



**Joint Research Centre & EUCERD Joint Action Workshop**

**on**

**Coding of rare diseases  
in health information systems  
with Orphacodes**

**Workshop Report**

**Ispra, 1-2 October 2014**

## DAY ONE

### CURRENT SITUATION OF CODING OF RARE DISEASES

#### INTRODUCTION

Jaroslav Waligora, Policy Officer at DG Health and Consumers at the European Commission, introduced the purpose of codification in the field of rare diseases (RDs). Discussions and substantial activities began several years ago on coding RDs as, in the absence of a uniform and precise coding system, RDs are not visible in health systems and data cannot be used for research or public health purpose. Health authorities therefore cannot adequately plan RD management.

With the rise in available treatments for RDs, the need to establish appropriate RD codification is becoming all the more urgent. Today's priority is therefore to discuss how to improve current status of rare diseases codification and which actions should be implemented on European and national level.

#### Output of preliminary steps to improve coding of rare diseases – *Ségolène Aymé*

Dr Ségolène Aymé, founder of Orphanet and leader of the Topic Advisory Group for Rare Diseases (RD TAG) for the revision of the International Classification of Diseases (ICD) at the World Health Organization (WHO), presented the situation concerning the ICD-11 revision process and terminology cross-referencing for RDs. A EUCERD Joint Action workshop took place in March 2014 to propose the use of Orphacodes by Member State health information systems. EC Expert Group on Rare Diseases (EGRD) discussions were held in July 2014, during which Member States expressed their interest in adopting Orphacodes and their wish to be guided on implementation systems, resulting in the present workshop to decide on next steps to implement Orphacodes at national levels.

Based on the current situation, in which different countries are using different adaptations of ICD-9 or ICD-10 and have different organisation of their coding process, it is necessary to explore what can be shared and what can be achieved at this stage to complement the coding systems in place with Orphacodes, the only option to date to achieve a homogenous and precise coding of RD. A draft recommendation of the EC Expert Group on Rare Diseases will be proposed for adoption in November 2014.

#### Discussion

The question was raised of whether RD data is being collected at the Commission. While RDs are not within the scope of EUROSTAT data collection, the Commission will examine the possibility to include RDs in data collection since codification allows this to be done.

On the question of mapping ICD-11 with Orphacodes, it was responded that Orphacodes are aligned with ICD11 beta version. However ways to organise the future update of ICD11 to maintain this alignment are not organised despite a proposal from Orphanet and an in-principle agreement from WHO

## **Presentation of the draft recommendations from the EC Expert Group on Rare Diseases: “Recommendation on the codification of rare diseases” - Jaroslaw Waligora**

The draft document has been presented at the Expert Group in July. Further discussion and possible adoption is planned for next Expert group meeting to be held on 12-13 November.

So far, only a few RDs are visible in national health systems and yet RD coding is essential to improve patient treatment pathways and clinical research. The lack of data on RDs is a problem for healthcare systems and needs to be addressed and improved over the next few years. There is a need to analyse RDs already coded in national health systems. In the draft text expert group on rare diseases is advising MS interested in using the Orphacodes to create working parties to discuss implementation possibilities.

Since the meeting in July 2014, the Expert Group is a step closer to evaluating ways to proceed with implementation. The next meeting, will allow the group to push discussions further on ways to improve and adopt codification in health information systems. The ECEGRD recommends that Member States consider adopting Orphacodes and explore possible implementation processes. It is as well suggested that an analysis of the number of rare diseases currently coded by each ICD national extension system should be conducted to have a documented picture of the coding situation.

Orphacodes are available freely under the Creative Commons Attribution-NoDerivs Licence and fully aligned (one-way alignment) with ICD-10, ICD-11 beta version, SNOMED-CT, MeSH, MeDRA and OMIM. Orphanet provides them in different formats depending on IT systems (xml, OWL, obo) at [www.orphadata.org](http://www.orphadata.org).

## **Introduction to concepts about Coding and Classification and about Orphacodes - How to code rare diseases with international terminologies? – Dr Ana Rath**

Dr Ana Rath, Director of Orphanet, presented the need to develop a common language to increase resource inter-operability and share clinical data between health care centres, databases and registries within countries and healthcare systems between countries. Concrete and detailed ways to implement Orphacodes in information systems in each country need to be explored. While no system will be exhaustive immediately, implementing Orphacodes will help gain knowledge on RD epidemiology, care pathways and public health impact.

Very few RDs are represented in current systems. WHO’s ICD-11 revision is not expected until 2017. SNOMED-CT is a multi-hierarchical, semantic coding system for electronic health records (EHR) and available in different languages for licensed users only. However, IHSTDO is starting to introduce Orphacodes into SNOMED-CT, which will be good for countries using SNOMED-CT. ICD-10 is mono-hierarchical (whereby diseases are classified once) and used essentially for statistical purposes. Over 80% of Orpha entries have been attributed an ICD-10 code. OMIM, available in English only, is used mostly for research on genetic diseases.

Each country must clarify their main objectives and reasons to incorporate Orphacodes. Strategies to implement these codes will depend on the way their coding system is organised and on the objectives of coding of RD (epidemiology, health policy, etc.). Countries must differentiate between coding (the act of introducing a code) and utilisation (deriving conclusions from the code and classifications).

