Orphanet Europe Joint Action

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TRAINING REPORT

Paris, 5 & 6 June 2013

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19h Dinner: Restaurant "Au Moulin Vert", 33 rue du Moulin Vert, 75014 PARIS

Thursday 6 June 2013

Room Primevère (ground floor)

9h-9h30 OrphaNetWork

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15h45-16h30 Coffee & feedback session/ general discussion (partners'survey, past and future distant trainings….)

Participants

Sandro Coelzer (AT)

Paz Urbina (BE)

Annelies Mallezie (BE)

Nancy Anoja (CA-QC)

Susan Rogers (CA-QC)

Béatrice Geissbuhler (CH)

Ana Vera Ruiz de Castaneda (CH)

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Leena Toivanen (FI)

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Mai Tanabe (JP)

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Imane Cherkaoui-Jaouad (MA)

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Patricia Arinto (PT)

Rula Zain (SE)

Mina Mansoor (SI)

Ales Maver (SI)

Gabriela Nagyova (SK)

Idoia Gomez-Paramio (UK)

Observers

Gail Ouellette (CA-QC)

Loredana D'Amato-Sizonenko (CH)

Petra van Overveld (NL)

Speakers

Ana Rath

Charlotte Gueydan

Virginie Hivert

Sylvie Maiella

Claude Barrère

Marie-Pierre Becas-Garro

Bénédicte Belloir

Sandra Peixoto

SESSION I

JUNE 5TH, 2013

The power point presentation of this session is available here.

Orphanet

During this session a brief presentation of Orphanet was given by Virginie Hivert.
She stressed that Orphanet is a registered brand name owned by INSERM. She also reminded everyone that it is an international website that is freely accessible and that has national entry points from all countries in which a team is appointed. The site is dedicated to a broad range of end-users from patients to healthcare professionals, regulators, and pharmaceutical companies. It is also a communication tool used to share and spread expertise via the Orphanews newsletter. Behind the website there is a very complex relational database which also represents a pre-competitive tool highly-valuable to R&D in the field of rare diseases (data available for download at www.orphadata.org). Moreover, new services and collaborations are developed regularly to resolve the issue of information dispersion and to address the specific needs of the different stakeholders. The ongoing international partnerships were described (refer to the 2012 Activity report for more details here). The number of partnerships developed grows every year because of the international influence that Orphanet has, notably: Orphanet is highlighted as the reference resource for rare diseases (RD) in the documents of the European commission on RD, it is involved in the WHO's ICD 11 revision as well as SNOMED CT nomenclature revision, with IRDIRC (see page 13), we also collaborate with European Medecine Agency in establishing consistent RD prevalence figures, and with ECRIN (Rare diseases hub for European clinical research infrastructures network) and with EBI and it is part of Elixir's French node.

The scientific content of Orphanet was also presented. Literature is surveyed daily by a dedicated professional, which facilitates the collection of information on newly or further described RD, RD epidemiology & genes involved in RD. It also allows the revision and updating of the in house produced RD inventory and classifications. There is also a massive in house encyclopaedia production (of summary abstracts on RD but also emergency guidelines, general public texts) which is very appreciated by the end users (as shown by the users' survey). RD are indexed with ICD-10, OMIM, and cross-referenced with MeSH descriptors, UMLS, MedDRa and SNOMED CT. Genes are cross-referenced with HGNC, UniprotKB, Genatlas, OMIM, ensemble, Reactome and IUPHAR. One of the added-values that Orphanet features is that all the information produced in-house is expert validated.

This topic was followed by a short description of the Orphanet Directory of resources (expert centres, medical laboratories, patient organisations, research projects, patient registries, mutation registries, biobanks, clinical trials and networks), stressing that the establishment of the directory of resources can only be achieved thanks to the work of the national teams in the different countries of the consortium. A brief overview of the organisational chart was then given.

