The background features a light blue gradient with two concentric circles. Two blue arcs, each with a small dark blue sphere at its end, curve across the top and bottom of the slide.

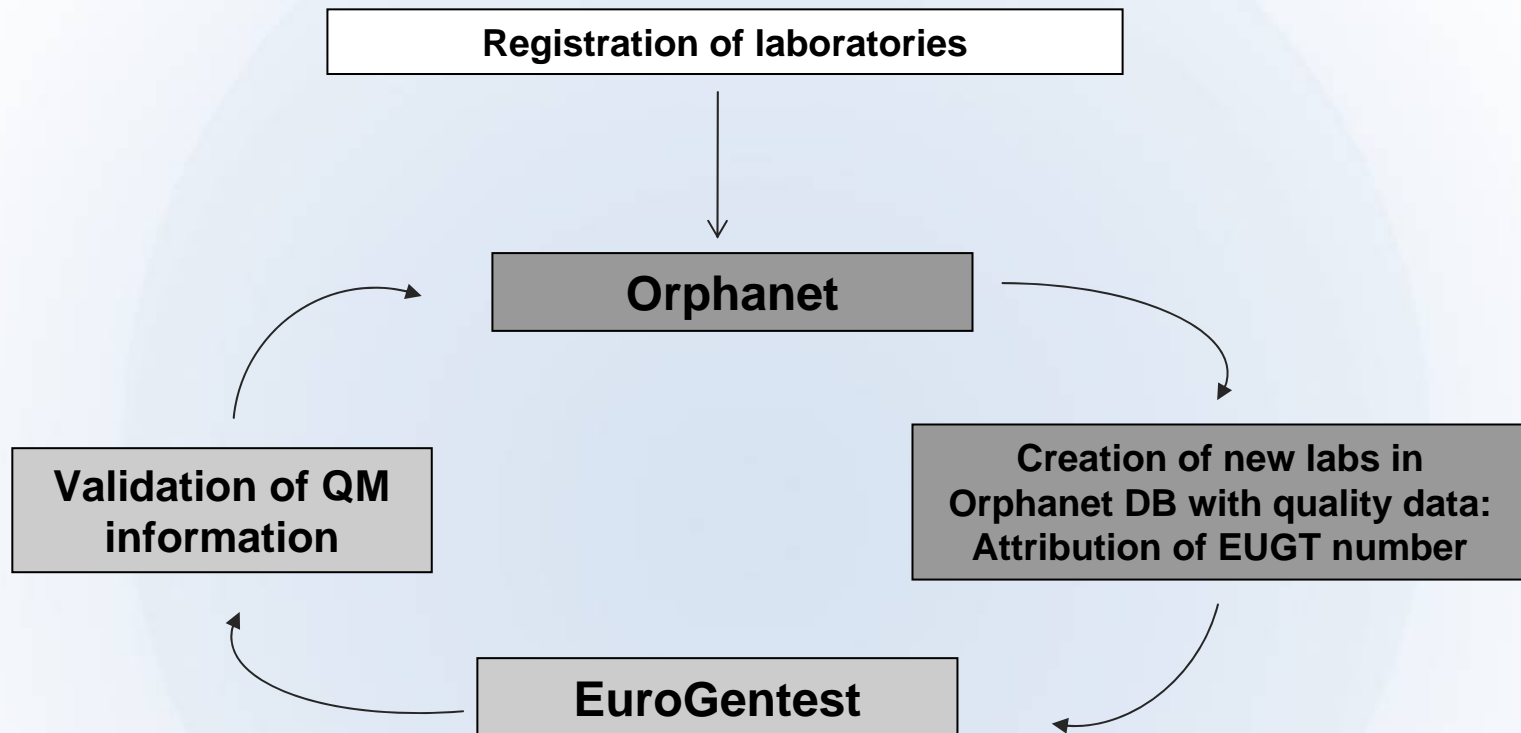
EuroGenTest / Orphanet database of medical laboratories