Examples of utilisation were proposed, depending on the purpose of coding RD Codification for statistical purposes requires a mono-hierarchical classification, such as ICD-10. Orphacodes, ICD-11 and SNOMED are multi-hierarchical and will require linearisation for exploitation. Some degree of granularity will be needed to capture all cases since different professions code with a different degree of precision. Orphacodes offer several levels of granularity and can capture information whilst respecting classification systems needed for other types of use. The efforts required include nomenclature translation into other languages, data integration and tooling, coding systems alignment and coding instructions and training.

Orphanet produces a nomenclature and classifications specific to RD, based on literature and expert advice. Since the beginning of this year, the Orphanet nomenclature became an ontology, the Orphanet Rare Disease Ontology (ORDO), in collaboration with the European Bioinformatics Institute (EBI). It is mapped to several other nomenclatures. This multi-hierarchical classification is built on clinical criteria. The Orphacode number does not indicate the position in the classification and is stable, i.e. it is never reused when the nomenclature is updated. Behind each number is always the same concept, even if the nomenclature changes or adapts to modifications requested by the evolution of knowledge.

When there is no specific ICD-10 code for a disease, an Orphacode will complement the ICD10 unspecific code used. Orphanet offers a procedure to attribute Orphacodes in a consistent manner. Linearisation, attributing a preferred classification for each RD according to rules, is provided in Orphadata. While regular updates are necessary, codes must be stable for at least one year and sustainable over time for coding purposes. Traceability between different versions is necessary and a transparent and easily processed updating procedure should therefore be established to ensure visibility and sustainability for users.

## Discussion

Questions concerning the adaptability to real-life situations, the stability and sustainability of a tool to implement Orphacodes raise the issue of the need for funding to equip institutions with the necessary tools for coding RDs. Each situation must be analysed to quantify the needs of coders and adapt the tool accordingly. Certain real-life situations will nevertheless remain problematic and will have to be addressed.

## Approaches to using Orphacodes in national health information systems: the experience of Germany – Dr Stefanie Weber

Dr Stefanie Weber, Head of the Medical Classifications Unit at the German Institute of Medical Documentation and Information (DIMDI <https://www.dimdi.de/static/en/index.html>), explained how DIMDI developed a modification of ICD-10, the ICD-10-GM. Modifications from the ICD-10-AM (Australian Modification) have been incorporated and due to special requirements of the German DRG-System and data security and protection guidelines new categories have been introduced or existing categories of ICD-10 have not been included into ICD-10-GM. DIMDI provides the classification file and updates it annually.

It is up to the software companies and users to develop tools to support the coding with this classification. ICD-10-GM is now used in all areas of the German health system, from ambulatory care to quality assurance, morbidity statistics for epidemiology and research purposes to reimbursement.

Germany's National Action League for People with Rare Diseases (NAMSE <http://www.namse.de/english.html>) proposed adopting a uniform coding scheme for all RD patients using Orphacodes in parallel with ICD-10 German Modification (GM), until ICD-11 becomes available.

DIMDI decided to code all rare diseases from Orphanet according to ICD-10-GM and to link the Orpha number according to the diagnosis name to the ICD-10-GM-Code and to publish it in the electronic file of the Alpha-ID. This will allow DIMDI to provide the Alpha-ID file with the relevant codes from both systems to all users of ICD-10-GM in Germany. It is intended that this will standardise coding of rare diseases in Germany by providing electronic files for easy implementation. DIMDI plans to assist special centres for rare diseases in selecting the correct codes from both systems and to provide easy access to rare diseases information to medical doctors within existing medical information systems through this file.

Orphadata contains some 13,027 terms, while the alphabet-database of the ICD-10 GM file contains around 80,000 entries, many of which however are not rare diseases. The comparison between the two coding systems permitted to flag RD needing to be added, but also divergences in ICD-10 assignment. DIMDI began giving feedback in July 2013 to Orphanet on problems encountered, and will as well give feedback to WHO on ICD-10 updates and ICD-11 development.

The project, funded by the German Ministry of Health, started in July 2013 and will last 3 years. Evaluation of the project and of its implementation within the German health system will be performed during the third year. Within the project's first year, future users of files were interviewed to identify needs and potential benefits. Missing (German) terms were added to the ICD-10-GM alphabet-database. A new dataset is expected to be published in the package during the second week of October 2014, with which users will be able test usability. DIMDI will collect feedback to assess whether datasets are really used and whether the investment is justified.

Over the next months, updated files need to be assessed to take into account all the changes introduced since the last comparison. Orphadata should provide a way to track changes to help maintain the German file. Linkage to Alpha-ID numbering is not necessary in other countries and remains specific to Germany. The linkage is based on the diagnosis text and the Orphacode which is sufficient to uniquely identify each entry. The alpha-ID was chosen for Germany as it was an already established file-format for use in IT-systems. Experiences from Germany can be shared with all countries wishing to implement OrphaCodes. This could be an action point of the forthcoming Joint Action.