The Orphanet coordinating team roles and description

A reminder of how the international coordinating team works was given. The team is supervised by the chief medical officer and Orphanet deputy director Ana Rath. The quality manager, Charlotte Gueydan, is in charge of the technical supervision of the national teams. Virginie Hivert, the expert resources and pharmaceutical affairs team manager is in charge of the scientific support. Valérie Lanneau, the Client Project Manager, is in charge of IT development follow-up while Sylvie Maiella is the International coordinator in charge of following the ongoing contracts, the user rights, the translation workflow and the administration of the internal website, OrphaNetWork.
More generally, the coordination comprises: coordination of data collection at a global level, training and supervision of national teams, supervising overall quality of the database (between types of data, countries...) and also collection of information concerning transnational data.
It was also reminded that communication between teams is equally ensured by the collaborative tools that have been established or improved. OrphaNetWork consists of a website plus an internal newsletter (intended to communicate and share information; all the documents needed at a national level are uploaded onto the site). The Quality Assurance Review (QAR) is intended to assist and to guide the information scientists (IS) in data collection and a translation report is issued regularly, containing the necessary information to carry out translation activities.
Deeper insight on how the expert resources team works at the coordinating level was given: Virginie supervises the team. Data collection and registration is provided by Claude Barrère (expert centres & patient organisations), Bénédicte Belloir (diagnostic tests & labs quality), Marie-Pierre Becas-Garro (drugs & clinical trials), Sandra Peixoto (research projects, registries and biobanks) and Marie Villemeur, who assists them with their tasks.

Information scientist's key roles

The principal topic of the first session was then addressed and the key roles of the information scientists (ISs) were described: they are in charge of identifying the sources of information in the country and of collecting the information about expert centers, laboratories with diagnostic tests, patient organisations, clinical trials, patient registries, mutation registries, biobanks, research projects and platforms (according to the technical SOPs here). They also have to follow the process of validation of the collected data according to the workflow established by the Country Coordinator, ensure the publication of the validated data on Orphanet (using the registration tool Major, that since June 2013 is available for all Information scientists after the appropriate training) and update the data once a year. Other roles are communication with the coordinating team, regular reporting to the country coordinator, reading of the Orphanetwork internal newsletter and the Orphanet Quality assurance review (and the translation report when applicable) and carrying out the action points posted in them, contributing to the OrphaNetWork internal newsletter by sending short reports on activities performed by the national team and, for some, to manage the country website and find specific sources of information on rare diseases at the national level. The information scientist's tasks and national team roles are detailed in the SOPs available here.

Publication of quality ensured data & Collector tool

It was reminded that in order to publish data which are relevant and accurate, a process to ensure quality assured data has been established. The general process and the different actors involved were then described as it is detailed in the SOPs.
It was stressed that the priority list of data collection suggested in the SOPs should be adapted to the national situation in terms of resources & policy and that it is also dependent on the objectives of data collection (ex: if a national research agency transmits a list of research projects in the frame of the IRDiRC, it should be entered in the database in priority).
Then it was reminded that the country coordinator is responsible for the data quality management of expert resources in the country, and he/she should check the relevance of the national data prior to its release online. It is of paramount importance that the country coordinator validates the data collected by the information scientist prior to its online release, because currently the quality control is performed on data which are already available online. This implies that if the entered information is not relevant it will still be displayed until QC is carried out at the coordinating level. A pre-release validation can reduce the risk of publishing non relevant data.
With the availability of the new registration tool COLLECTOR (see below), all the steps of data validation, including QC, will be performed before online publication.
Finally it was reminded that for the post-release quality control, mini directories are issued through a tool called Exor which allows for the production of data directories that can be submitted to the scientific advisory board or the person in charge of PRQC. (Guidelines to assist the persons in charge of PRQC are available here).
It was also stressed that it is of paramount importance that sources of information and selection criteria be advertised on the national website as decided by the Orphanet management board.

A detailed description of the Collector tool was given by Ana Rath.
It was explained that it is an integrated tool for data management and quality assurance.
It was decided to develop it in order to face several challenges that go along with the development of the consortium such as: to make the national team autonomous, to simplify the dataset collected (some data are too difficult to obtain while some of the collected data are never used) and to adopt the UTF8 standard (for the website and the database). This tool has also been developed to follow the evolution in RD health & research policies such as the definition of shared quality standards used to collect expert resources, data normalisation to ease data exchange with other databases, quality improvement of the expert resources database (in order to achieve coherence and to make the database exploitable for analysis by a third party and for decision support).
Currently the data collection integrates different job expertise and the management of this integration is hard and time-consuming, often resulting in loss of information in the decision cycle. Therefore several needs have been identified, such as the integration of different jobs in a single data-management tool, the possibility of tracking decisions and sources, the implementation of quality rules BEFORE publication and to facilitate quality control AFTER publication.
In parallel, because of the increase in scientific information available on RD, the scientific projects also need to have a structured literature survey and a tracking of decisions and sources. In addition, to address the increasing demands to produce high-quality, structured,

