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ORAL PRESENTATIONS

NATIONAL PLANS AND STRATEGIES FOR RARE DISEASES

O1
The German plan for rare diseases: a development in progress
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In the year 2008, the German Federal Ministry of Health initiated a comprehensive study on the topic, “Activities for the improvement of health care for people with Rare Diseases”. The study was motivated by recommendations on Rare Diseases by the European Council of Health Ministers. One important aim of the Council is to develop strategies and plans to improve the situation for patients with Rare Diseases at a national level.

In summer 2009, we published the aforementioned study about the situation of people with Rare Diseases in Germany, in which we made recommendations to improve their health care and life situation. One major conclusion was the creation of an institution working on a national plan for Rare Diseases in Germany.

In consideration of this result, the German Federal Government announced the foundation of NAMSE (National coalition for people with Rare Diseases) in March 2010. The main purpose of NAMSE is to finalise a German plan for Rare Diseases until the year 2013. Other goals are to concentrate current initiatives and to encourage pilot projects in the field of Rare Diseases. In order to secure the implementation of different perspectives and point of views, NAMSE’s members are the most important stakeholders of the German health care system. This helps to ensure the feasibility of the national plan for Rare Diseases. The constituent meeting will take place in summer 2010. NAMSE is separated into a steering committee, which is the decision and controlling board, and working groups, which provide functional and continual work on particular questions. The activities of NAMSE are patient information/transfer of knowledge, diagnostics and therapy (including development of guidelines), health care and quality assurance, national and European networks of Reference and Expertise Centres, research as well as topics, suggested by the steering committee.

O2
State of the art of services in Europe: where are the problems?
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With regard to centres of expertise, the Orphanet database lists 378 centres of expertise, mainly concentrated in Belgium, France, Germany, Italy, Spain and the UK. They cover very different realities as the qualifying criteria to define a centre as an expert centre differ from one country to another, both in terms of the mission and in terms of resources. Some experimental European reference networks of centres of expertise have been established and funded for a three year period only, a time period too short to allow any assessment of the added-value of these networks. With regard to genetic tests, only testing for Cystic fibrosis is provided by every country and over 500 diseases are testable in only one country in Europe. The test offer differs greatly from one large country to another: Germany (1,141 genes), France (874 genes), Italy (825 genes), Spain (582 genes), United Kingdom (414 genes). This situation explains the large cross-border flow of specimens, highlighting the need to provide access to services in other countries when necessary, especially for very rare diseases. With regard to the provision of information to patients and professionals, the difficulty is to maintain updated information about several thousands of diseases and to provide this information in languages understandable by the end users. With regard to funding for research on rare diseases, the multinational common calls for proposals E-Rare now covers Austria, France, Germany, Greece, Israel, Italy, Spain, Turkey, The Netherlands and Portugal. Regarding patient registries, the main problem they face is their sustainability as they are long term projects and most funding sources only support short term projects. The way forward is to establish a public/private partnership in this area with the support of regulatory agencies. Among MS, major disparities in access to treatment are also observed.
**O3**

Recommendations for the development of national plans for rare diseases

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Rare diseases are a threat to the health of EU citizens, so far as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity. Despite their rarity, there are so many different types of rare diseases that millions of people are affected. The focus on rare diseases and the recognition of the fact that rare diseases have common issues in a public health perspective and require specifically targeted policies, is a relatively new achievement in most EU member states. There is at present great variability among countries about the type of services provided to rare disease patients and the accessibility to these services. Following the Council Recommendations, each Member State should (preferably by the end of 2013) establish and implement plans or strategies for rare diseases at the appropriate level. The aim is to ensure that all patients with a rare disease in Europe have equal access to high-quality care, including diagnostics, treatments and rehabilitation. The European Project for Rare Diseases National Plans Development (EUROPLAN), has the task of elaborating documents to facilitate the establishment and implementation of National Plans or Strategies. EUROPLAN has defined a National Plan or Strategy as a set of integrated and comprehensive health and social policy actions for rare diseases, to be developed and implemented at national level, and characterised by identified objectives to be achieved within a specified timeframe. The EUROPLAN guidance document (recommendations) provides a set of “tools and examples” on how activities for rare diseases can be organised at national (and European) level on different areas, e.g. the coding (and traceability) of rare diseases, research, centres of expertise and the empowerment of patient organisations for rare diseases.

(http://www.europlanproject.eu)

**CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS**

**O4**

The added value of centres of expertise for rare disease patients in Europe

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In many European Union (EU) Member States (MS), rare disease (RD) patients are subject to marginalisation in classic healthcare systems designed for non-rare diseases. As a result they do not experience equal access to timely, high quality health services they deserve. The restructuring of healthcare systems to better reflect the values of equity and solidarity amongst RD patients, professionals, and policy makers across Europe can be accomplished through the establishment of Centres of Expertise (CoE).

Although no official definition of CoE exists, some MS have established physical expert structures for the management and care of RD patients at the national level. Specialising in a single RD or a group of RDs, CoE are ideally care centres that bring together a group of multidisciplinary, specialised competencies and ensure timely diagnosis and appropriate follow-up care by aiming to improve the continuity and coordination of care through the implementation of healthcare pathways.

In addition to providing an added value to the quality of local healthcare services, CoE are also key determinants for research on rare diseases, and support the optimisation of healthcare spending in the current economic climate. Stakeholders across Europe in the rare disease community have worked together to establish several key policy elements synthesized in the Eurordis Policy Factsheet on CoE.

