WHAT IS IT AND WHY IS IT NEEDED

Dre Loredana D'Amato Sizonenko, MD
Genetic Medicine Division - HUG
Coordinator Orphanet Switzerland

Martin Arles
Orphanet Switzerland project manager

Inselspital, Bern - 13th February 2020

Hôpitaux Universitaires Genève

Coordination nationale des maladies rares

What is Orphanet?

Orphanet is **the leader in collecting, integrating, producing and disseminating added-value information and data in the field of rare diseases**.

- The added-value comes from the data being **expert-reviewed, manually curated and integrated** to other resources.
- Thus, Orphanet is a tool both for **healthcare** and for **research**.

Orphanet is the reference for the **nomenclature and classification of rare diseases** (it's the only specific resource) and has a normative role in this domain.

Orphanet was established in 1997 to address identified problems

Lack of information	→	Encyclopaedia of rare diseases
Scarce expertise	→	Directory of experts/clinics
Too few collaborations	→	Directory of research projects
Difficult recruitment	→	Directory of clinical trials
Isolation of patients	→	Directory of patient organisations

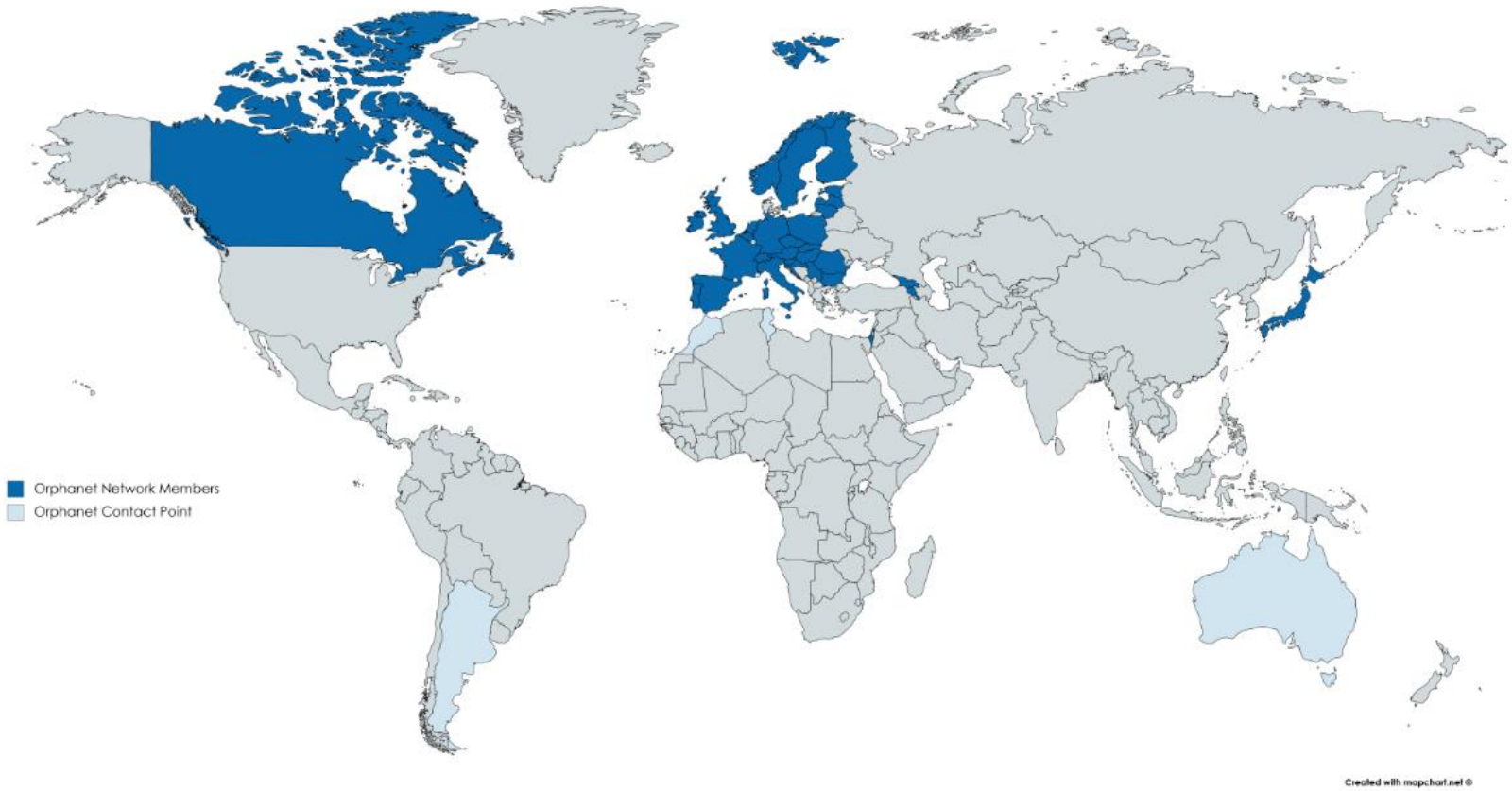
orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Orphanet network