## Discussion

The question of obligation to code with ICD-10 GM in Germany was raised. By law, physicians must code with ICD-10 GM for in- and out-patient care. They are not obliged to use Alpha-ID. These are used only by a handful of software tools, many of which are in fact implemented in general practices and hospitals. Many physicians use the diagnosis texts from the Alpha-ID file to identify ICD-10 codes. The use of Alpha-ID is essentially for e-health purposes and for electronic data exchange, although they are frequently used for other purposes as well. Physicians code through a text search which assigns semi-automatically the right ICD-10 GM code. The Alpha-ID code is collected somewhere in the IT system as it is attributed to the ICD-10 GM information. The physician may therefore never actually see the Alpha-ID codes collected into the system. Alpha-ID codes are used only for electronic exchange purposes.

Instructions for coding are included in the ICD and coding guidelines for in-patient coding. Clinicians will therefore follow WHO coding guidelines. Hopefully these rules will not be affected by new ICD-10 GM files. Coding should not be a burden on physicians and they will continue to search Orphacodes as usual, by entering text to identify the right Orphacode. The German national plan may not last long enough as to impose the use of Orphacodes to all health professionals but DIMDI expects that it will eventually become compulsory for expert centres to use coding files in their systems.

## Coding rare diseases in real practice, the French experience – *Dr Rémy Choquet*

Dr Rémy Choquet, National Rare Diseases Database (BNDMR) operational Director, presented the French coding systems which includes ICD10 for coding hospital-based electronic health records (EHR) and the Common Classification of Medical Procedures (CCAM). In 2005, in the frame of the first national plan for RD, a national registry system to record data from patients seen in reference centres was established, called CEMARA. The registry was used as a generic registry tool for RD but also for research purposes. At the time of the second national plan, this project was extended to become BaMaRa which had, in addition, the task to define a minimum RD data set that every centre of reference/competence should provide. Thus BaMaRa extends CEMARA to gather the minimum data set from the electronic health records in clinical practice. This was established to identify all RD patients referred to RD centres, to track RD clinical activity and to contribute to research activities. Today CEMARA includes 1,000 users and over 250,000 diagnoses, 54% of which are confirmed. CEMARA is still a standalone system and is not incorporated into the general health information system. Data must therefore be incorporated separately into the two systems, which is doubling

the workload for professionals. Currently 42% of RD centres use Orphacodes. The quality of the coding depends on centres and their sites, and coding and recording homogeneity is lacking.

It is now planned to have an integrative national strategy, to connect BaMaRa to EHRs to minimise data re-entry for health professionals. Rollout for these actions is due to begin at the start of 2015. The integrative approach will interconnect public health epidemiology (via BaMaRa), care (via EHR) and research (via RaDiCo <http://www.radico.fr/fr/>, rare disease cohorts).

A second action of the second national plan was to integrate all Orphacodes into all health information systems. The ministry of health passed an instruction in December 2012 requiring that all RD centres code RDs using Orphacodes in addition to ICD-10. However, the directive was not well prepared thus not well executed and only 26,000 hospitalisations were coded in 2013. Main major issues listed are: lack of instructions for coding, no tools to help coding, no annual version and distribution mechanism of a list of codes, granularity and missing diseases were highlighted as a major problem as well as double-coding (Orpha + ICD). These problems need to be addressed to continue further with this project.

The plan has shifted its project objectives. Whilst deploying the minimum data set to all RD centres (through EHR links), the incorporated RD coding will be made possible at large scale and eventually at deep granularity. Now the second action on coding will enable to link the patients seen and labelled by RD centres with the French national database for ambulatory care (SNIIRAM). The coding project now aims at elaborating the right resource, which might enable capturing RD coding diagnosis outside the RD centres at a second stage. Once the thesaurus evolves and the Orpha nomenclature starts to be used more widely, its quality will also evolve. A French nomenclature expert group for RD (including members of BaMaRa, Orphanet, ATIH, ICD/SNOMED users, hospital coders, engineers and DIMDI) will be soon established to assess the current Orphanet resource and propose an operational adaptation to render Orphacode nomenclature efficient in a broader clinical practice but for RD centres first. The work will assess the needs for advice of RD centres, organise feedback to and from Orphanet and stay in line with ICD-11. Further tools are needed to operate inside hospitals to assist clinicians. The LORD (<http://enlord.bndmr.fr/#homepage>) tool helps visualising the classifications to help selecting the appropriate code RD. Future challenges include making sure Orphanet remains in capacity to maintain and distribute its coding system to the International community.

### **Coding rare diseases in real practice, the Italian experience – Dr Paola Facchin**

Dr Paola Facchin, Head of the Veneto Region Coordinating Centre-Registry of Rare Diseases, presented the RD classification system in Italy where physicians have to record patients with a RD as they are entitled to special dispositions regarding the reimbursement of care. In the Italian registry for RDs, clinicians need only enter the name of the syndrome, selecting the appropriate one from a list, without having to enter a code, nor to select the corresponding ICD and Orphacode. Thanks to this procedure, it is easy to select all the cases with a given disease. From a public health point of view, it is possible to evaluate globally how many patients with the RD have experienced a particular event. The difficulty of distinguishing and listing disease subgroups should be overcome, over time, by querying and elaborating lists with all the Orphacodes which should be aggregated. The limit of this approach concerns the impact assessment in terms of health service intervention planning, since this process is related to phenotypic manifestations of diseases.

