


October 2007

 **This month's TO-DO list** 

## **V4 database evolution**

The V4 database is progressing slowly, but safely.

The Informatics team works hard: first data migration (to provide material for the first floor Major V4 tests), adjustments after first tests, screens development...

Database and editorial teams in Paris are testing Major V4: first floor tests were done mid-October, second floor tests are on their way.

All existing V3 data will be transferred to V4 (diseases, clinics, research projects, clinical tests, support groups, contact persons, addresses etc.) and only new data will be injected. Members of the database and editorial team in Paris are creating the new tables for genes, genetic testing quality data, epidemiology and functional classifications. They will be injected after the definitive migration.

As soon as V4 is available and effective throughout the whole database, you will all test the new version before its release.

One question that you might have asked yourself is: “when should we stop registering new data and updating in V3?” The answer is: just before the final migration.

The informatics service will inform us early enough when Major V3 registering is to be frozen, currently estimated by the end of December, too.

## **This month's TO-DO list**

### **Use of Orphanet database agreement**

We remind you that we have prepared an [agreement](#) in order to specify the rules of use of the files containing country-specific data and the complete list of diseases (data/5files).

This agreement is **to be printed and filled in duplicate**, signed by the duly authorized representative of the partner institution (not by the Orphanet national coordinator), and sent to the French team in order to be signed by Inserm (one copy will be sent back to the partner).

It is very important to sign this agreement since the country-specific data and the complete list of diseases are very precious data that must be protected. The agreement specifies that they can be freely used by the partners only within the framework of their activity on behalf of Orphanet. However, partners are encouraged to offer these data to national institutions in order to promote fruitful partnerships. A number of "derivated products" (i.e. classifications of diseases by medical specialty) can also be used to create partnerships with different kinds of organisations.

In any case, and as specified in the agreement, the partnership must receive the authorization of the Orphanet executive management board, and an agreement should be signed with the third party.

### **Orphanet partners' meeting on Monday 26 November in Lisbon**

The second partners' meeting in the framework of our RD portal contract will take place the 26th of November in Lisbon.

You will find the [agenda](#) and the [list of participants](#) enclosed.



## What we did



*Estonia:* Prof. Andres Metspalu, the Orphanet coordinator, and Prof. Tiina Talvik, the Chairman of the National scientific advisory committee, were asked by the Estonian Ministry of Social Affairs to provide **Estonian suggestions for future policies on rare Diseases** in the European Union. They both filled in the questionnaire for the Public Consultation on the Commission Communication on rare diseases in the framework of the EU Task Force on Rare Diseases.



*Italy, 21-23 September:* the second meeting of the **INSIEME project** took place in Rome. Prof Bruno Dallapiccola made a speech entitled ‘La Malattia Rara: la difficoltà di ricevere una diagnosi tempestiva’ (Rare Disease: the difficulty to have a tempestive diagnosis). The project ‘INSIEME – Giornate di socializzazione per malati affetti da patologie rare’ (in English: TOGETHER – Days of socializing for rare diseases patients) had been proposed by the Italian Federation for Rare Diseases – UNIAMO FIMR onlus – at the occasion of the 2007 notification of the Ministry of Social Solidarity for Associations for Social Promotion. This initiative received funding for the realisation of three weekends of socializing for rare diseases patients and their families, one for each macroarea of Italy (North: Milan, June 29-30 and 1 July; Centre: Rome, September 21-23; South: Messina, October 26-28).

*Milan, 4-6 October:* Prof. Bruno Dallapiccola took part in the fourteenth national convention of S.I.O.H., ‘**Società Italiana di Odontostomatologia per l’handicap**’ (‘Italian Association for Disability and Oral Health’, IADH). He made a speech entitled ‘Basi genetiche dei difetti cranio-facciali e odontostomatologici’ (Genetic bases for craniofacial and odontostomatologic defects). The convention was dedicated to the promotion of the oral health in disabled people.

*Bologna, 6 October:* Prof. Bruno Dallapiccola attended the convention ‘Il trattamento multidisciplinare delle dismorfosi cranio e dento-facciali in età evolutiva’ (**Multidisciplinary treatment for cranio and dentofacial dysmorphology in evolutive age**) making a speech entitled ‘Le basi genetiche dei difetti craniofacciali’ (Genetic bases for craniofacial defects). The objective of the convention is to provide updated information to specialists about the multidisciplinary treatment of these pathologies and to point out the role of associations and the psychological aspects concerning the patient and his/her relatives.