37 countries worldwide

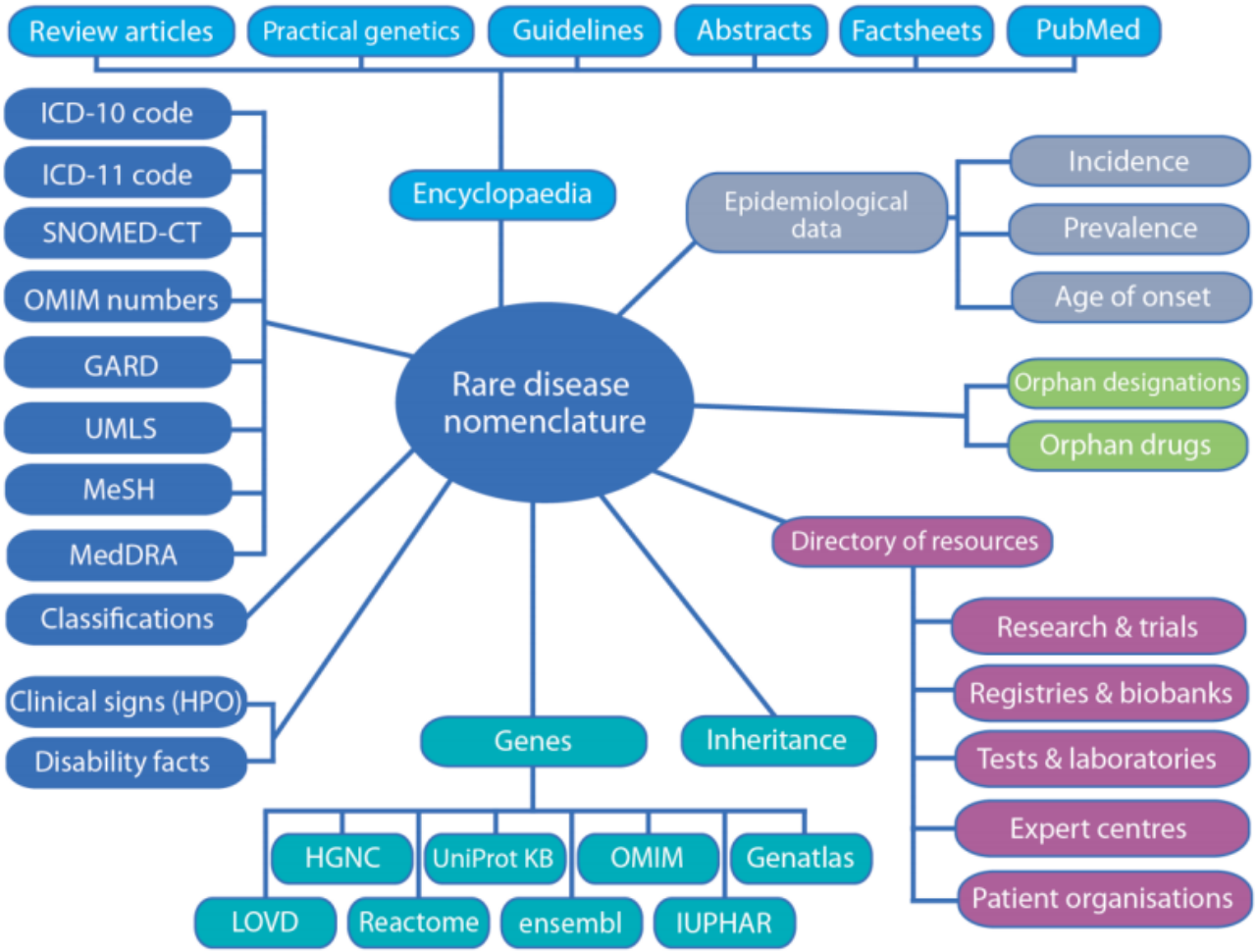


orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Database structure



orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Orphanet's missions

- Improve the visibility of rare diseases in healthcare and research systems

- Provide high-quality information and expertise on rare diseases

- Contribute to generating knowledge on rare diseases

→ piecing together the parts of the puzzle to better understand rare diseases

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Orphanet nomenclature and classification

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

The Orphanet nomenclature

- Each disease is assigned **a <u>unique</u> and <u>stable</u>** ORPHA code
- Cross-referenced with other international terminologies
- The only nomenclature **specific for rare diseases**
- Nine languages (Cz, En, Es, De, Fr, It, Nl, Pt, Pl… current translations JP, CN)

ORPHA number	Preferred label	Synonyms
ORPHA:893	**WAGR syndrome**	Del(11)(p13)
		Deletion 11p13
		Monosomy 11p13
		Wilms tumor-aniridia-genitourinary anomalies-intellectual disability syndrome
ORPHA:231169	**Usher syndrome type 1**	USH1

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

The Orphanet nomenclature

- Each disease is assigned **a unique and stable** ORPHA code
- Cross-referenced with other international terminologies
- The only nomenclature **specific for rare diseases**
- Nine languages (Cz, En, Es, De, Fr, It, Nl, Pt, Pl… current translations JP, CN)

→ Providing **a common language** across the RD field: different systems can work together

→ Improving **the recognition of rare diseases** in healthcare and research systems

The Orphanet RD classification

Example: Stickle syndrome (ORPHA:828)

Characterized by a distinctive facial appearance, eye abnormalities, hearing loss, and joint problems.

Multi-dimensional

Stickler syndrome

- Orphanet classification of rare developmental anomalies during embryogenesis
- Classification of sucking/swallowing disorders
- Orphanet classification of rare genetic diseases
- Orphanet classification of rare eye diseases
- Orphanet classification of rare bone diseases
- Orphanet classification of rare otorhinolaryngological diseases
- Orphanet classification of rare surgical maxillo-facial diseases

Multi-hierarchical

Orphanet classification of rare otorhinolaryngological diseases

- Rare otorhinolaryngologic disease ORPHA:98036
  - Syndrome or malformation associated with head and neck malformations ORPHA:156237
    - Rare disease with Pierre Robin syndrome ORPHA:138044
      - Pierre Robin syndrome associated with collagen disease ORPHA:138041
        - Stickler syndrome ORPHA:828