Ségolène Aymé

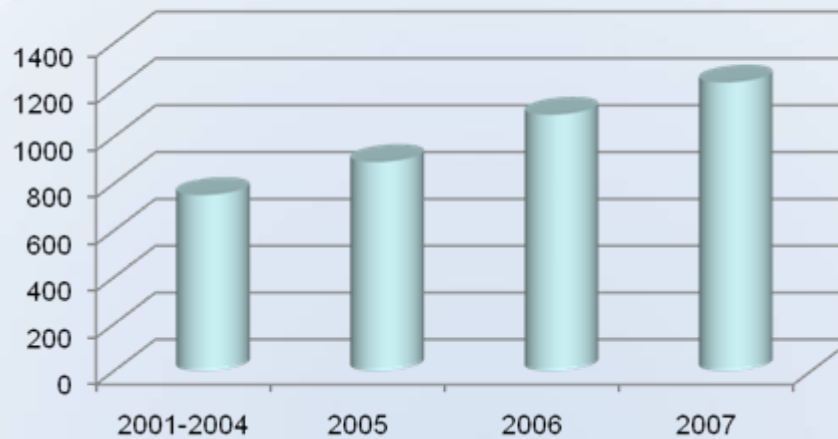
Progress report 2008

- Data collection in 36 countries
 - Algeria, Austria, Belgium, Bulgaria, Croatia, Cyprus, Czech Republik, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Israel, Italy, Latvia, Lebanon, Lithuania, Luxembourg, Malta, Morocco, Netherlands, Norway, Poland, Portugal, Romania, Serbia, Slovakia, Slovenia, Spain, Sweden, Switzerland, Tunisia, Turkey, United-Kingdom
- Information on quality management
 - Accreditation, Certification, licensing
 - Major differences between countries
- Possibility to query by gene
- Link to SwissProt and gene databases