*L’Aquila, 20 October:* Prof. Bruno Dallapiccola attended the **fourth update course of pediatric surgery** ‘Le sindromi malformative: quale iter diagnostico?’ (Malformative syndromes: which diagnostic procedures?). He made the speech entitled ‘Genetica e difetti congeniti’ (Genetics and congenital defects) and took part as moderator in two round table discussions: ‘Sindrome di Noonan: approccio multidisciplinare’ and ‘Sindrome di Williams: approccio multidisciplinare’ (Noonan syndrome: multidisciplinary approach; Williams syndrome: multidisciplinary approach).



*United-Kingdom, 5 October:* Emma Gillaspay attended **the Dyscerne Network Launch Meeting** and presented Orphanet to the partners. Several ideas for collaboration between the initiatives were suggested during the launch meeting. An official partnership has been agreed upon, creating strong links between the European Network of Centres of Dysmorphology and Orphanet.

***About Dyscerne:** This DG Sanco funded project, headed by Prof Jill Clayton-Smith, will run for three years and comprises six designated Centres of Expertise in Dysmorphology (UK, Belgium, France, Italy, Netherlands and Poland). The University of Manchester will be the coordinating and managing centre for the network. The main aims of the project are to raise current standards for the diagnosis and management of rare dysmorphic syndromes, and to improve dissemination of information on these conditions. The project will identify existing centres of expertise for dysmorphology, and create a formal network. A web-based electronic dysmorphology diagnostic system will be established, enabling clinicians to submit difficult to diagnose cases electronically to an expert diagnostic panel. Recommendations and opinions from the panel will be collated and sent back to the referring clinician. Dyscerne will also serve as a model for future EU Networks of Centres of Expertise. For more information on the project, contact Pam Griffiths ([pam.griffiths@cmmc.nhs.uk](mailto:pam.griffiths@cmmc.nhs.uk)).*

*19 October:* Emma Gillaspay was interviewed by Isabel Pereira Santos from the Portuguese national television about Orphanet. This interview formed part of the preparation for several news reports concerning health in the European Union.

*October:* Emma Gillaspay negotiated a hyperlink to Orphanet directly from the front page of the British Society of Human Genetics website (<http://www.bshg.org.uk/>).



*Spain, 25 October:* Orphanet has been the focus of an [article](#) published in “El Pais”, the leading national newspaper.



*Paris, Hôpital Necker, Centre of reference for juvenile arthritis, 12 October:* Orphanet-France was invited to the Reference Centre for Juvenile Arthritis annual meeting at Necker Hospital, to present services that can be provided for rare diseases centres of reference and patients' support groups. The meeting attendees included doctors from several French reference centres and representatives of patients' support groups. The French language presentation given by Dr. Ioana Caron is available [here](#).

*France, St Malo, 13 October:* Orphanet, represented by Ségolène, was the guest star of a session at the Annual Meeting of the French Syndicate of Pharmacists in Private Practice. This session was dedicated to the launch of a foundation intended to support the dissemination of information on rare diseases to the public. The foundation plans to distribute the Orphanet directory to every pharmacy store and to display the Orphanet website address and "Maladies Rares Info Service" helpline number. As, each day, pharmacists come in contact with over 1 million people, this is a great opportunity to distribute RD information to a large audience.

*Paris, Palais des Congrès, 18 October:* Orphanet was invited to the INSERM stand at **Wonca Europe 2007**, the first European conference of general practitioners in France, a meeting that brings together about 4000 physicians from Europe and overseas.

Wonca stands for "The World Organisation of National Colleges, Academies and Academic Associations of General Practitioners/Family Physicians" or in short "The World Organisation of Family Doctors": it is a world-wide organisation representing general practitioners and family doctors.

This was an opportunity to present Orphanet services via a continuous French/English presentation throughout the days. Leaflets describing Orphanet Encyclopaedia, Directory of Services, OrphanXchange and the newsletters Orphanews and Orphanews Europe were handed out to general practitioners who visited the stand. The Internet connection permitted to present all these activities live.

*Italy, Trento, 19 and 20 October:* Ana Rath reported the Orphanet experience at the **2nd MAGI International Congress on Medical Genetics**. MAGI, an international association on medical genetics lead by Dr Matteo Bertelli and based in Lago del Garda in Northern Italy, helps patients' support groups working together and funds research projects on genetic diseases. This second congress dealt with progress in diagnosis and therapeutics for genetic and rare diseases. Ana presented the different Orphanet services and contents, as well as their ongoing evolution. It was the opportunity to meet very different people with an interest on rare diseases as their common denominator: researchers, oncologists, genetic counsellors, dentists ... as well as professionals working on rare diseases patients' information in Italy, i.e. the Mario Negri Institute.