- Rare otorhinolaryngologic disease ORPHA:98036
  - Rare deafness ORPHA:68361
    - Syndromic genetic deafness ORPHA:90642
      - Stickler syndrome ORPHA:828

orphanet

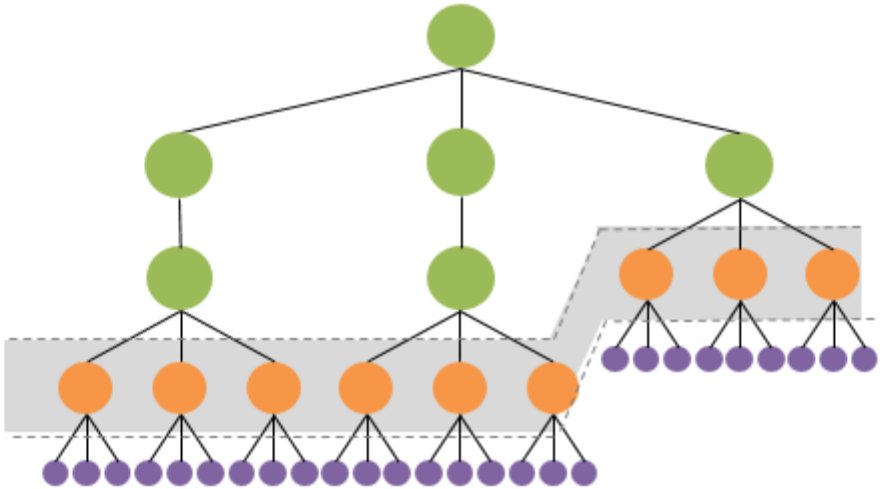
HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

The Orphanet RD classification

- 55 Classification of rare metabolic diseases (Functional) (Rch,Dgs,Cns,Ass)
  - 10507 Rare inborn errors of metabolism [Category] ORPHA68367
    - 10506 Lysosomal disease [Category] ORPHA68366
      - 21365 Glycoproteinosis [Category] ORPHA309279
        - 11238 Mucolipidosis [Category] ORPHA79212
          - 27 Mucolipidosis type II [Disease] ORPHA576
          - 28 Mucolipidosis type III [Disease] ORPHA577
            - 23158 Mucolipidosis type III alpha/beta [Clinical subtype] ORPHA423461
            - 23159 Mucolipidosis type III gamma [Clinical subtype] ORPHA423470
          - 29 Mucolipidosis type IV [Disease] ORPHA578
        - 11241 Oligosaccharidosis [Category] ORPHA79215
          - 3 Alpha-mannosidosis [Disease] ORPHA61
            - 21366 Alpha-mannosidosis, infantile form [Clinical subtype] ORPHA309282
            - 21367 Alpha-mannosidosis, adult form [Clinical subtype] ORPHA309288
          - 5 Aspartylglucosaminuria [Disease] ORPHA93
          - 7 Beta-mannosidosis [Disease] ORPHA118
          - 13 Fucosidosis [Disease] ORPHA349
          - 498 Galactosialidosis [Disease] ORPHA351
          - 673 Alpha-N-acetylgalactosaminidase deficiency [Disease] ORPHA3137
            - 11305 Alpha-N-acetylgalactosaminidase deficiency type 1 [Clinical subtype] ORPHA79279
            - 11306 Alpha-N-acetylgalactosaminidase deficiency type 2 [Clinical subtype] ORPHA79280
            - 11307 Alpha-N-acetylgalactosaminidase deficiency type 3 [Clinical subtype] ORPHA79281
          - 21368 Sialidosis [Clinical group] ORPHA309294
            - 26 Sialidosis type 1 [Disease] ORPHA812
            - 11801 Sialidosis type 2 [Disease] ORPHA87876
              - 12321 Juvenile sialidosis type 2 [Clinical subtype] ORPHA93399
              - 12322 Congenital sialidosis type 2 [Clinical subtype] ORPHA93400

The Orphanet RD classification



Group

Disorder

Subtype

Group: Category, clinical group

Disorder: Disease, clinical syndrome, malformation syndrome, morphological anomaly, biological anomaly, particular clinical situation

Subtype: Etiological, clinical, histopathological

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

Scientific annotations

Natural history data	Number of groups of disorders, disorders and sub-types
Average age of onset	5,879
Mode of inheritance	5,341

Epidemiological data	Number of groups of disorders, disorders and sub-types
Point prevalence	5,722
Prevalence at birth	507
Lifetime prevalence	46
Annual incidence	575

As of January 2019.

European Journal of Human Genetics

Article | Open Access | Published: 16 September 2019

Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database

Stéphanie Nguengang Wakap, Deborah M. Lambert, Annie Olry, Charlotte Rodwell, Charlotte Gueydan, Valérie Lanneau, Daniel Murphy, Yann Le Cam & Ana Rath

European Journal of Human Genetics (2019) | Download Citation

3541 Accesses | **123** Altmetric | Metrics »

- **6172** unique rare diseases (number stable over time)
- 86% are prevalent diseases (long term, chronic illnesses)
- 14% are incident diseases (cancers, infections, poisonings)

What is the age of onset?

- 70% only in childhood
- 12% only in adulthood
- 18% either in childhood or in adulthood

- 72% of rare diseases have a genetic basis

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

Orphanet inventory of genes

Genes involved in rare diseases are entered in the database and updated regularly according to **new scientific publications**.

The relationship between a gene and a disease is **manually qualified** according to the role that the gene plays in the **pathogenesis** of the disease:

- Causative
- Modifiers
- Major susceptibility factors
- Playing a role in the phenotype

Genes:

5,340 genes linked to 3,832 rare diseases
5,337 genes interfaced with HGNC
4,847 genes interfaced with OMIM
4,967 genes interfaced with Genatlas
4,863 genes interfaced with UniProtKB
5,103 genes interfaced with Ensembl
689 genes interfaced with IUPHAR-DB
3,992 genes interfaced with Reactome

Orphanet nomenclature annotated with phenotypic traits (HPO)

- From 2015 Orphanet disorders are annotated with the **Human Phenotype Ontology**, a standard and controlled terminology covering phenotypic abnormalities in human diseases, recognised as the reference in the domain.

- Each phenotypic term is associated with the frequency of occurrence (obligate, very frequent, frequent, occasional, very rare, excluded), and whether the annotated HPO term is a major diagnostic criterion or a pathognomonic sign of the rare disease.