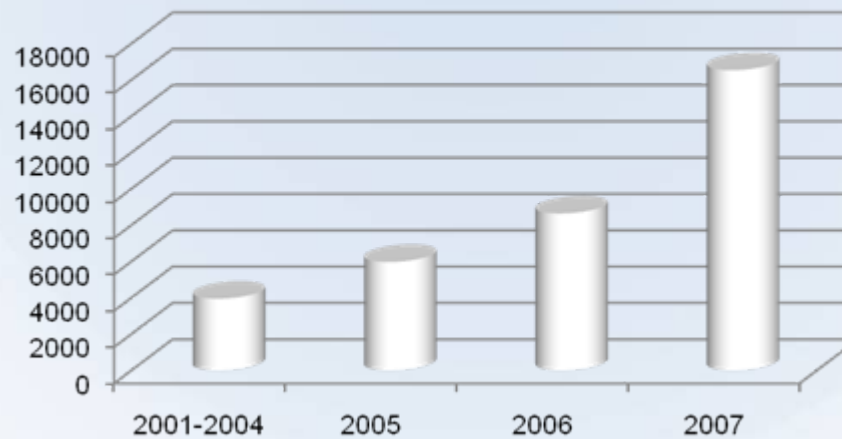
Orphanet - EUGT Unit 1



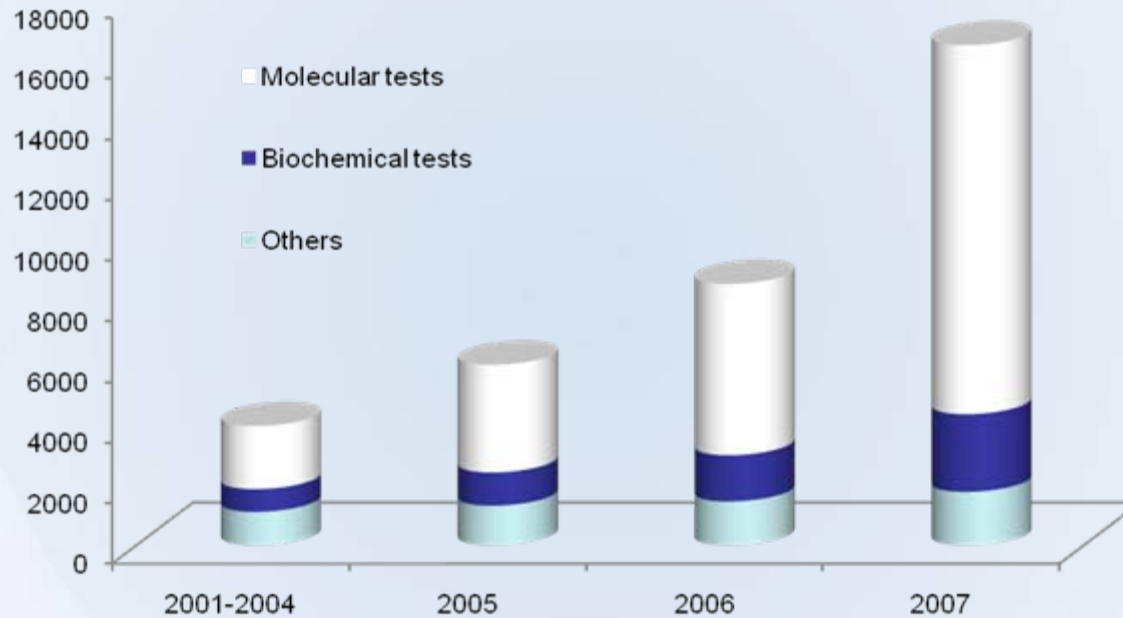
Medical laboratories



Diagnostic tests



Diagnostic tests




Content of the database

- 7,110 diseases
- 1,613 genes linked to 1,283 diseases
- 8,519 abstracts in 5 languages
- 713 review articles
- **1,232 medical labs testing 2,397 diseases**
- 5,222 research projects about 2,197 diseases
- 1,740 patients organizations linked to 2,605 diseases
- 3,184 specialized clinics
- 11,102 professionals

Three ways to search clinical tests/ laboratories

Languages Français | **English** | Español | Deutsch | Italiano | Portuguese

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RARE DISEASES | **ORPHAN DRUGS** | **CLINICS** | **CLINICAL LABORATORIES** | **RESEARCH AND TRIALS** | **PATIENT ORGANISATION** | **DIRECTORY OF RESOURCES** | **EDUCATION**

SIMPLE SEARCH
Enter keyword
> Alphabetical list of rare diseases

OTHER SEARCH OPTION(S)
> Orphan drugs > Patient organisations
> Research and trials > Clinics
> Clinical laboratories > Directory of resources

ORPHANET TODAY
Diseases : 7242
Clinics : 3017
Laboratories : 2838
Professionals : 10136
Daily visitors : 22558

RARE DISEASES
> [Information about a disease](#)
> [Alphabetical list](#)
> [Search by sign](#)
> [Emergency guidelines](#)
> [Patient encyclopaedia](#)
> [Professional encyclopaedia](#)
> [About Rare diseases](#)

RESOURCES DIRECTORY
> [Clinics](#)
> [Reference centres](#)
> [Clinical laboratories](#)
> [Research projects](#)
> [Registries / databases](#)
> [Professionals](#)
> [Patient organisations](#)
> [EuroGentest](#)
> [Register your activity](#)


Improve the quality of medical care for Rare Diseases.
Provide adapted services to the rare diseases community
[About Orphanet](#) | [Quality charter](#)
[Register your activity](#) | [Support Orphanet](#)

Services for professionals
> [Professional encyclopaedia](#)
> [Search by sign](#)
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Services for patients
> [Patient encyclopaedia](#)
> [Information about a disease](#)
> [Research projects](#)
> [Contact other patients / families](#)
> [Training sessions](#)

NEWS
.....
[Orphanet's 10th birthday Conference presentation highlights](#)
.....
[EC Rare Disease webpages](#)
.....

EuroGentest link



Simple search: by gene name

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Search Encyclopaedia for patients Encyclopaedia for professionals Emergency guidelines Search by sign

Homepage » Rare diseases » Search

SIMPLE SEARCH

MECP2

☐ Disease name
☒ Gene name or symbol
☐ MIM number
☐ ICD 10 code
☐ Orpha number

OTHER SEARCH OPTION(S)
> Alphabetical list

→ OK

::: 1 Result(s)

Methyl CpG binding protein 2 (Rett syndrome)

Print

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Homepage » Rare diseases » Search

SIMPLE SEARCH

MECP2

☐ Disease name
☒ Gene name or symbol
☐ MIM number
☐ ICD 10 code
☐ Orpha number

OTHER SEARCH OPTION(S)
> Alphabetical list

→ OK

::: 5 Result(s)