*Italy, Trieste, 30 October:* Segolene was invited to participate in a WHO meeting to present the methodology put in place to revise the current International Classification of Diseases in the field of rare diseases. She presented the current content of the Orphanet database which is going to be the basis for the proposed new classification. The task is, of course, enormous, but very exciting nevertheless.

*France:* Orphanet achievements and future developments were presented by Ségolène at three regional meetings, organised by the French patients' umbrella organisation "Alliance Maladies Rares" ; in Angers on 11 October, in Marseille on 25 October and in Nice on 8 November. These meetings intend to facilitate a public debate on the effects of the National Action Plan for Rare Diseases at the regional level in France. Additional regional meetings are scheduled for 2008.



*Copenhagen, 18-19 October:* The 2007 EPPOSI partnering workshop took place in Copenhagen on 18 and 19 October. One session was dedicated to the assessment of the number of treatable rare diseases, during which Ségolène presented the epidemiology data collected by Orphanet. The Orphanet teams were represented by Annick Dubosq and Louise Taylor from France, Kathrin Rommel from Germany, Loredana d'Amato-Sizonenko from Switzerland, and Karen Brondum from Denmark. Next year's EPPOSI partnering workshop will be held in Paris in October: a workshop not to miss!



### Who we met



*Switzerland, Lausanne, 15 October:* Dr Loredana D'Amato Sizonenko, met the vice president of "AEMO - Association Enfance et Maladies Orphelines" in order to discuss a Swiss National Alliance of Rare Diseases. Within the framework of the data update of Orphanet Switzerland, the contact with the representatives of several patients' support groups permitted the observation that there is much interest for a national alliance. AEMO aims to inform the public on rare diseases and to collect funds in order to identify and financially support families affected by rare diseases. The association is willing to establish a partnership with Orphanet Switzerland.

### **Stormy period at Orphanet-Portugal**

The Portuguese Orphanet team is facing difficulties due to the restructuration of genetic services in the context of a complete change in the organization of the whole Public Administration. In this context the Institute de Genetica Medica Jacinto Magalhães in Porto has been closed and fused to the National Institute of Health Ricardo Jorge, and will be called the Centre of Medical Genetics JM. The Director has not been nominated yet. The Genetics department will be run by Dr Luis Nunes from Lisbon who will run it while assuming his other positions (Director of Genetics Service at Hospital D Estefania, Professor at the National Public Health School, consultant at General Directorate of Health, President of Portuguese SHG,...). He is trying to get a consensus between Porto and Lisbon..In this context , Daniel Osorio left the Orphanet team in May for his PhD, and in July Jorge was dismissed from the institute (as well as many others now...). Margarida Lima is the only professional to confront the work load. She is expecting to be able to recruit someone remaining who currently teaches information technology at a high school in Porto. His contract is delayed because of the institution's fusion and administrative changes. We wish a prompt recovery to the team !

### **New team organisation at Orphanet-France**

As announced in the last edition, Ioana Caron is taking over the position of Valerie Thibaudeau as Coordinator of the European teams. If you have not heard from her yet, don't be alarmed! The Paris team is heavily involved in the testing of the V4 website and in the preparation of new datasets for the database. In addition, the replacement for Ioana's previous position (in charge of clinics and patients organisations), will not be able to join the France team until January 2008 so she has twice the amount of work!



### *Fundings, projects, software evolution...*

#### **The European Commission thanks Orphanetplatform partners for their work!**

We received the letter of acceptance after the second report of the contract between INSERM and the European Community covering the period from 01/04/05 to 30/06/06 (you can download it on the [Orphanplatform website](#)). The letter was attached to an email of the European Commission "thanking the whole consortium for the work performed, and the results achieved through this project, which is important for the field of rare diseases".

## Here comes Switzerland again with its 26 cantons!

In order to assure the 2008 funding, Orphanet Switzerland's activity report for the year 2007 was sent to the 26 cantons + Liechtenstein. To date, approximately 10 cantons have not yet assured their financial support. This entailed a time consuming administrative task because every file included a personal letter to each of the 26 Cantonal Ministers of Health and... had to be translated in German when needed, that is for 20 cantons!



## New Orphanet data

### *Disease creation and correction*

We remind you that the disease database is frozen until V4 is launched.