- 3,312 diseases were annotated with HPO terms (as of January 2019).

Orphanet's missions

- Improve the visibility of rare diseases in healthcare and research systems

- Provide high-quality information and expertise on rare diseases

- Contribute to generating knowledge on rare diseases

→ piecing together the parts of the puzzle to better understand rare diseases

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

The Orphanet Professional Encyclopedia

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

The Orphanet Professional Encyclopedia

- Summary texts:
  - Disease definitions
  - Abstracts (8-10 sections)
- Intended for healthcare professionals
- Clinical emphasis
- Based on review of biomedical literature
- Use of HPO terms
- Validated by internationally recognized experts

The Orphanet Professional Encyclopedia

Farber disease

Suggest an update

Disease definition

A subcutaneous tissue disease characterized by a spectrum of clinical signs ranging from the classical triad of painful and progressively deformed joints, subcutaneous nodules, and progressive hoarseness (due to laryngeal involvement) that presents in infancy, to varying phenotypes with respiratory and neurologic involvement.

ORPHA:333

Classification level: Disorder

Synonym(s):
Acid ceramidase deficiency
Farber lipogranulomatosis

Prevalence: <1 / 1 000 000

Inheritance: Autosomal recessive

Age of onset: Infancy, Neonatal, Childhood, Antenatal

ICD-10: E75.2

OMIM: 228000

UMLS: C0268255 C2936785

MeSH: C537075 D055577

GARD: 6426

MedDRA: -

Summary

Epidemiology
Approximately 200 cases of Farber disease have been reported worldwide in the literature to date.

Clinical description
A high clinical variability is seen between patients. The classic phenotype presents at around 3-6 months of age with painful, swollen and stiff joints of the hands and feet, prominent subcutaneous nodules over pressure points, and progressive hoarseness leading to aphonia due to vocal cord infiltration. Patients can also develop cardiac, pulmonary and neurological defects. Progressive neurological deterioration can be marked in some forms with seizures, paraparesis and developmental delay. The most severe neonatal form presents at birth with hydrops fetalis, lethargy, and failure to thrive, as well as hepatosplenomegaly, rapid neurological deterioration, and granulomatous infiltrations to various other organs (i.e. liver, spleen, lungs). Milder forms have also been described with no neurological defects and a longer life-expectancy. In some patients, the disease manifests in childhood as a spinal muscular atrophy associated with progressive myoclonic epilepsy (SMA-PME) in the absence of subcutaneous nodules.

Etiology
Farber disease is caused by mutations in the N-acylsphingosine amidohydrolase (*ASAH1*) gene (8p22) which encodes acid ceramidase, a lysosomal enzyme that hydrolyzes ceramide into sphingosine and free fatty acid. A deficient activity of this enzyme leads to an accumulation of ceramide in most tissues.

Diagnostic methods
Diagnosis is based on clinical and laboratory findings by assaying the activity of acid ceramidase in peripheral blood leukocytes, cultured lymphoid cells or cultured skin fibroblasts. Alternatively, diagnosis can be performed by determining ceramide concentration in cultured cells or tissues or by studying lysosomal ceramide catabolism in cultured living cells. Identification of mutations in the *ASAH1* gene by molecular genetic testing usually allows for diagnostic confirmation.

Differential diagnosis
Differential diagnoses include juvenile idiopathic arthritis, stiff skin syndrome and lethal restrictive dermopathy. Encephalopathy due to prosaposin deficiency should also be excluded.

Antenatal diagnosis
Prenatal diagnosis by DNA testing is possible in families with a known disease-causing mutation. Alternatively, prenatal diagnosis can be performed by measuring acid ceramidase activity in cultured amniotic fluid cells or chorionic villi.

Genetic counseling
The pattern of inheritance is autosomal recessive. Genetic counseling should be proposed to affected families informing them of a 25% risk of transmitting the disease where both parents are unaffected carriers.

The Orphanet encyclopaedia contains the following summary texts:

5,574	English
3,736	French
3,484	Spanish
3,465	Italian
3,322	German
2,261	Dutch
1,173	Portuguese
1,088	Polish
422	Greek
255	Russian
166	Finnish
95	Slovak

Management and treatment
There is currently no effective specific therapy for Farber disease, and symptomatic treatment is based on analgesics, corticotherapy, and plastic surgery. However, allogeneic hematopoietic stem cell transplantation provides a promising approach for patients with limited neurological involvement.

Prognosis
The prognosis varies, with some patients dying within the first few days of life (severe neonatal form) and others living until adolescence or early adulthood (milder forms).

Expert reviewer(s): ***Pr Thierry LEVADE*** *- Last update:* ***April 2019***

Detailed information

Professionals

› Clinical genetics review
English (2018)

Additional information

Further information on this disease
- › Classification(s) (7)
- › Gene(s) (1)
- › Clinical signs and symptoms
- › Publications in PubMed
- › Other website(s) (9)

Health care resources for this disease
- › Expert centres (314)
- › Diagnostic tests (41)
- › Patient organisations (65)
- › Orphan designation(s) and orphan drug(s) (1)

Research activities on this disease
- › Research projects (36)
- › Clinical trials (0)
- › Registries/biobanks (37)
- › Network of experts (2)

Specialised Social Services
- › Eurordis directory

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

The Orphanet Professional Encyclopedia

In-house produced texts

- Articles for the general public
- Emergency guidelines
- Disability factsheets

Links to external texts

- Articles for the general public
- Review articles
- Clinical practice guidelines
- Practical genetics articles
- Clinical genetics review
- Anaesthesia guidelines
- Guidance for genetic testing