up-to-date data, it was necessary to improve data structure in terms of concepts and relationships for disorders, genes, drugs and phenotypes. It was also necessary to produce new data on epidemiology, analytes and cross-referencing. The needs identified were exactly the same as those associated with the data collection of expert resources.
This is the reason « CollectOR » became an umbrella term, but the tool has two inter-connected sides with one more related to the expert resources/international coordination and the other to the scientific resources. The integration of data and workflows in the same tool requires some changes, such as evolution of the Orphanet database as a whole, moving to UTF8 , creation of new tools and the update of the existing tools and, of course, mobilisation of all Orphanet staff.

From a technical point of view, this ambitious project has been divided into several smaller projects to be more manageable. The different 6 sub-projects were presented and described, in particular:

- The sub project 1 includes development of Professor (internal name not to be used for external communication purposes), a tool allowing annual and continuous updates for professionals (it will be explained in full length during D2).
- The sub-project 2 includes creation of a new database (including revision of the dataset) and update of the international site and of the extraction tool. This will allow the presentation of new data (epidemiology, diseases, analytes, techniques and purpose of diagnostic tests…), the production of automatic definitions for certain disorders (moved-to, etiological sub-types, groups…), and the improvement of the presentation of expert resources and networks of expert resources (clinical trials, networks of expertise …). It will also allow for improved searching via the website (advanced search for tests & labs…)
  A first version of the definitive tool (Collector light) will be available. It is a web based interface replacing MAJOR. At this time, an updated version of Professor (V2) will be connected to the new website
- The sub project 5 includes the final version of Collector. This platform allows for: management of the professional's demands, publication of validated data (after national validation and QC) and it keep tracks of the decisions made and the data life cycle. It will also manage exchanges between teams and it will present the role and responsibilities of each position.

An overview of the general process was given:
The professional must log in to update or register new data (please refer to the Professor tool description in the second section of this report). Selection of the country in which he/she is will define the attribution to a Country workspace. The entered data then undergoes the above mentioned steps of validation. It has to be noted that according to the kind of modification (major or minor) and according to the sensitivity of the data, different actors may be concerned for validation (IS, country coordinator and QC coordinating team).
For example, IS can modify and validate Persons (Prs), Hosting institutions (Adr) and Departments/laboratories (Loc), whilehile officially designated status, person functions, network coordinating role for an expert resource and funding data may need further validation.

Questions (Q) & answer (A)

Q: How long will it take between the collection of new data and its online publication when the new tool is in use?
A: Currently QC is slowed down because it is looking at massive datasets and because of the difficulty in integrating the different jobs participating in the workflow with the different teams involved. Moreover, currently the QC team is also checking for relevance of the data when this should be the national coordinator's job. With the new tool the procedure will change: all the demands will be treated one by one and weekly meetings will be held where entries are looked over. Probably the slower step will be the national validation carried out by the coordinator who is in charge of checking the relevance of the data.

Q: What is the point in training for the use of Major with the arrival of the new tool?
A: To manage the 2013 update and because Collector will be similar but more user-friendly; what has been learned for Major will be applicable to Collector as it is about entering data in the database.

A. Directory of expert resources: criteria for data collection, dataset, quality control and feedback.

It was reminded that the aim of data collection is to have a comprehensive directory of expert resources for rare diseases in a given country.
Criteria used for the data collection of each resource were explained (please refer to the technical Standard Operating Procedures (SOPs) https://network.orpha.net/network/cgi-bin/articles.php?lng=en&pg=68 for more information). It was stressed that these criteria will also be explained in the Terms & Conditions page which has to be signed by the professional upon registration when using the new tool.

General recommendations on Data registration

- All information should be checked by you and the country coordinator (relevance).
- Specify if it is an update of existing information or a new data submission.
- Remember to check if a person is already registered because many duplicates currently exist.
- Mapping with diseases and genes is crucial – take some time to check the classification in Arbor 2! It is of paramount importance that the disease link is correct, as activities displayed on the website are now sorted by specificity of the disease link you select, therefore mistakes in the link will result in the display of wrong information and this could result in consequences for patient referral.
- Validate the complete list of diseases you have chosen with the professional, whenever the initial submitted list was too vague or too specific. You can export an ARBOR classification and submit it to him/her.
- If a disease or a gene is missing, contact the coordinating team and explain why you asked for a creation, and add corresponding references in the literature. The demand will be treated at the monthly disease meeting and, if relevant, the disease/gene missing will be added in the database.