The Italian solution to some of the health service issues using this approach consists in elaborating a hybrid system, using ICD non-specific code and Orphanet branches. According to this method, when classifying a rare entity through ICD-10 non-specific code, the ICD code is integrated to different branches of the Orphacode classification, in which the syndrome is included. The code selects a branch of the hierarchy according to the clinical profile of the patient. In this way, the multi-dimensional aspect of this classification is preserved, and the true value of Orphacode classification is exploited. This system can be used in research, clinical practice and public health. In practice, this automatic tool, using an informative system that is

mandatory in parts of Italy, can help diagnosis of all RD patients in all expert centres. The system, run in the Veneto region, was extended to other regions, to assess and diagnose patients with rare diseases in a population of around 25 million inhabitants. Clinicians need only select the name of the diagnosis, without any code, i.e. Alport syndrome. The system automatically shows the possible general branch in which Alport syndrome is represented. The clinician then selects the branch that best represents the clinical representation of the patient. Automatically, the ICD code and Orphacode for Alport syndrome appear in the patient form that the clinician can visualise and use to fill hospital records. All the information collected and coded by ICD and Orphacodes are registered in a databank. This method makes it possible to register a single disease or disease subgroup, or a group of diseases, depending on the granularity of the Orphacode and inform on the health needs of the patient depending on the branch selected.

So far the system has been used to register and classify around 83,000 RD patients. Studies are being conducted to evaluate whether this method can be extended to general hospitals and general clinicians. The system has been tested in general hospitals, in which 2.75 million hospital records have been processed by this tool. Out of these, 96,000 RD diagnoses records have been processed, concerning 66,000 RD patients. The result of this work is that the use of this hybrid integrated system increased by 85% the identification of RD patients, compared with the single use of ICD. This testing field demonstrates that the use of this system is feasible to increase results in terms of patient identification and health needs.

Possible future ideas include sharing and testing the available tool in new areas and comparing it with other approaches. Secondly, the orphan classification structure could be improved, not through the general linearisation process, but through identification of redundant branches. Thirdly, some branches could be simplified. Finally, it is necessary to focus on support, testing and coding tools. An automatic tool should also be developed supporting data analysis

## Discussion

The question was raised concerning the reasons for showing the classification when coding with ICD and Orphacodes. One could obtain the same result if only the syndrome were selected and the ICD code shown. What is the use of showing the Orphacode as well? It was explained that this is a general problem for diagnosis processes. A good diagnosis depends on the quality of the clinical process. When the clinician defines a clinical diagnosis, he defines this through a classification using the code. He then selects the branch, because only with the branch selection he can identify the phenotype and establish which care pathway the patient needs. If the physician uses only the specific Orphacode, only the ICD logic uni-dimensional classification will appear which does not show the full phenotypic scope.

Another question was raised on how the Italian system uses Orphacodes attached to ICD-10. When Orphanet attributed ICD codes, it was decided not to attribute ICD-10 codes to groups because the diseases under a group are heterogeneously linked to ICD-10 and it is impossible to attribute all the codes to ICD-10, therefore groups are not linked to ICD-10. It was explained that it is essential to maintain the multi-dimensionality of Orphacodes and granularity inside ICD codes. Orphacodes should be connected to ICD because ICD is a general instrument and all patients are classified with ICD. Therefore RD patients are not in a completely separate system and diagnosis can benefit from ICD granularity not present inside Orphacodes.

The issue of clinical training was raised. Since the system was applied to expert centres for RD, physicians have been trained to use the coding instrument adequately to establish patient care pathways. A big effort is also being made to train all professionals involved in RD activities. Italy's disease branch selection system allows physicians to direct patients to the right treatment centre.

### **Coding rare diseases in real practice, the Belgian experience – Dr Ingrid Mertens**

Dr Ingrid Mertens, from the Federal Public Service for Health in Belgium, explained the two mandatory plans in Belgium: the new Belgian plan for Rare Diseases and the Belgian e-Health Roadmap, important for changes conducted in Belgium. The national plan states that Orphacodes will become mandatory for Belgian reference centres, as Orphanet provides the most comprehensive list which has currently been constructed, this list is regularly updated and it is the foundation for the future ICD11 chapters on rare diseases. The roadmap states that patients will be recorded only once for multiple uses, excluding any double coding. Presently, hospitals still code using ICD-9 but Belgium will move to ICD-10 in January 2015. The plan is to migrate also to SNOMED in the future.

Coders are beginning to code with ICD-10 for diagnosis and procedures, and will begin grouping in Diagnosis Related Groups (DRGs) for hospital administrative purposes. System testing will begin in 2015. In the future, Belgium intends to use ICD-10 and DRGs, via SNOMED. The system needs coders and is semi-automatic. In the future, the intention is to develop a fully automatic system.

The Belgian Scientific Institute of Public Health is conducting a survey of reference centres to assess the progress in coding. Out of eight of these centres, five use currently Orphacodes. Members of the Institute not using OrphaCodes state that Orphacodes are insufficient and they have to use additional codes (SNOMED, HPO, OMIM, ISCN). Although these concerns have to be addressed, it is necessary to maintain the decision to have a unique coding system, which will be SNOMED for coding health events in electronic health records and ICD10 + Orphacodes for final diagnosis.