Angelman syndrome

Autism

Mental retardation - progressive spasticity, X-linked

Mental retardation X-linked - psychosis - macroorchidism

Rett syndrome

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Homepage » Clinical laboratories » Search

SIMPLE SEARCH

rett-syndrom

☒ Disease name
☐ Gene name or symbol

Country: **All countries**

☐ Accredited laboratories
☐ EQA participating laboratories
☐ Certified laboratories

:76 Result(s)

Sort by Quality management ☐ Location ☒

Caption : Accredited=■; Certified=●; EQA=▲;

AUSTRIA	WIEN	WIEN	Molekulare Diagnostik Martin GENCIK More information
BELGIUM	ANTWERPEN	EDEGEM	Molecular diagnosis Antwerp University Katrien STORM More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	Molecular diagnosis Hôpital Erasme Pascale COCHAUX More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	Molecular diagnosis Université catholique Nathalie LANNON More information
BELGIUM	HAINAUT	GOSSELIES	Molecular diagnosis

Homepage » Clinical laboratories » Search

SIMPLE SEARCH

rett-syndrom

☒ Disease name
☐ Gene name or symbol

Country: **BELGIUM**

☐ Accredited laboratories
☐ EQA participating laboratories
☐ Certified laboratories

:7 Result(s)

Sort by Quality management ☐ Location ☒

Caption : Accredited=■; Certified=●; EQA=▲;

BELGIUM	ANTWERPEN	EDEGEM	Molecular diagnosis of Rett syndrome (MECP2 gene) Antwerp University hospital Katrien STORM More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	Molecular diagnosis of Rett syndrome (MECP2 gene) Hôpital Erasme Pascale COCHAUX, Catherine RYDLEWSKI, Jasmine PARMA More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	Molecular diagnosis of Rett (MECP2 gene) Université catholique de Louvain Nathalie LANNON More information
BELGIUM	HAINAUT	GOSSELIES	Molecular diagnosis of Rett syndrome (MECP2 gene)

Karin SEGERS
[More information](#)

BELGIUM	OOST-VLAANDEREN	GHEENT	▲ Molecular diagnosis of Rett (MECP2 gene) Ghent University Hospital Kathleen CLAES, Elfride DE BAERE More information
---------	-----------------	--------	---

Click! for more information on this diagnostic test

**Click! for
'more information about
this location'**

[Orphanet](#) | [Rare diseases](#) | [Orphan drugs](#) | [Clinics](#) | [Clinical laboratories](#) | [Research projects](#) | [Research and trials](#) | [Patient organisations](#)

::Laboratory for molecular genetics

Print 

Center for medical genetics
Ghent University Hospital
De Pintelaan 185
9000 GHENT
BELGIUM

Phone: 32 9 2403603
Fax: 32 9 2404970
[Website](#)

Type(s): -
Status : Public hospital
Other services provided : -
EUGT number : EUGTBE55694

CONTACTS

Director of laboratory : [Pr Paul COUCKE](#)
Director of department : [Pr Anne DE PAEPE](#)
Quality manager : [Ms Greta VAN DER CRUYSEN](#)
Other professionals : [Dr Kathleen CLAES](#), [Pr Paul COUCKE](#), [Dr Elfride DE BAERE](#)

Quality management

Accreditation and EQA participations are validated by EuroGentest

■ Accreditation

Accreditation [ISO 15189](#) by [BELAC](#) in 2004 (on-going)

● certification

certification [Government authorization](#) by [Government](#) in 1987 (effective)

▲ EQA

[CF](#) : participation 2004, organised by [CF Network](#)

[CF](#) : participation 2005, organised by [CF Network](#)

[DNA sequencing](#) : participation 2004, organised by [EMQN](#)

[FRAXA](#) : participation 2004, organised by [EMQN](#)

[HD](#) : participation 2004, organised by [EMQN](#)

[HFE](#) : participation 2004, organised by [EMQN](#)

[HNPCC](#) : participation 2004, organised by [EMQN](#)

[PW/AS](#) : participation 2004, organised by [EMQN](#)