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

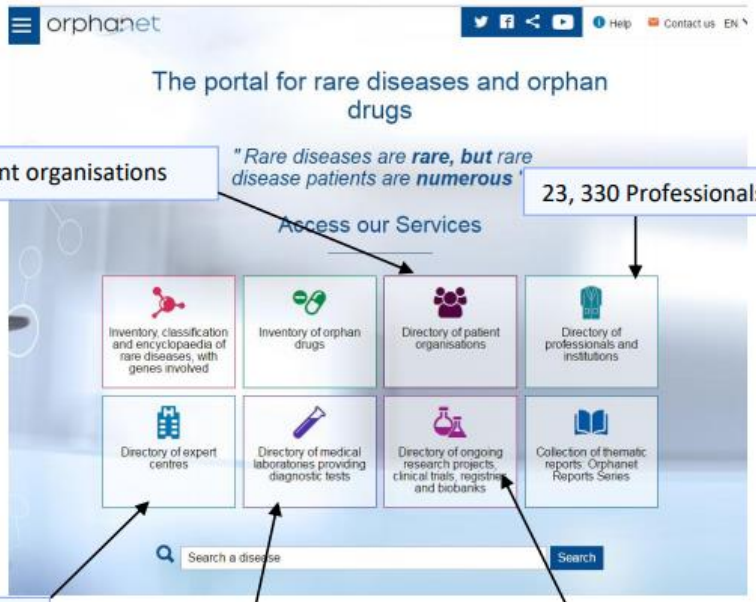
Directory of expert resources

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Directory of expert resources



2,636 Patient organisations

23, 330 Professionals referenced in the database

8,556 Expert centres

1,648 Medical laboratories dedicated to diagnosis

43,836 diagnostic tests linked to 4,819 diseases and 5,067 genes and 1,390 panels of genes

2,281 Research laboratories

3,090 Sites conducting 2,545 ongoing research projects on 1,927 diseases

3,111 Sites conducting 2,723 ongoing clinical trials for 890 diseases

777 Patient registries

270 Mutation databases

161 Biobanks

Figure 9 Directory of expert services (January 2019)

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

Neurometabolic and Metabolic clinic

Interdisciplinary metabolic Team
University Hospital Inselspital
Freiburgstrasse 18
3010 BERN
SWITZERLAND

More information

Head of clinic : Dr Jean-Marc NUOFFER
Phone : **0041(0) 31 632 22 01**
Additional Phone : -
Fax : -
Website
Contact : karinbalmer@insel.ch
Age range : **For Adults *and* Children**
Type(s) : **Medical management**

Team

- Dr Matthias GAUTSCHI (Clinical expert)
- Dr Alexander LÄMMLE (Clinical expert)
- Dr Jean-Marc NUOFFER (Clinical expert)

Description of the activity

Last update: February 2020

Part of

SWITZERLAND
BERN

SGIEM Swiss Group for Inborn Errors of Metabolism
SGIEM

More information

Additional information

Further information on this clinic

- Disease(s)/group of diseases (2)
- Networks (1)

SGIEM Swiss Group for Inborn Errors of Metabolism

Type of network : **Expert centres**

Geographic coverage : **National**

Funding body(ies) : -

Website

Coordinator of expert centre network

Dr Matthias GAUTSCHI

Universitätsklinik für Kinderheilkunde Inselspital

SGIEM

Freiburgstrasse 15

3010 BERN

SWITZERLAND

More information

Phone : 0041 (0)31 632 52 45

Fax : 0041 (0)31 362 84 24

Website

Contact secretary :

metabolik@insel.ch

Last update: January 2020

Members

SWITZERLAND BASEL	Inborn errors of metabolic diseases **Universitätsspital Basel**	More information
SWITZERLAND BERN	Rare metabolic diseases clinic **Inselspital, Universitätsspital**	More information
SWITZERLAND BERN	Neurometabolic and Metabolic clinic **University Hospital Inselspital**	More information
SWITZERLAND GENÈVE	Paediatric metabolic diseases clinic **Hôpital des Enfants - Hôpitaux Universitaires de Genève HUG**	More information
SWITZERLAND GENÈVE	Adult clinic of inborn errors of metabolism **Hôpitaux Universitaires de Genève HUG**	More information
SWITZERLAND LAUSANNE	Adult clinic of inborn errors of metabolism **Centre Hospitalier Universitaire Vaudois CHUV**	More information
SWITZERLAND ST. GALLEN	Metabolic diseases Clinic **Ostschweizer Kinderspital**	More information
SWITZERLAND ZÜRICH	Lysosomal diseases clinic **Universitäts - Kinderspital Zürich - Eleonorenstiftung**	More information
SWITZERLAND ZÜRICH	Metabolic diseases clinic **Universitäts - Kinderspital Zürich - Eleonorenstiftung**	More information
SWITZERLAND ZÜRICH	Rare metabolic diseases clinic **UniversitätsSpital Zürich**	More information