### **Coding rare diseases in real practice, the Bulgaria experience – Prof. Rumen Stefanov**

Prof. Rumen Stefanov, Director of the Institute of Rare Diseases, presented the situation concerning coding RDs in Bulgaria. A new ordinance (16/30.07.2014) about the designation of centres of expertise, reference network and national register of rare diseases in Bulgaria has been recently adopted by the Ministry of health. According to this ordinance, a new commission on rare diseases is expected to be established at the Ministry of Health with legal functions to take responsibility for actions needed towards RD patients. This commission, officially responsible for RDs, intends to define the list of prioritised RDs, recommend the designation of centres of expertise, evaluate activities of the national RD registry and centres of expertise, advise on RD management and national healthcare services, and collaborate with governmental bodies.

Concerning codification, RDs included in the national list of RDs will be coded using ICD-10, officially accepted for medical practice in Bulgaria. If specific ICD-10 codes are not available, Orphacodes will be used to complement the classification. The national registry of RDs will use in parallel Orphacodes, which will be used by expert centres for epidemiological and research purposes.

Several major concerns, expressed by various experts in Bulgaria were presented, among which was the question of Orphanet's ownership, sustainability and intellectual property. Issues, regarding the warranty, technical support, costs for implementation and training, EUROSTAT synchronisation were also commented upon. A feasibility study about the use and effectiveness of Orphacodes for public health needs in national health information systems in EU MS was proposed.

### **Discussion**

One major concern, expressed by various government officials, remains Orphanet's sustainability and intellectual property. This last point is refuted as Orphanet belongs to a large and reliable government organisation, guaranteeing its future existence and long term funding. It is added that WHO's ICD-11 project, in comparison, has received very little funding. Regarding the IP rights on Orphacodes, although they are at the INSERM, their use is free without any restriction under the "creative commons" framework. This situation is similar to the situation of ICD as WHO has the IP on the ICD codes which does not prevent anyone to use the coding system.



Discussions have already been conducted with the European Commission concerning Orphanet and Orphacode sustainability, including possible support for Orphanet and its inclusion in an ERIC. Discussions were not conclusive and solutions still need to be discussed together with Member States. The Commission believes that Member States should also be engaged to work on a European project. The idea was expressed that Member States might possibly consider paying a member fee for long term use of Orphacodes.

### **Coding rare diseases in real practice, the Hungary experience – Prof. Janos Sandor**

Prof. Janos Sandor, Representative at the Commission Expert Group on Rare Diseases for Hungary, presented the Hungarian project to introduce Orphacodes into the nationwide discharge record system, maintained by the national health insurance fund. Because it is a nationwide system, there is a strong legal basis behind the project. The ministry of health is convinced of the project and is waiting for the Commission Expert Group on RDs to develop the project. A structured and organised project was initiated to achieve the expected goals. Using existing resources in Hungary, the project intends to define ICD codes, currently used to code diseases in the system. The national health insurance fund has already accepted that ICD codes be completed by Orphacodes for reimbursement processes.

To be convincing during this process, the Hungarian project must demonstrate that the indicators prepared using Orphacodes in databases are more useful than indicators without Orphacodes. A stepwise introduction should be conducted as it is not possible to introduce all Orphacodes into the whole system at once.

A nationwide mortality statistical analysis was launched to demonstrate that the public health survey of RDs has dramatically changed over the past decades. Existing discharge records attempt to trace patients and demonstrate that Hungary is very centralised in terms of RD patient care organisation. Distinction between RDs with existing ICD-10 codes is currently useful to study a number of diseases. Other diseases, however, are under-registered in the system, which could become a problem when integrating new diseases registered using Orphacodes.

Concerning very RD patients, the question of cost effectiveness was raised and whether it is economically viable to use Orphacodes. Clear rules and training must be established to address multi-systemic and complex diseases. Furthermore, the Hungarian project considers that the legal background of the Orphanet coding system needs to be defined.

### **Discussion**

The question was raised about the pertinence of testing Orphacodes in mortality data, which is the area in which it is not expected to be useful. The Hungarian project wanted to demonstrate that the public health importance of RDs is significant and is growing. If different areas of RDs are separated, it is clear that there are fields in which the life expectancy of RD patients has improved significantly over the past couple of decades, whereas life expectancy has not improved in other disease areas. This mortality study was conducted only to make these observations.

## DAY TWO

### NEXT STEPS FOR THE IMPLEMENTATION OF ORPHACODES IN NATIONAL HEALTH INFORMATION SYSTEMS AND FOR EXPLOITING CURRENTLY AVAILABLE DATA

Chaired by Stefanie Weber, Paola Facchin and Patrice Dosquet, from the French Ministry of Health, the second day of the workshop was dedicated to discussing the next steps needed to proceed and align resources to make sure Orphacodes are implemented in health information systems. Since numerous Member States are interested in introducing Orphacodes, it is essential to improve Orphacode knowledge and training for RD reference centres and the broader public. Ways to ease the implementation of Orphacodes via the joint action in preparation were discussed.

