yp	Orphanet	
Definition	Published	581271	Cramp-fasciculation syndrome	Orphanet	
Definition	Published	576074	Middle East respiratory syndrome	Orphanet	
Definition	Published	573253	Split cord malformation type II	Orphanet	
Definition	Published	573278	Split cord malformation	Orphanet	
Definition	Published	572773	Microcephaly-short stature-limb abnormalities syndrome	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Published	572768	Microcephaly-micromelia syndrome	Orphanet	
Definition	Published	572333	Blepharophimosis-ptosis-epicanthus inversus syndrome plus	Orphanet	
Definition	Published	100054	F12-related hereditary angioedema with normal C1Inh	Orphanet	
Definition	Published	645749	Congenital esophageal stenosis	Orphanet	
Definition	Published	645393	Hemi-myeloschisis	Orphanet	
Definition	Published	645388	Hemi-myelomeningocele	Orphanet	
Definition	Published	645401	True myeloschisis	Orphanet	
Definition	Published	645398	Myeloschisis	Orphanet	
Definition	Published	645367	Conus spinal cord lipoma	Orphanet	
Definition	Published	645362	Dorsal spinal cord lipoma	Orphanet	
Definition	Published	645383	True myelomeningocele	Orphanet	
Definition	Published	645378	Myelic limited dorsal malformation	Orphanet	
Definition	Published	645354	Saccular limited dorsal myeloschisis	Orphanet	
Definition	Published	645359	Intramedullary non-dysraphic spinal cord lipoma	Orphanet	
Definition	Published	645343	Non-saccular limited dorsal myeloschisis	Orphanet	
Definition	Published	645337	Terminal myelocystocele	Orphanet	
Definition	Published	645340	Non-terminal myelocystocele	Orphanet	
Definition	Published	645325	Isolated filum lipoma	Orphanet	
Definition	Published	645334	Retained medullary cord	Orphanet	
Definition	Published	645319	Saccular spinal dysraphism with a stalk to the dome	Orphanet	
Definition	Published	645322	Isolated transitional filum lipoma	Orphanet	
Definition	Published	645300	Lipomatous non-saccular limited dorsal myeloschisis	Orphanet	
Definition	Published	645310	Fibroneural non-saccular limited dorsal myeloschisis	Orphanet	
Definition	Published	645294	Posterior extramedullary conus spinal cord lipoma	Orphanet	
Definition	Published	645297	Extramedullary conus spinal cord lipoma	Orphanet	
Definition	Published	645288	Terminal extramedullary conus spinal cord lipoma	Orphanet	
Definition	Published	645291	Transitional extramedullary conus spinal cord lipoma	Orphanet	
Definition	Published	645285	Chaotic conus spinal cord lipoma	Orphanet	
Definition	Published	645282	Anomaly of the filum	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Published	645279	Fibrolipomatous filum anomaly	Orphanet	
Definition	Published	645276	Spinal cord lipoma	Orphanet	
Definition	Published	645273	Dysraphic spinal cord lipoma	Orphanet	
Definition	Published	645270	Open spinal dysraphism with a posterior meningocele	Orphanet	
Definition	Published	645202	Closed spinal dysraphism	Orphanet	
Definition	Published	645196	Limited dorsal myeloschisis	Orphanet	
Definition	Published	645193	Dysraphism with stalk	Orphanet	
Definition	Published	645188	Spinal dermal sinus	Orphanet	
Definition	Published	643549	Hao-Fountain syndrome	Orphanet	
Definition	Published	643503	Marfanoid habitus-facial dysmorphism-skeletal abnormality-heart defect syndrome	Orphanet	
Definition	Published	648919	Idiopathic catatonia	Orphanet	
Definition	Published	648581	Digenic hemochromatosis	Orphanet	
Definition	Published	648681	Immune-mediated scleritis	Orphanet	
Definition	Published	648675	Idiopathic scleritis	Orphanet	
Definition	Published	99912	Ovarian dysgerminoma	Orphanet	
Definition	Published	647811	Cardiac-urogenital syndrome	Orphanet	
Definition	Published	647804	Combined immunodeficiency due to FCHO1 deficiency	Orphanet	
Definition	Published	656417	Embryonal tumor with multilayered rosettes	Orphanet	
Definition	Published	653712	CHD4-related neurodevelopmental disorder	Orphanet	
Definition	Published	652519	Cleft palate-congenital heart defect-intellectual disability syndrome	Orphanet	
Definition	Published	652532	Adult-onset progressive leukoencephalopathy-early-onset deafness	Orphanet	
Definition	Published	659756	Oroya fever	Orphanet	
Definition	Published	659759	Verruga peruana	Orphanet	
Definition	Published	631248	Mitchell Syndrome	Orphanet	
Definition	Published	634475	Mosaic NF2-related schwannomatosis	Orphanet	
Definition	Published	634492	Mosaic schwannomatosis	Orphanet	
Definition	Published	634511	Mosaic Legius syndrome	Orphanet	
Definition	Published	634461	Mosaic neurofibromatosis type 1	Orphanet	
Definition	Published	637064	Isolated optic nerve aplasia	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Published	637061	Isolated optic nerve hypoplasia	Orphanet	