The current database structure was described in comparison with the next version of the database. In particular, the dataset has been reviewed to evolve towards the new database, each dataset will move from the current infrastructure to the new one.
It was reminded that some resources have funny looking names because they are a shortened version of the French name (Ass for "associations" which is patient organizations in French…).
Regarding the contemplated changes in the datasets:

- ✓ Address will be kept but renamed "Hosting institution" with Town, Region (if relevant), Country
- ✓ Location will be kept but renamed « Department/Laboratory », it is an additional piece of information always associated with a hosting institution and linked to an expert resource.
- ✓ 'Person' will be split in two tabs: Person & Legal entity (Person is a professional or a stakeholder in the field of rare diseases). It will be linked to an expert resource or an abstract and is defined by a function in the database.

Questions (Q) & answer (A)

Q. The editorial teams often create duplicates of expert addresses.
A. This problem has been addressed. A big clean up of the addresses associated with the experts involved in the editorial process has been performed and great attention is now given on not creating duplicate addresses.

Q. The problem with the registration of the same patient organisation in countries with multiple languages.
A. It will be possible for a patient organisation to have their name in several languages.

Q. Why are telephone number, email and precise address are not collected anymore for locations?
A. IS lose too much time collecting this info that is too heavy to update. Since our role is to give visibility of the expertise, a link to the website of the hospital is all that is needed (as on their site the information will always be updated).

Q. It is often a request of the professional to have the precise address of the centre listed as often the hospitals are located in very different buildings or not even in the same area.
A. It will be specified in the terms and conditions of activity registration and the team can always explain the reason why this choice has been made.

Q. The postal code and region field are not mandatory
A. It is possible for the region, however either the postal code or the suffix should be entered.

Q. Is it possible to combine titles for example Prof & MD, as in some countries this is very important?
A. No it is not possible.

Q. What happens in case of homonymy of persons when they register?
A. This case has been investigated, there are very few homonyms in the database, and the difference will be made thanks to the email address.

Q. What happens to patient organisations that are also charities?
A. In this case they could be registered as both a patient organisation AND a legal entity. It will be the only duplicate allowed.

Q. What is the utility of having funding bodies?
A. They have a utility for the users using OR to support health policy decisions. They can be attached to the different resources and at national level they allow for statistical questions on health policy matters to be answered (for example which disease is more funded).

Q. Is the Involvement of funding bodies possible for the PRQC?
A. Yes it is possible, for example for research projects.

For each resource the definition and selection criteria were given as stated in the technical SOPs available here.
In addition some specific points were further discussed:

Expert centres

- ✓ Particular attention was given on the selection of expert centres, this argument will be further addressed during the 2013 management board to reach a definitive consensus. Check the meeting report here.
- ✓ Guidelines on how to assess the questionnaire will be produced according to the discussion held at this year's meeting. Check the meeting report here.
- ✓ It was stressed that validation of the relevance of a selected centre by the country coordinator is of paramount importance.
- ✓ Selection criteria, in particular if an official designation exists in the country, has to be decided jointly with the health authorities and explicitly stated in the national country website.
- ✓ Some important improvements with the new database will be the possibility of flagging a centre as both Genetic counseling and Medical management. Also, if the center is a coordinator of a network, it is possible to link 0 to *n* expert centre(s) to i .

Diagnostic tests

- ✓ Diagnostic tests for chromosome number anomalies and ring chromosomes done by FISH (e.g. Trisomy 11) are not registered in Orphanet, as their detection does not require a specific expertise in molecular cytogenetics.

Research projects:

- ✓ When research projects are completed, they should be flagged as « completed ».
- ✓ The consequence is that they would not be visible on Orphanet anymore and they will be removed from the database: except for those funded by a member of IRDiRC from 2010 on.