#### Ways to improve knowledge of and access to Orphanet classifications – *Ana Rath*

**Ana Rath** introduced the topic of ways to improve the knowledge and access to Orphacodes. Ways are different when addressing the needs of health authorities, of coding agencies, or of ultimate coders. The question was raised on how to bring authorities and decision makers to recognise the benefits of using Orphacodes in their information systems. This question will be answered by the recommendations to be adopted in November by the EC Expert Group on RDs which should be then made known to appropriate authorities. The second question is more technical and concerns ways in which the codes should be implemented and analysed. An exchange of the experience of countries implementing Orphacodes and a round table of MS authorities/coding agencies involved in the process of implementing Orphacodes was proposed.

**Patrice Dosquet** presented the French point of view. France was an early adopter of a national plan, but still needs data for research and administrative purposes. However, it is not easy to collect all the necessary data for multiple reasons. An instruction from the ministry of health, which was not perfectly written or clear and not prepared with coders, was launched very rapidly at the end of 2012. Physicians at reference centres were ready to code, but about 50% were not equipped to implement the additional coding system. In French hospitals, coders are not necessarily the physicians who follow the patients. Rather, they belong to a special and powerful administrative entity who code in the context of reimbursements and hospital financing only. These coders consider only the financial aspects and not the epidemiological purposes nor care follow-up purposes of coding. The Ministry of Health was willing to launch this national plan action early, but it was not adequately prepared. It is therefore important to prepare the administration, physicians, coders and hospital administrators, and clearly write up administrative instructions. Then tools and training are necessary to help coders from all backgrounds use the codes.

**Paola Facchin** explained that in the Italian experience, the instrument to implement codes depended on a particular condition in the Italian law, whereby RD patients must go through RD reference centres to obtain a diagnosis and have their health cost supported by the system. This law was used to implement a dedicated health care system, not only the coding with Orphacodes, but also the entire modality to create regional centres of reference. The national law demands that an official diagnosis be made in one of these reference centres, networks of which must be set up in regions. Based on this obligation, Italy could initiate a national

information system. This is an opportunity that other countries could put in their RD national plan, as this approach permits to capture RD patients, but also helps identify good RD centres. Once patients leave the reference centres, they keep this unique non-nominative identification number, which gives them access to appropriate healthcare outside the reference centre.

Two instruments are used: the first during the diagnostic process at the reference centres and the second to flag the patient and follow how the patient evolves through the care pathway. It is therefore possible to gain a more complete picture of a patient needs and care consumption. This could be strategic for other countries to identify RD centres according to the type of care they can offer to particular RD patients. The centre's mission is to first diagnose and then define patient care and treatment plans. Treatment and care can be conducted in other centres, far from the initial reference centre that performed the diagnosis. Other professionals involved in patient care will be able to see online what needs to be done and the centre will monitor the patient's treatment pathway.

**Ana Rath** introduced the second part of the discussion on how to improve codification. If Member States have drawn up a national plan, coding is embedded in the plan, requiring no further need to convince of the benefit of identifying RD patients. The example of Italy – where all RD patients must be registered through centres of expertise – does not apply to other countries. Even if the national plan defines the need to code all RD patients, what measures should be introduced to help users put this into practice?

**Stefanie Weber** suggests that because the situation is different in all countries – Germany for instance does not have a national identifier for patients, it is therefore not possible to follow a patient throughout the healthcare pathway – experts need to explore possibilities that could be adapted to each Member States. It would be useful to show the code users the benefits of using the classification, starting with the centres of expertise and progressively extending this to practitioners and hospitals. Demonstrating the utility of codes to obtain information on treatment and care would help practitioners and hospitals manage very rare diseases, many of which may be unknown to them. A minimum set of standardised coding, adopted by all the countries, would improve the flow of information and benefit patients, allowing them to move around more freely and obtain treatment in other centres and other countries if necessary. Member States need to agree on a feasible system for all countries.

The question was raised about conducting a feasibility study to establish the use and validity of such an approach and the possibility to apply it to all Member States. It might indeed be reasonable to test this system in a few countries first to validate its utility before applying it to all Member States. It is suggested however that experiences in certain countries may not be transposable to other countries due to very different healthcare systems. The experiences are good in theory, but they are not scientific proof of the feasibility of the system in another context. In response to these doubts, experts should remember that it has been demonstrated that RD are currently poorly coded with existing systems and represent a burden for healthcare systems. It is therefore essential to monitor RD evolution and obtain data to make decisions on how to optimally organise the healthcare system. Demonstrating the necessity to code RDs adequately is well established and supported by a recommendation from the Expert Group on Rare Diseases to be adopted in November. Concerning ways to improve the system, there are few options because of doubts around ICD-11 and its date of release and adoption. In the meantime, the unique solution is to adopt Orphacodes that are aligned with the future ICD-11. Users of Orphacodes will therefore be prepared and gain time when switching from ICD-10 to ICD-11 once it becomes available.

Concerning cost effectiveness of such a system, further data is needed to convince users of the benefits of implementation. Because ICD coding does not offer information and data on the patient, no current alternative exists besides Orphacodes to gain more accurate RD patients' figures.

From the patient point of view, the most important task is to make ministries understand and find a way to make this solution economically feasible. Today, IT systems are very effective and not overly burdensome.