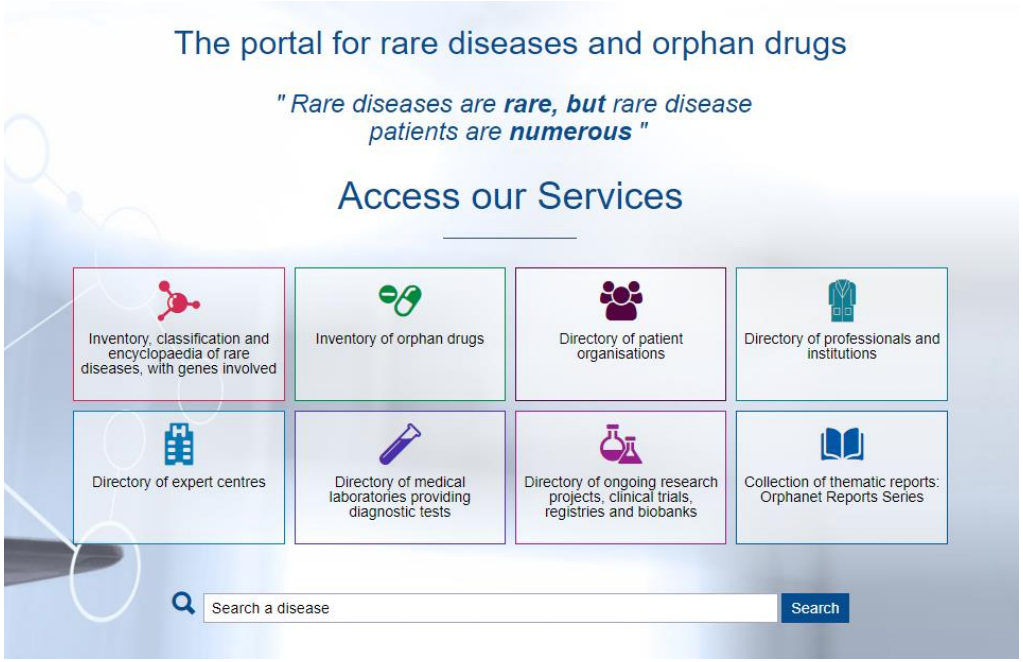
orphanet

Hôpitaux Universitaires Genève

kosek

Coordination nationale des maladies rares

Orphanet website



- Freely accessible site
- Available in 8 languages
- Dedicated to a broad range of users, from patients to healthcare professionals, researchers, policy makers, pharmaceutical companies, etc.

ORPHANET SITE in NUMBERS (2018)
32 million pages viewed
12+ million unique visitors

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Orphanet's missions

- Improve the visibility of rare diseases in healthcare and research systems

- Provide high-quality information and expertise on rare diseases

- Contribute to generating knowledge on rare diseases

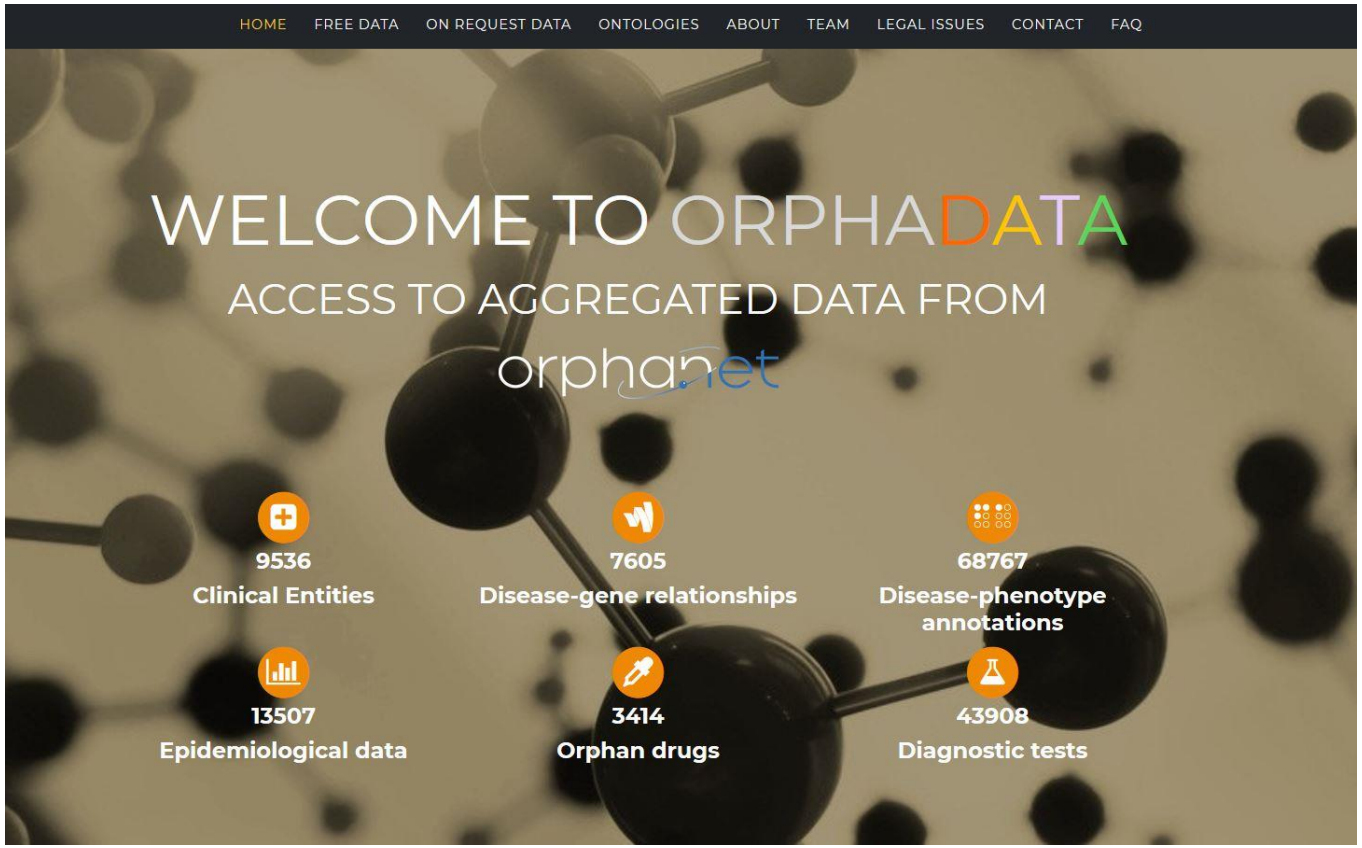
→ piecing together the parts of the puzzle to better understand rare diseases

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Orphanet, a contributor to generating knowledge on RD



www.orphadata.org

Massive aggregated data in a computer-friendly format

Integrable, reusable, essential for research

orphanet

HUG Hôpitaux Universitaires Genève

kosek

Coordination nationale des maladies rares

Orphanet, a contributor to generating knowledge on RD

RARE DISEASES AND CLASSIFICATIONS

Orphanet maintains the Orphanet nomenclature of rare diseases, essential in improving the visibility of rare diseases in health and research information systems: each disease in Orphanet is attributed a unique and stable identifier, **the ORPHA number.** The nomenclature is organised in a poly-hierarchial classification, and data set includes: types of disorders, flags of disorders, new relations between disorders, and characterisation of the alignments between disorders and external terminologies or resources (OMIM, ICD-10, MeSH, UMLS, MedDRA and GARD). The alignments are characterised in order to indicate if the terms are perfectly equivalent (exact mapping) or not. For analysis purposes, each disorder is attributed to a preferred classification by linking it to the head of classification entity.