■ ■ ■

RELEVANT INFORMATION

additional information

- > clinical laboratories (54)
- > research projects (1)
- > clinical trials (1)
- > registries/databases (0)

related services

- > [eurogentest](#)
- > educational tools

getting involved

- > register your activity

SIMPLE SEARCH

cystic fibrosis

☒ Disease name

☐ Gene name or symbol

Country

☐ Accredited laboratories

☐ EQA participating laboratories

☐ Certified laboratories

Quality management definitions:

- Accreditation
- Certification
- External Quality Assessment
- Licensing

Simple search: This screen gives access to lists of clinical laboratories by country and the type of quality management (accredited, certified, EQA participating). You may use truncated names. A list of all names may be obtained by clicking on the "All countries" button.

Search by region: This screen gives access to lists of clinical laboratories by geographical area. You must select a country and a region. You may also query a laboratory speciality (instead of a region).

Search a laboratory: This screen provides access to lists of clinical laboratories by geographical area, name, or number. The country has to be selected first. You may also query a laboratory speciality (instead of a region).

Search a professional: This screen provides access to lists of clinical laboratories by geographical area, name, or number. The country has to be selected first. You may also query a laboratory speciality (instead of a region).

Search by licensing: This screen provides access to lists of clinical laboratories by geographical area, name, or number. The country has to be selected first. You may also query a laboratory speciality (instead of a region).

Clinical laboratories listed in *Orphanet* are those offering a test for the diagnosis of a rare disease or of a group of rare diseases and those performing genetic testing whatever the prevalence of the disease. The lists are established in collaboration with *EuroGentest* which is also in charge of the validation of the data on accreditation and on *EQA* (External Quality Assessment) of the laboratories. Data on certification are based on self-reporting of the laboratories.

The information which is displayed is provided by the scientists in charge of the activities. They must apply to have their laboratory activity listed. The applications are reviewed by the member of the *Orphanet* scientific advisory board of the country in charge of the relevant speciality.

Accreditation is a "procedure by which an authoritative body gives formal recognition that a body or person is competent to carry out specific tasks" (*ISO 9000, 2000 Quality management systems – fundamentals and vocabulary*).

Accreditation standards related to clinical laboratories (for example, *ISO15189*) place emphasis on having an effective quality insurance system in place, on a commitment to meeting needs of patients and their doctors as users of laboratory services and a need for continuous cycle of quality improvement at the centre of all policy making an operational decisions. Data on accreditation have been validated by *EuroGentest* before their release on *Orphanet*.

Certification is a procedure by which a third party gives written assurance that a product, process or service conforms to specific requirements. (*ISO 9000, 2000 Quality management systems – fundamentals and vocabulary*).

EQA (External Quality Assessment, also known as proficiency testing) is "a system whereby a set of reagents and techniques are assessed by an external source and the results of the testing laboratory are compared with those of an approved reference laboratory" (*WHO*). It allows a laboratory to compare its performance for an individual test or technique against that of other laboratories. Data on *EQA* participation have been validated by *EuroGentest* before their release on *Orphanet*.

Licensing is a legal permit or formal permission from a constituted authority or governmental agency to operate a laboratory. It may involve documenting the existence, institutional accountability and the activities of the facility, for example, the type of service provided. In return the laboratory is officially registered and may be publicly listed.

Data collection takes place in European and surrounding countries and is currently ongoing. The database cannot yet be regarded as comprehensive. If an expert clinical laboratory is not listed in a region or a country, it may not have been identified or the laboratory may have refused to be listed. It is also possible that it does not exist as diagnostic tests and genetic testing facilities do not exist for all for all rare diseases or in every region.