Implementing tools will be expensive, but done correctly, coding will be easy and adding Orphacodes to existing systems can be done easily.

The main question during this discussion was how implementation should be conducted in different countries. Each Member States has to decide if it wants to add Orphacodes to ICD codes when unspecific, if they want and can develop an electronic support to help the coders to use the Orphacodes (example of the visualisation tool in France) if they want to make references to hierarchy branches to enrich the possibility to exploit the data, which level of granularity they want, etc. Further questions were raised around the workshop's main objectives. What is the best practical system? Where to start? Should they start with expert centres, where experts are more likely to code correctly? How can a minimum set of codes be generated? The majority of Member States wish to implement, but are uncertain of the best practical ways to do so. It was suggested that Member States should clearly define what they want to achieve. All interested Member States should have the opportunity to share their experience with other countries so as to demonstrate the benefits and challenges of implementation. Member States may need to agree on a minimum set of Orphacodes to implement at a European level that all countries could adopt. In some countries, such as Bulgaria, authorities want to know how much this system will affect healthcare costs and what results can be expected in five to ten years. The recommendation was made that, due to the significance of this project, it is recommended to start coding with Orphacodes in centres of expertise to demonstrate the added value of the system.

Another recommendation was made to collect cost-effectiveness data in countries implementing Orphacodes. It was answered that no cost effectiveness studies were conducted when countries migrated from ICD-9 to ICD-10. While it is difficult to study cost effectiveness at this stage due to the lack of data, costs alone should be assessed before effectiveness can be analysed. Such a study of the costs of implementation could be proposed, although difficult to manage and probably not transposable to other MS.

### **Ways to implement Orphacodes in Member State countries (IP issues, update process, file availability, etc.) – Stefanie Weber**

Stefanie Weber presented the German model in the context of the e-Health as an example. The epSOS project (<http://www.epsos.eu/>), which ended in June this year, aimed to design, build and evaluate a service infrastructure to demonstrate cross-border interoperability between electronic health record systems in Europe for emergency patients. The epSOS team defined a master file used to describe the data to be transmitted with emergency patients in order for physicians from one country to access patient data in a centralised database. The epSOS file is now an open source, freely available tool for all users. The master file cannot, however, be used in Germany due to licensing issues. From this experience, if a project is to work across Europe, it should be made as simple as possible. The responsibility for file contents should be given to each country to make sure they cover their local needs, since international standards may not be applicable in all countries.

In order to constitute a master file, Member States need to validate what is available in their country, concerning the ICD or other coding in use, and the existence of potential licensing problems. A prototype could be created, such as Orphanet's link to ICD-10, as a starting point for testing. The granularity of the file should be discussed to address the number of diseases to include. Should the master file contain all the diseases in Orphanet or should it start with the most common ones? Should all diseases eventually go into the files and should it become mandatory for all Member States to use the file or would this represent too much administrative burden?

The long-term maintenance of this tool needs to be discussed and must be guaranteed. How will the file be maintained once it is put into use? If national versions of classifications are added to the file, attention must be given to national languages, translations and choice of terms. If countries use coding systems other than

ICD or Orphacodes, these could also be integrated into the master file. Because the file should be freely available, licensing issues must be solved in advance. A group of experts from different countries should collaborate to design the file, code entries from the various countries, avoid double coding and maintain the file. The file can be used by countries who want to code ICD-10 and Orphacodes separately to check plausibility and accuracy of matches.

Overall, it seems important to establish a long-term expert board to ensure long term sustainability and maintenance of the file. As well it should be possible for all user countries of the file to propose changes to the file, be it on structure or content. These proposals will have to be discussed by the expert board which might result in changes to the file or even in recommendations to change some Orphanet entries, if necessary. It is essential therefore to define a clear one-year update cycle. This approach has begun in Germany, but it should be discussed with other experts to assess whether this could be developed at European level.

## Discussion

In terms of emergency and unplanned care, experts believed epSOS could be a solution. Besides the problem of sustainability, such a tool deals with interoperability, which is needed at the European level and should be addressed early on. Experts from Member States can share their experiences as they develop the project, providing a more practical and realistic approach to constructing the file. In terms of file content, it would be preferable to map the English text and the Orphacode. National codes should, however, be attached to translations to ensure that all diseases and local language versions are included. It was recommended that the file be maintained on an annual basis, derived from the Orphanet alignments, but separate from the Orphanet central facility because of the different frequencies of versioning.

A second recommendation was to bring together experts from different countries to agree on a first list of diseases and draw up a working plan, starting with prevalence for instance, followed by progressive information accumulation. The master file is one part of the plan since it does not directly address the implementation issue of coding in different countries.

Experts need to establish objectives at the country and European level, and share consistency and past or current experiences between Countries. Member States must find a common denominator that is easy to implement and mutualise between countries, addressing each country's specificities. Two objectives should be clearly stated to capture the data at country level and synchronise implementation in all countries.

A recommendation was made that all European centres must use the master file in order to be part of a reference centre network. Experiences with ICD-10 coding illustrate problems encountered concerning the quality of collected data. It is therefore essential to provide an up-to-date master file freely, based on concurrent sets of data, to avoid different countries using and generating different data. Varying levels of information would prevent data pooling and analysis of some very rare diseases.