Definition	Published	637051	Borna virus encephalitis	Orphanet	
Definition	Published	637013	SMARCA2-related blepharophimosis-intellectual disability syndrome	Orphanet	
Definition	Published	641829	Neonatal compartment syndrome	Orphanet	
Definition	Published	641496	Childhood-onset schizophrenia	Orphanet	
Definition	Published	641390	PsAPASH syndrome	Orphanet	
Definition	Published	641380	PAPASH syndrome	Orphanet	
Definition	Published	641385	PASS syndrome	Orphanet	
Definition	Published	641353	Infantile neurodegeneration-progressive spasticity-intellectual disability-white matter lesions syndrome	Orphanet	
Definition	Published	641361	Neurodevelopmental delay-hypotonia-cerebellar ataxia-cardiac conduction defects syndrome	Orphanet	
Definition	Published	642965	Autosomal recessive ataxia due to PEX2 deficiency	Orphanet	
Definition	Published	642954	Autosomal recessive ataxia due to PEX16 deficiency	Orphanet	
Definition	Published	642763	Autosomal dominant intellectual disability-craniofacial dysmorphism-macrocephaly-hypotonia syndrome due to H1-4 mutation	Orphanet	
Definition	Published	642747	PUM1-related cerebellar ataxia	Orphanet	
Definition	Published	642691	Fragile X-associated primary ovarian insufficiency	Orphanet	
Definition	Published	642675	CHD8 overgrowth syndrome	Orphanet	
Definition	Published	642099	Spondyloepimetaphyseal dysplasia with joint laxity, Beighton type	Orphanet	
Definition	Published	642085	EXOC6B-related spondyloepimetaphyseal dysplasia with joint laxity	Orphanet	
Definition	Published	615964	Acute reversible leukoencephalopathy with increased urinary alpha-ketoglutarate	Orphanet	
Definition	Published	617449	Congenital aphakia-iris hypoplasia-microphthalmia-microcornea syndrome	Orphanet	
Definition	Published	617408	Classic eosinophilic pustular folliculitis	Orphanet	
Definition	Published	617440	Painful legs and moving toes syndrome	Orphanet	
Definition	Published	617304	Amniotic fluid embolism	Orphanet	
Definition	Published	617294	Twin anemia-polycythemia sequence	Orphanet	
Definition	Published	617301	Selective intrauterine growth restriction	Orphanet	
Definition	Published	617297	Twin-reversed arterial perfusion sequence	Orphanet	
Definition	Published	617910	Conjunctival malignant melanoma	Orphanet	
Definition	Published	619972	CADINS disease	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Ongoing	658913	Paragonimiasis	Orphanet	
Definition	Ongoing	658909	Fasciolopsiasis	Orphanet	
Definition	Ongoing	658917	Clonorchiasis	Orphanet	
Definition	Ongoing	621758	Fibrosis-neurodegeneration-cerebral angiomatosis syndrome	Orphanet	
Definition	Ongoing	617919	F12-associated cold autoinflammatory syndrome	Orphanet	
Definition	Ongoing	619948	Early-onset autoimmunity-autoinflammation-immunodeficiency syndrome due to SOCS1 haploinsufficiency	Orphanet	
Definition	Ongoing	653709	Cone rod dystrophy-short stature syndrome	Orphanet	
Definition	Ongoing	653728	Congenital insensitivity to pain syndrome, Marsili type	Orphanet	
Definition	Ongoing	79492	Pili gemini	Orphanet	
Definition	Ongoing	482	Kimura Disease	Orphanet	
Definition	Ongoing	1685	Distomatosis	Orphanet	
Definition	Ongoing	213504	Adenocarcinoma of ovary	Orphanet	
Definition	Ongoing	424982	Biliary cystadenocarcinoma	Orphanet	
Definition	Ongoing	618891	Chronic neurovisceral acid sphingomyelinase deficiency	Orphanet	
Definition	Ongoing	209989	Non-papillary transitional cell carcinoma of the bladder	Orphanet	
Definition	Ongoing	646139	Dysplastic cortical hyperostosis	Orphanet	
Definition	Ongoing	87884	Non-syndromic genetic deafness	Orphanet	
Definition	Ongoing	83642	Microcytic anemia with liver iron overload	Orphanet	
Definition	Ongoing	663	Mitochondrial DNA-related progressive external ophthalmoplegia	Orphanet	
Definition	Ongoing	2148	Lissencephaly type 1 due to doublecortin gene mutation	Orphanet	
Definition	Ongoing	162	Cataract-glaucoma syndrome	Orphanet	
Definition	Ongoing	75373	Progressive bifocal chorioretinal atrophy	Orphanet	
Definition	Ongoing	599519	Factor V short isoforms-related bleeding disorder	Orphanet	
Definition	Ongoing	90970	Primary lipodystrophy	Orphanet	
Definition	Ongoing	631095	Spinocerebellar ataxia type 44	Orphanet	
Definition	Ongoing	631103	Spinocerebellar ataxia type 48	Orphanet	
Definition	Ongoing	2356	Arachnoid cyst	Orphanet	
Definition	Ongoing	2021	Fibrochondrogenesis	Orphanet	

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Text type	Status	ORPHAcode	Name of the entity	ERN	Type of abstract
Definition	Ongoing	1048	Isolated anencephaly/exencephaly	Orphanet	
Definition	Ongoing	1172	Autosomal recessive cerebellar ataxia	Orphanet	
Definition	Ongoing	674	Accessory pancreas	Orphanet	

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