Homepage » Clinical laboratories » Search

SIMPLE SEARCH

cystic-fibrosis

Disease name
Gene name or symbol

Country: All countries

Accredited laboratories
EQA participating laboratories

→OK

OTHER SEARCH OPTION(S)

- > Search by region
- > Search by city
- > Search by laboratory
- > Search by professional
- > Search by licensing

287 Result(s)

Sort by

Quality management
Location

→OK

Caption: Accredited=■; Certified=●; EQA=▲;

AUSTRIA	OBERÖSTERREICH	LINZ	Molekulare Diagnostik der Zystischen Fibrose A. o. Krankenhaus der Elisabethinen Helmut MITTERMAYER, Christoph LUGER More information
AUSTRIA	SALZBURG	SALZBURG	Molekulare Diagnostik der zystischen Fibrose Paracelsus Medizinische Privatuniversität Salzburg Olaf RITTINGER More information
AUSTRIA	STIEBERMARK	GRAZ	Molekulare Diagnostik der zystischen Fibrose Medizinische Universität Graz Klaus WAGNER, Peter KROISEL, Hannelore ZIERLER More information
AUSTRIA	TIROL	INNSBRUCK	Molekulare Diagnostik der Zystischen Fibrose Leopold-Franzens-Universität Innsbruck Barbara GÜNTHER, Gerd UTERMANN More information
AUSTRIA	TIROL	INNSBRUCK	Molekulare Diagnostik der zystischen Fibrose Medizinische Universität Innsbruck Wolfgang VOGEL More information
AUSTRIA	WIEN	WIEN	Molekulare Diagnostik der zystischen Fibrose Medizinische Universität Wien Christa FONATSCH, Robert WIESER More information

Homepage » Clinical laboratories » Search

SIMPLE SEARCH

cystic-fibrosis

Disease name
Gene name or symbol

Country: BELGIUM

Accredited laboratories
EQA participating laboratories
Certified laboratories

→OK

OTHER SEARCH OPTION(S)

- > Search by region
- > Search by city
- > Search a laboratory
- > Search by professional
- > Search by licensing

8 Result(s)

Sort by

Quality management
Location

→OK

Caption: Accredited=■; Certified=●; EQA=▲;

BELGIUM	ANTWERPEN	EDEGEM	▲ Molecular diagnosis of cystic fibrosis (CFTR gene) ● Antwerp University hospital Wim WUYTS, Katrien STORM More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	Molecular diagnosis of cystic fibrosis (CFTR gene) Hôpital Erasme Pascale COCHAUX, Catherine RYDLEWSKI, Jasmine PARMA More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	Molecular diagnosis of cystic fibrosis (CFTR gene) Universitair Ziekenhuis Brussel Ingeborg LIEBAERS More information
BELGIUM	ARRONDISSEMENT BRUSSEL HOOFDSTAD	BRUSSELS	▲ Molecular diagnosis of cystic fibrosis (CFTR gene) ● Université catholique de Louvain Xavier PEPEMANS More information
BELGIUM	HAINAUT	GOSSELIES	▲ Molecular diagnosis of cystic fibrosis (CFTR gene) ● Institut de Pathologie et de Génétique Pascale HILBERT More information

Search by professional

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RARE
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SEARCH BY PROFESSIONAL

Search by professional:

Country:

[→OK](#)

OTHER SEARCH OPTION(S)

[> Simple search](#)

::: Dr Pierre HUTTER

[Print](#)

Activities for Orphanet:

pierre.hutter@ichv.ch

Phone: -

Fax: -

::: 2 Result(s)

[Dr HUTTER Pierre](#)

[Pr KOHLSCHÜTTER Alfred](#)

Laboratoire de Diagnostic
Oncogénétique
Laboratoire de Référence Suisse
Romand pour le Cancer Colorectal
Familial
Centre Médical Universitaire
1 Rue Michel Servet
1211
GENEVE
SWITZERLAND

[More informations about this location](#)

Phone : + 41 (0)22 379 58 13
Fax : + 41 (0)22 379 58 97

Division de Génétique Médicale
Institut Central des Hôpitaux
Valaisans (ICHV)
86, avenue Gd-Champsec, Case
Postale 520
1951
SION
SWITZERLAND

[More informations about this location](#)

Phone : 00 41 27 603 48 53
Fax : 00 41 27 603 49 95
[Website](#)

RELEVANT INFORMATION

additional information

- > clinics (0)
- > clinical laboratories (11)
- > research projects (0)
- > patients organisations (0)

article(s) for orphanet

- > article(s) (0)
- > summaries (0)