As some decisions could be made somewhat arbitrarily, we have written a set of rules to make sure attributions are consistent. Hence, users are allowed to access to 3 differents data sets: **Rare diseases and cross referencing, Classifications of rare diseases and Linearization of disorders.**

Orphanet nomenclature files for coding

Cross-referencing of rare diseases

Classifications of rare diseases

Linearisation of disorders

orphanet

Hôpitaux Universitaires Genève

kosek

Coordination nationale des maladies rares

Orphanet, a contributor to generating knowledge on RD

elixir Core Data Resource

ORPHADATA ONTOLOGIES POWERED BY orphanet

An ontology is a structured vocabulary that describes concepts within the same domain, capturing relationships between these concepts.

ORDO

The Orphanet Rare Disease Ontology (ORDO) was jointly developed by Orphanet and the European Bioinformatics Institute (EMBL-EBI) to provide a structured vocabulary for rare diseases, capturing relationships between diseases, genes and other relevant features, forming a useful resource for the computational analysis of rare diseases.

Sparql endpoint

Our freely available data sets can be queried using this SPARQL endpoint.

HOOM

The HPO – ORDO Ontological Module (HOOM) qualifies the annotations between a clinical entity (from ORDO) and phenotypic abnormalities from HPO (Human Phenotype Ontology) according to frequency and by integrating the notion of diagnostic criterion.

www.orphadata.org

Scientific aggregated data in a semantic format

Orphanews newsletter

orphaNews

The newsletter for the rare disease community

WELCOME TO ORPHANEWS

OrphaNews is a freely available, twice-monthly electronic newsletter presenting an overview of scientific and political news about rare diseases and orphan drugs. OrphaNews is produced by Orphanet is intended for the rare disease community. It is supported by the European Commission's DG SANTE (PP-1-2-2018-Rare 2030) and the French Muscular Dystrophy Association (AFM).

In each new issue, OrphaNews reports the latest developments in the field of rare diseases and orphan drugs, including new syndromes, new genes, basic and clinical research, national and international policy, disease surveillance, clinical trial updates, orphan drug approvals, funding opportunities, ethical, social and legal issues, news from the patient associations, upcoming events, and new publications. OrphaNews is suited to all sectors of the rare disease and orphan drugs community - including policy makers, scientists, health professionals, patient representatives, geneticists, members of the biopharmaceutical industry and anyone interested in staying informed of important developments and new initiatives in the field of rare diseases and orphan drugs.

Disclaimer :

The content of newsletter represents the views of the Editorial Board only and is his/her sole responsibility; it can not be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.

Inserm
La science pour la santé
From science to health

orphanet

Co-funded by the Health Programme of the European Union

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Orphanet Switzerland

Hôpitaux Universitaires Genève

Since 2001 member of the Orphanet network

Team:

Coordinator: Dre. Loredana D'Amato Sizonenko
Project manager: Martin Arles
Information scientist: Béatrice Geissbuhler

TELETHON
FONDATION TELETHON ACTION SUISSE

GDK Schweizerische Konferenz der kantonalen Gesundheitsdirektorinnen und -direktoren
CDS Conférence suisse des directrices et directeurs cantonaux de la santé
CDS Conferenza svizzera delle direttrici e dei direttori cantonali della sanità

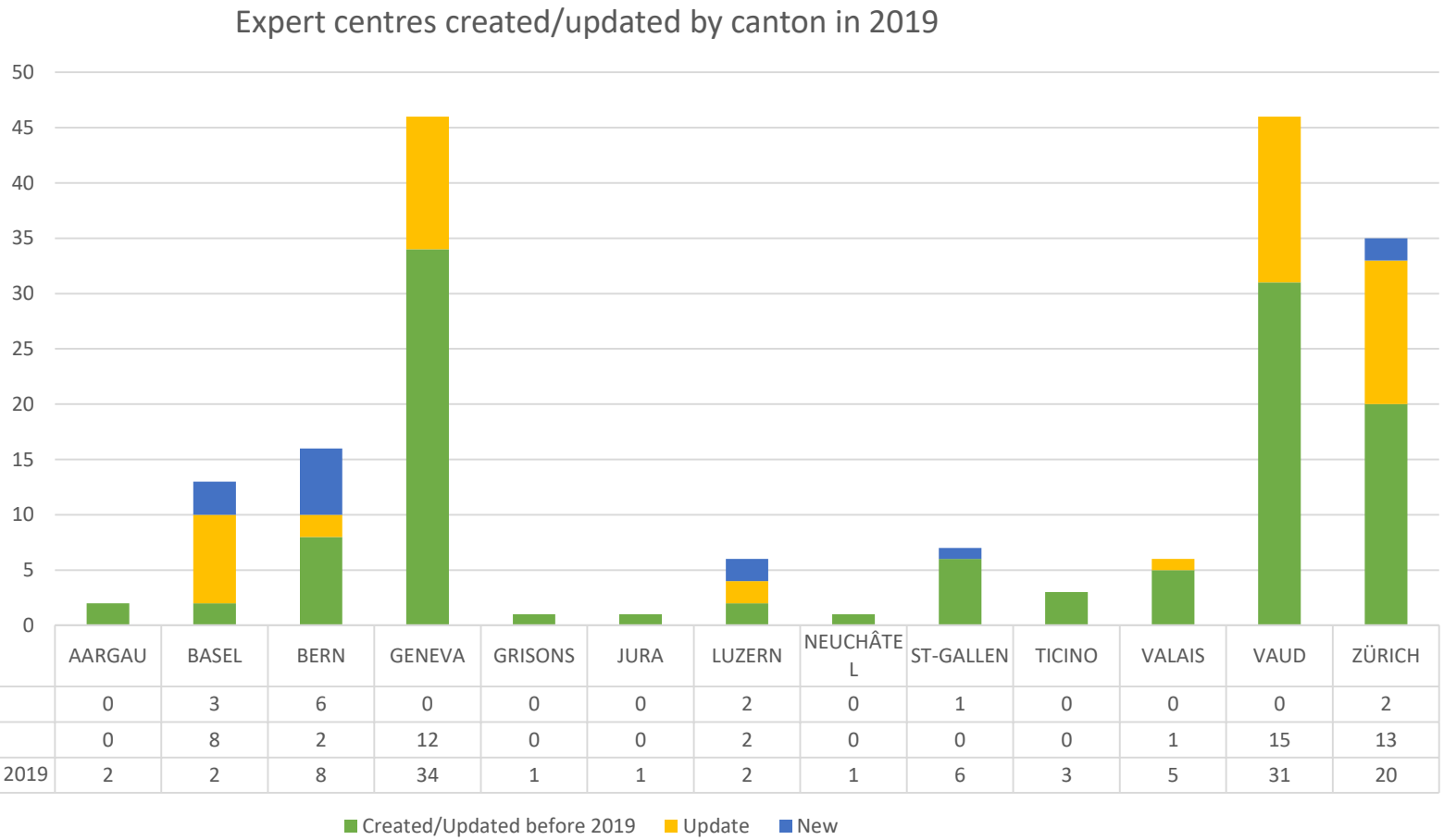
kosek
Coordination nationale des maladies rares