Tips

- ✓ **To avoid duplication between Registry and Research**
  - Research projects may concern the construction of a registry as long as the registry is not yet established
  - Research project may concern an already-established registry as long as it implies an actual research project conducted with the participation of patients of the registry
  - Collection of data corresponding to the expressed goal of a registry should not be entered as an additional research project.
- ✓ **To avoid duplication between diagnostic tests performed in a clinical setting and research projects aimed at developing a diagnostic test**
- ✓ A diagnostic test with clinical setting has to be registered only in the diagnostic test section and should be removed from the research project section.
- ✓ **To avoid misregistration between Clinical trials and Research**
  - Clinical research projects have to be registered as research projects not as clinical trials.
  - Clinical trials are mostly interventional studies, with a phase and a registration number from a national or international drug agency.
  - Clinical research projects are mostly observational and are performed with ethical committee authorisation but are not registered by a national or international drug agency (but they are sometimes listed in clinicaltrials.gov).

Quality control of the data entered

Quality control of all entered data is carried out monthly by the coordinating team.
Charlotte Gueydan presented the type of modifications or mistakes occurring frequently and their evolution over time for each resource. (This information is provided monthly to the Information scientists in the QA review; analysis per country has also been carried out and is available upon request).
It must be kept in mind that currently errors are classified into 5 categories:

- ✓ **Relevance:** when an entry does not meet the Orphanet inclusion criteria (e.g. expert center for a non rare disease)
- ✓ **QC-failed**: when assessment of the entry could not be performed as the associated information was not sufficient (e.g. research project without a description of the research aim or medical management expert centre with no addendum questionnaire)
- ✓ **Mapping**: when there is a discrepancy between the actual mapping with the Orphanet nomenclature of diseases and a more relevant one (i.e. wrong diseases and gene links)
- ✓ **Other amendments**: when items other than the disease mapping should be corrected (e.g. flags, types of tests or research projects, labels…)
- ✓ **Classification**: when modifications in the Orphanet nomenclature of rare diseases, genes, or classification should be performed to meet the needs of the current state-of-the-art in rare diseases, as suggested by the corresponding expert resource.

It was stressed that most of the corrections suggested are related to mapping problems. This is particularly unfortunate as wrong mapping results in an erroneous sort by specificity,

therefore irrelevant information will be displayed on the website. This could also result in a further diagnosis delay and misreferrals.
QC often failed on expert centres because no sufficient info is available, notably, it is often that the addendum questionnaire is missing or it is not validated by the country coordinator.

International Rare Disease Research Consortium

It is an initiative between the European Commission & the American NIH. Members are funding bodies spending a minimum of 10 millions USD over 5 years in research contributing towards IRDiRC objectives (200 new therapies by 2020).

Orphanet is the reference portal for R&D information for IRDIRC.

Implications for Orphanet's workflow and database:

- ✓ The scientific secretariat of IRDiRC is located in Paris, in Orphanet's office. One of our roles is to analyse the RD research landscape by analysing the research projects funded by IRDiRC members.
- ✓ The IRDiRC team keeps record of everything to be able to perform analysis
- ✓ Every Orphanet partner team takes care of its Research data
- ✓ Sandra takes care of FR, US and EC-funded projects
- ✓ Quality Control is performed by the coordination team of Orphanet

SESSION II

JUNE 6TH, 2013

The power point presentations of this session are available here.

Orphanetwork website

It was reminded that ORPHANETWORK is a site dedicated to Orphanet's national teams. It houses a wide range of tools and informative documents, as well as all the technical standard operating procedures regarding the information scientists' tasks. It is accessible with a login and a password (one login/team).

https://network.orpha.net/network/cgi-bin/index.php?lng=en

In order to improve the website, partners were surveyed for suggestions on what could be done to make it more useful for them. Many suggestions were received (refer to the 2011 & 2012 Partners'survey here) and all the improvements that were possible using the current administration tool were implemented. It was stressed that there are some limitations in the implementations suggested because this site is administrated thanks to a free tool due to there being no dedicated funding for this activity. However, it was decided that it is important to implement a better search tool allowing searches within PDFs documents, therefore some resources will be dedicated for this.

MAJOR workshop

All the participants from countries where access to the current registration tool was not granted were trained to use it. This approach will make national teams autonomous regarding the management of updates.
A survey to collect feedback on the workshop and suggestions for the next distant training and for the annual training was launched.

Description of the Professor tool

Professor is the new tool that allows professionals to register or update expert resources.
Please note that this is an internal name that should not be used for external communication purposes.
The interface for professionals will be available by clicking on the tab «Register your activity" found on the international website.
The link to this interface can also be sent by email whenever an update campaign is launched or it can be bookmarked in the browser of professionals.
An important feature is that this interface can be available in all the languages, the professional will be able to choose from a drop down menu. All 7 Orphanet languages will be

made available progressively, plus all the other languages for which the teams are willing to carry out the translation.