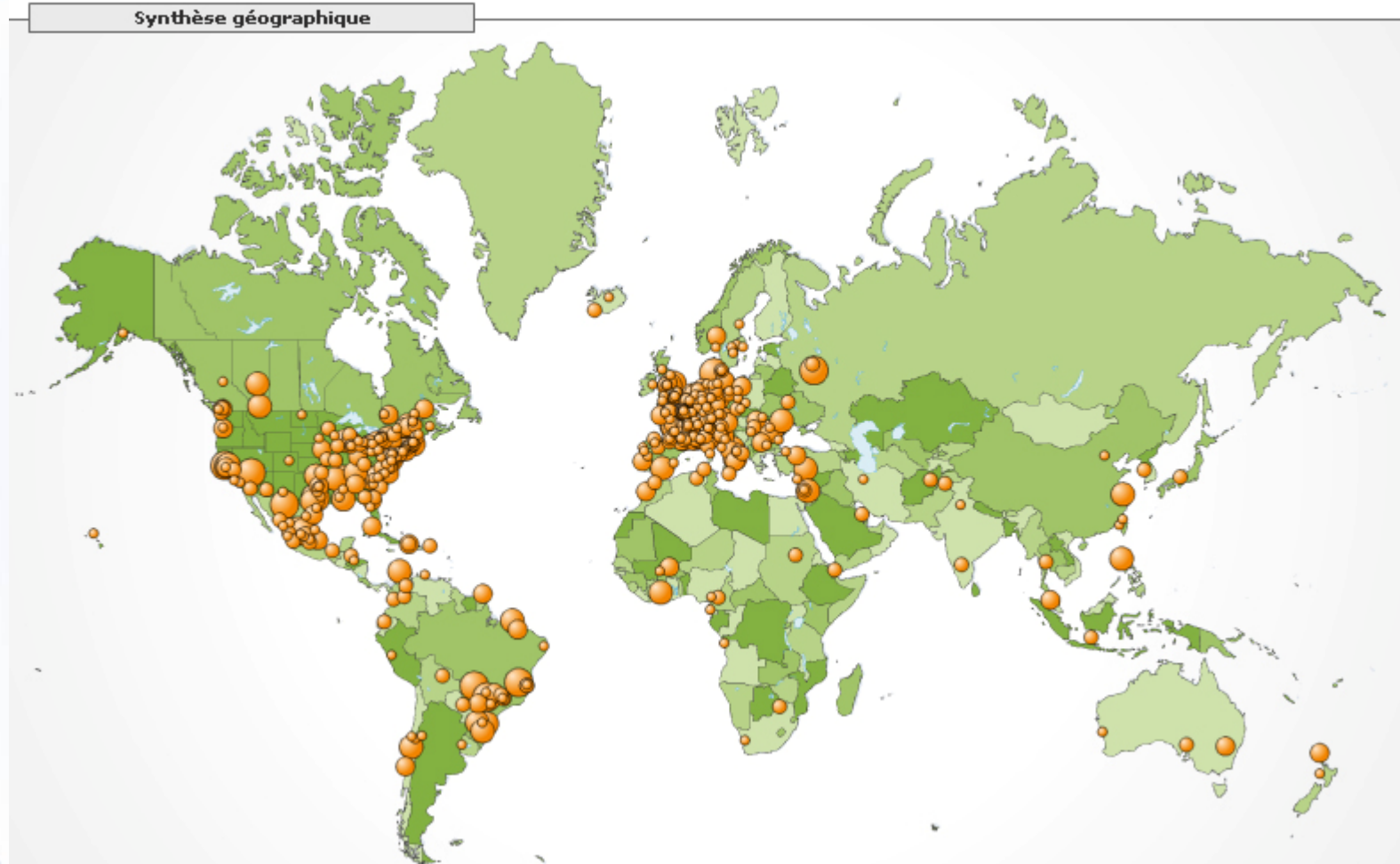
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Orphanet Worldwide users: 20,000 daily



Partnership with GeneTests

- Exchange of files for quality control
 - Couples gene-disease
- Comparison of tests offered
 - To identify tests only offered on one continent
- Results
 - Very few discrepancies
 - Difficulties due
 - to differences in classification
 - Separation of molecular and biochemical tests in Orphanet



Further analysis for final conclusions

Report at the next meeting !