orphanet

Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Switzerland: Number of expert centres by canton



	AARGAU	BASEL	BERN	GENEVA	GRISONS	JURA	LUZERN	NEUCHÂTEL	ST-GALLEN	TICINO	VALAIS	VAUD	ZÜRICH
New	0	3	6	0	0	0	2	0	1	0	0	0	2
Update	0	8	2	12	0	0	2	0	0	0	1	15	13
Created/Updated before 2019	2	2	8	34	1	1	2	1	6	3	5	31	20

orphanet

Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Registration of Expert Centres in Orphanet for Switzerland

A – Orphanet eligibility criteria

The expert centres should deliver a service of indisputably higher quality than a standard hospital service in the relevant speciality.

As there are no centres officially designated by the Swiss health authorities yet, the centres should fulfil most of the Orphanet eligibility criteria (see the registration form), adapted from the European Union Committee of Experts on Rare Diseases recommendations.

The expertise has to be demonstrated by number of rare diseases patients seen, expert advice/second opinion provided, multidisciplinary facilities, referrals inside the country and from abroad, quality management procedures, collaborations and networking, implication in research and systematic data collection, and grants and publications.

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

Registration of Expert Centres in Orphanet for Switzerland

B – Required information (mandatory*)

- Name of the expert centre in local language and in English*: *The name should include the disease/group of diseases covered by the centre (e.g. "Specialized clinic for rare epilepsies" or "centre for neuromuscular diseases")*
- The disease(s) or group of diseases covered by the expert centre*: *you don't need to list all the diseases, the Orphanet Swiss team will send you a proposal based on the Orphanet classification.*
- Indication whether the expert centre is intended for children, adults or both*.
- Indication whether the expert centre is a genetic counselling clinic, a medical management clinic or both*.
- Name and details (email address and phone number, not mandatorily published online), of at least one expert centre coordinator*
- Team members: *a maximum of 3/4 professionals who are directly involved in the clinic*.
- Name and address of the hospital/institution and of the department/service responsible of the expert centre*
- Orphanet eligibility criteria form* (based on EUCERD recommendation)
- Website of the expert centre

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

Orphanet eligibility criteria form

European Union Committee of Experts on Rare Diseases

EUCERD RECOMMENDATIONS

QUALITY CRITERIA FOR CENTRES OF EXPERTISE FOR RARE DISEASES IN MEMBER STATES

orphanet

HUG Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares

Registration of Expert Centres in Orphanet for Switzerland

C - Steps to be followed to register an expert centre in Orphanet

- Contact the Orphanet Swiss team through contact@orphanet.ch to receive a registration form.
- Complete the form with the required information listed above.
- Once the form completed, the Orphanet Swiss team will assess it and, if necessary, will contact you to clarify some data (particularly the list of diseases to be linked based on the Orphanet classification).
- Once validated by the Swiss team, the form will be assessed by the Orphanet coordinating team.
- Once validated by the Orphanet coordinating team, the centre will be published in the Orphanet website and you will be informed.

Registration of Expert Centres in Orphanet for Switzerland

1) How many patients did you see with this disease or group of diseases last year?

- Total number of patients seen last year * :
- Number of new cases last year :
- Percentage of patients from other regions from the country :
- Percentage of patients from abroad :

2) Do you provide expert advice/second opinion to other clinicians (mail, telephone)? * Yes No

Number of expert opinion given last year:

3) Is your centre multi-disciplinary, integrating medical, biological, paramedical, psychological and social needs (such as a rare disease board)? * Yes No

Please explain your answer :

4) Does your centre organise collaborations to ensure the continuity of care between childhood, adolescence and adulthood, if this is relevant? * Yes No

5) Does your centre have appropriate arrangements in place for referrals within your country and from/to other EU countries (if applicable)? * Yes No

if yes, please describe :

orphanet

HUG Hôpitaux Universitaires Genève

kosek Coordination nationale des maladies rares

Thank you for your attention

Contact:
loredana.damatosizonenko@orphanet.ch
martin.arles@orphanet.ch

orphanet

Hôpitaux Universitaires Genève

kosek
Coordination nationale des maladies rares