According to the discussion held at the 2013 training and also at the 2013 meeting it was decided that:

Upon accessing the interface the professional will have to:

- ✓ verify he/she is not already registered in Orphanet
- ✓ then he/she will access the login screen
  - a button for if you have forgotten your username or your password will be available
  - a "contact us" button will also be available

If the professional is not registered

- ✓ He/she has to create an account. The system offers two basic request choices:
  - Declare a new expert resource
  - Declare being involved in an expert resource already referenced

If the professional is already registered

- ✓ Either he/she is linked to at least one expert resource, in which case the system offers him four object requests:
  - Update my personal record
  - Declare a new expert resource
  - Declare being involved in expert resource already referenced
  - Update my expert resources
- ✓ Either he/she is not linked to any expert resource, but it is linked to at least one abstract (so it has the function "expert reviewer in Orphanet "), in which case the system offers him three object requests:
  - Update my personal record
  - Declare a new expert resource
  - Declare involvement in an expert resource already referenced

In the forms, the professional will have to select, from a scroll down menu, the disease to which the resource he/she is registering should be linked to (the list corresponds of course to the Orphanet rare disease inventory). If the disease is not present in the list there is a comment box that allows for the addition of this information.

The ISs will have access to these forms through their back office/workspace for pre-release validation of the data.
The professional is the only one able to submit the demand by clicking the "submit" button.

The ISs should regularly check his/her workspace in order to follow up any new form submitted by the professionals and then proceed to the pre-release quality control of the data and registration in the database before online availability.

In Collector, when the form is submitted by the professional, the information scientist receives a notification in his/her workspace.

Please note that:

- ✓ If the professional is registering an expert centre it will also have to fill in questions that are presently collected through the addendum questionnaire.
- ✓ If the professional has indicated a disease not available in the scroll down menu the IS has to check for relevance of the disease and then when applicable, submit it for creation of a new disease in the inventory of RD.
- ✓ The forms in Professor have three possible states:
  - Saved : the form is not yet ready to be submitted, but it is stored in the professional's workspace
  - Submitted: the professional has validated the information entered by him/her
  - Treated: the demand has undergone pre-release validation by the national team

An important feature of this tool is the traceability of the exchanges.

Additional feedback from the IS:

- ✓ in the field "are you an officially designated expert centre" it should be specified "for RD" to avoid confusion as in some countries all the hospitals are officially designated but not necessarily for RD.
- ✓ add a comment box at the end of the registration form
- ✓ for translation by the national teams it was suggested to send screen shots to help the translator contextualize
- ✓ having automatic emails sent to the professional is a possibility. However it could be better to have template emails available in the workspace that can be customised by the information scientists (for example in the case where a professional has updated 20 tests at the same time, if it is an automatic generated email, the professional will receive 20 times the same email for each test modified, clearly in this case the IS can send just one email recapping the 20 tests). In both cases a validation by the IS will be required before sending the email to the professionals.

Annual update

It was reminded that we are obliged by contract to have annual updates. This year's update of data will be launched through PROFESSOR. All registered professionals in the database will receive an email containing a link to the new interface asking them to create an account and to check their data.

The ISs will access these forms through their backoffice space. The modifications shall be entered in the database trough the MAJOR tool by the IS. A button will be created in the backoffice "treated".

The update has to be launched before the migration of the datasets to the new version of the database.

Post release quality control

It was reminded that published data on expert resources should be quality controlled in each country in order to be sure that:

- ✓ all the relevant expert resources are there (**completeness** for a country at a given time)
- ✓ no superfluous resource is present, which means, all the expert resources collected comply with the inclusion criteria according to the Orphanet standards (**relevance**)
- ✓ information given is exact (to the best of the validator's knowledge) (**accuracy**)

Performing PRQC is also necessary to have good-quality mini-directories for communication purposes thus it is an unavoidable step before disseminating the expert resources information in each country. It should be organized at the National level.

Mini-directories intended for quality control of the database content were generated by country and by expert resources, before the evolutions in the data model take place. These mini-directories are available for you to download at the following address:

https://backoffice.orpha.net/fichor/cgi-bin/views/

Guidelines describing how to perform PRQC have been produced to help validators in their task, they are available here. They should be sent together with the mini-directories to all validators.