So, we will not have the monthly disease meeting until November. However, you can always send suggestions of creations/deletions/modifications to [Marie](#) and [Ana](#). The Paris team is continuing to work in “off mode” and preparing the future disease meetings.

### *Clinics/classifications creations*

As you might have noticed, last month there was **no more creation of new types of clinics**. “Types of clinics” will no longer be used in V4, along with “pools” of diseases. The function of these two entities will be replaced by ‘functional classifications’. This change is meant to make for easier and more precise linking and searching for a resource on a disease or a group of diseases in the database.

As a matter of fact, as has been done previously with clinical tests, research projects, support groups... clinics will be linked to classifications directly to diseases. In a classification, each disease can be a ‘parent’ of one or more other disease, and, at the same time, can be a ‘child’ of one or many diseases. One link between an activity and a disease is distributed to all the ‘children’ of the disease.

### *New Orphanet publication*

#### **New Orphanet Report Series!**

Nine new Orphanet Report Series have been published this month. You can now read issue 6 of “The prevalence of rare diseases: A bibliographic study” in English, **French, German and Italian!** The updated “List of marketing authorised orphan drugs in Europe” is now available in English, **French, German, Italian and Spanish**. Please disseminate these important documents.



## Tools for your mission

### *Zoom on a procedure point*

#### **New V4 quality data on genetic testing**

As was written in the last OrphaNetWork (see column 'New identified data sources', p. 10) the new version of the database (V4) opens access to registration of new data.

In addition to this article, we'd like to focus on the collected information on quality control of listed labs that will permit **quality data on genetic testing** to be displayed in the next version of Orphanet.

Collecting quality data on genetic testing is the result of a close collaboration of EuroGenTest and Orphanet.

There are several types of quality data collected :

- **licensing** (the permission, permit from a governmental agency to operate a laboratory)
- **accreditation** (procedure by which an authoritative body gives formal recognition that a body or person is competent to carry out specific tasks)
- **EQA** (External Quality Assessment: a system of objectively assessing the laboratory performance by an outside agency. EQA 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. The main objective of External Quality Assessment is to establish inter-laboratory compatibility)
- **certification** (procedure by which a third party gives written assurance that a product, process or service conforms to specific requirements)

Maybe you have already collected this information in your registration forms, page 2-3.

If you haven't done so yet, please take a look at this form: for each quality data, if available, it has to specify the EQA provider, the accreditation body and the standard, the certification body and the standard.

When you contact clinical labs, please ask them if they have an accreditation, certification or another quality data. Meanwhile this is not mandatory for Orphanet registering.

Please, check if you have any quality data from downloaded forms that you never transmitted to Cécile and send it to [Valérie Lanneau](#).

For any further information on genetic testing quality data, please contact [Valérie Lanneau](#).

*Detailed response to a frequently asked question*

**Is the problem of accessing the Orphanet website resolved?**

As you all noticed, the Orphanet website was temporarily inaccessible in mid-October due to the steadily increasing volume of user traffic that overloaded the system. The Informatic team quickly mobilised to solve the problem. They re-programmed the request model of the database to improve the server response. Since, we recovered the previous rate of the website, even better!

*New documents published on Orphanplatform website*

**WONCA presentation**

The global presentation of Orphanet and its services given at the WONCA meeting in Paris can be downloaded from the Orphanet documents/powerpoints file on the [Orphanplatform website](#).



## Monthly statistics

Country Name	Total Data of services		New Data
	October 2007	September 2007	
Austria	616	616	0
Belgium	761	765	-4
Bulgaria	120	116	4
Cyprus	96	96	0
Czech Republik	169	169	0
Denmark	214	213	1
Estonia	74	74	0
Finland	260	260	0
France	8224	8223	1
Germany	4663	4551	112
Greece	266	266	0
Hungary	101	101	0
Ireland	163	162	1
Italy	4250	4099	151
Lebanon	21	8	13
Lithuania	8	8	0
Morocco	25	25	0
Netherland	839	839	0
Norway	136	136	0
Poland	12	12	0
Portugal	645	645	0
Romania	75	75	0
Slovakia	13	13	0
Slovenia	14	14	0
Spain	2082	2079	3
Sweden	19	18	1
Switzerland	620	614	6
Turkey	101	101	0
United Kingdom	2071	2019	52

**OrphaNetWork**, the Orphanet in-house newsletter

Editor-in-chief : Annie Olry (aolry@orpha.net)

Editorial board : Ségolène Aymé, Matthieu Levi-Strauss, Valérie Fonteny, Ioana caron, Ana Rath, Louise Taylor