## Road map at Member State level and at EU level including funding options – Ana Rath

Ana Rath led the final discussion concerning elaborating a roadmap at Member State and at EU level. The expert group must begin exploring country needs in order to establish a concrete roadmap. The group must first conduct a detailed collection of objectives, existing solutions, how solutions are translatable or not to other country situations and what can be mutualised into a master file. Technical solutions need to be elaborated to reproduce implementation in other countries. Experts should all agree on this first proposal.

## Discussion

Past experience of European action shows that it is preferable to observe, report and share data already available and mutualised, and allow Member States to decide on how they wish to implement codes. Many countries have already initiated projects and do not require detailed recommendations on implementation approaches.

A proposal was made to begin working with countries that have already initiated implementation. A working group, as opposed to a study group, should be constituted to draw up a pragmatic and adaptable tool. The Joint Action should propose concrete actions and a landscape of what is required financially and at country level within the next year. A European level project could be discussed later and become a separate additional project. Concrete implementation, including all IT and training problems to be solved, would require specific funding.

The need for a concrete strategy on how to spread implementation to the whole health system was expressed.

A budget would be needed to create a space for exchange among the working group. Another general objective of the group is to elaborate guidelines to help Member States implement tools. A suggestion was made to standardise reporting experiences, activities, concerns and implementing preferences in different countries. It was suggested that the European recommendation enter the implementation phase soon, before countries start implementing tools separately. Early adoption would avoid multi-phase development in different countries and data replication. Each country would integrate sections of the master file into national systems according to what concerns them only.

Countries could benefit from other experiences, such as lessons from France, Germany and Italy. It was suggested that the task force could be hosted in the scope of the next Joint Action. The cost of producing a master file was questioned. Countries interested in the project could also be invited to invest in the task force. The task force should therefore address financial modelling to maintain this project long term.

The example of Norway was presented briefly at this stage. Norway's ten expert centres, attached to one resource service for RDs, were instructed to start reporting their activities using Orphacodes as of this year. Two centres have the same electronic records systems, into which the Orphanet xml file was downloaded. Diagnosed patients are given the Orphacode, the English name, the adjoining available synonyms and the corresponding ICD-10 code. The work is due to start mid-October. The centres will be able to establish whether the ICD codes are very different to those already used in Norway.

While it is a good idea to implement the model progressively and in a country-by-country approach, the expert group must bear in mind that it is not possible to implement the German model in France for instance, due to technical issues. It was suggested that a sound approach would be to share the implementation processes, based on the tools available in each country. A roadmap should be proposed to outline clearly what needs to be achieved over the next three years. Implementing should however be conducted at the local level.

Problems to address at the European level will include making sure all countries are using the same version of the file in order to enable data extraction and analysis. The example of Norway raises the question of what version of ICD centres (including Orphanet) are using to code. When cross-referencing data and counting patients across different countries for instance, coders must make sure the leaves under a disease class have not been modified from one year to another. Experts were reminded that a master file is only part of classification of RDs. A master file is not an instrument for technical interoperability or a solution to all implementation issues, but rather a basis to compare and validate data.

The Joint Action cannot offer space to discuss projects at both European and national levels. However, this task force should focus on actions concerning technical issues that can be achieved at the European level rather than at the country level. Because different countries have different systems, it would be impossible to propose one tool to fit each country's needs at this stage. The Joint Action must agree on a mutual tool,

such as a master file, that all members can achieve together on a European level. The Joint Action is the only instrument available at the moment to conduct a project and the promotion of a master file should, during this first phase, be part of a work package in the Joint Action.

It is essential that countries agree to follow the implementation and maintenance of a master file even after the Joint Action ends. Focusing on the sustainability of the plan at Member State levels is one of the outcomes of this work package. The costs for IT, training, maintenance and sustainability of a master file, and burden of annual updates should not be under-estimated to avoid losing user motivation. Since launching its national plan in July, Ireland for example is keen to implement coding, but requires funding and support to conduct feasibility studies on implementation and long-term sustainability.

The next Joint Action could be the opportunity to promote the development of a master file as a common tool, taking into account two remarks. Firstly, the Joint Action should work on a master file, based on experiences of countries already using orphacodes with a maybe limited number of diseases shared by all the European countries. Secondly, Member States should ensure sustainability at country level before concentrating on sustainability at the European level. Orphacode implementation will require guidelines for coding in each country. The master file can be shared as a common tool, but the option on how to implement the tool will remain national for some time still.

### **Concluding remarks**

A draft proposal of a work package on codification must be drawn up soon and distributed to participating countries. The work package must be clear, concrete and built on experiences from all the countries, including doubts from countries hesitating to participate. As a result of this workshop, it was recommended that the contents of a work package be written up, integrating ideas from all the Joint Action members. Official nominations from Member States of the bodies to be part of the Joint Action are required by 20<sup>th</sup> October. The plan is also to share this draft with all Member State representatives in the expert group and propose additional ideas for the Joint Action concerning other work packages. Further discussions will be held at the next expert group meeting in November. The expert group will receive this draft recommendation, which will hopefully be adopted.