In order to best establish, manage, and sustain long-term functioning of CoE, monitoring systems must be established appropriate for each disease and national context. Building on the previous work of many stakeholders in this discussion, MS across Europe should work toward the establishment of CoE as possibly the most crucial elements in the context of national plans and strategies. Although much success has been observed in the establishment of CoE in several MS, several challenges for future national strategies on CoE remain and are identified here.

**O5**

Individual plans and coordinated services: an empowering process

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In Norway there are some 30,000 people with rare hereditary or congenital disorders. For many rare disorders, the needs they have can be a challenge for both the patient, the family and for the service providers. As a user of long-term, coordinated health and/or social services in Norway, you are entitled to an Individual Plan (IP). The right to an IP is not conditional on a particular diagnosis or age, and is mentioned in several Norwegian acts. One service provider (coordinator) has the overall responsibility for each person’s IP. To succeed as a coordinator it is essential to establish and maintain a relationship with the user based on trust and respect. An IP contains an outline of your objectives, your resources and the services you require. As a user you have the right to participate throughout the planning process. The users’ contribution to the plan is crucial for success. The individual plan should state when the different measures are to be carried out and who is responsible for this. An IP documents your actual situation and your need for measures and services, and it can also provide a basis for applying for services you do not have today. An IP may be a tool for you to reach your goals in life. It is documented that 87% of the professionals in local authorities and specialist healthcare think the working model with individual plans encourage cooperation between different parts of the services. In Norway there are 16 different state-financed resource centres for people with rare diseases. To date, such services have been established for over 300 diseases. These centres can be important contributors to an individual plan process, in addition to their advisory role to different parts of the help system.

**O6**

Building centres of expertise according to the Dutch model?

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Establishing the expertise for rare diseases started in the Netherlands by an inventory on existing expertise in 500 rare diseases at the Dutch Medical University Centres. The outcome of this survey was a vast list of persons with special knowledge of one or several of these rare diseases. The Dutch Steering Committee on Rare Diseases and Orphan Drugs concluded that differentiation within this list was needed by defining more specialised criteria for expertise. The criteria are based on European directives and are in line with a future perspective to improve quality of care. The highest distinguished level of expertise is an Expertise Centre where basic research, scientific output and professional training result in multidisciplinary high level care for people with complex rare diseases. Alongside this, Expertise Teams provide applied clinical research and high level care in local treatment centres. Currently, the Steering Committee is consulting professional groups, treatment centres and other stakeholders to gain support for this plan.

In 2011, the Steering Committee will advise the Ministry of Health about the organisation and the quality monitoring of the Centres of Expertise and Teams for various rare diseases. This advice will also contain a paragraph on a financing model concerning the organisation of care for rare diseases in the Netherlands.
French experience with rare diseases plans
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The first French national plan for rare diseases (2005-2008) laid the foundations for Rare Disease (RD) specific activities, mainly by creating centres of expertise. The second plan (2010-2014) aims at building the RD world of tomorrow on the foundations of the first plan taking into account its achievements and pitfalls.

RD patients are rare, as are RD experts. Building, maintaining and spreading expertise requires a constant equilibrium between up-hill and down-hill.

Up-hill is filling the gap between patient care and the tremendous fundamental progresses which can come from a close connection with fundamental research, and academic and/or industrial groups involved in orphan drugs or new therapeutic approaches. The main achievements of the first national plan have been a large support for research including therapeutic trials, for genetics labs and building animal models.

The main projects for the second plan are to create a foundation for scientific cooperation which deals with advising for scientific projects and grant submissions, of helping with access to highly specialised tools and promoting partnership between academic and industrial stakeholders.

Down-hill is the use of expertise for improvement in patient’s quality of life, from centres of expertise to the patient’s home. Spreading information and guidelines is time consuming, and involves several levels of actions. The first plan’s main achievement was the identification and support of 131 centres of expertise and their network of 500 «Competences» centres; the increased financial support to Orphanet; and the improved connection between patient organisations and experts.

The second Plan’s main projects are to establish national networks for dissemination of expertise, information, and therapeutic education as well as neighbouring assistance for home care and day to day problems. Moreover it has to be emphasized that expertise is bicephalous, involving both specific clinical management and biological assessments by highly specialised and constantly evolving techniques. The second plan will officially recognise such synergies.

Living with Progeria
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Children with Progeria can be an inspiration to those who meet them.

Mission: The Progeria Family Circle is a parents’ organisation and network that supports European Progeria children and their families in several ways. The objectives are fourfold:

Meetings: First, the foundation organises annual meetings for all European children and their families. These are important highlights, because of the rarity of the disease children never see other patients. Also for the parents, mutual contact proved very valuable. Meetings are a source of joy in which the children find much self-esteem. Also they have the opportunity to speak with specialist physicians, and to exchange actual information about Progeria.

Information: As a second objective, the Progeria Family Circle offers parents of European children advice and support where needed. The value of emotional support and assistance of parents with experience is difficult to estimate. We bring family doctors and physiotherapists into contact with specialists in the field of Progeria.

Support: Progeria families come from different countries with different social, political and religious backgrounds. It is sometimes a big problem for families to give their children enough care. We also look for solutions to individual households to help them as directly as possible. If necessary, we seek financial support, when needed for the welfare of the child.

Medical developments: The fourth objective focuses on the medical field. The aim is to support better and faster recognition of symptoms and problems in new, but also known patients. Scientific research and new therapies have made much progress in the past few years. An experimental treatment with Farnesyltransferase inhibitors (FTIs) started in Boston, and European children were offered a different therapy with a combination of statines and amino-biphosphonate in Marseille. The last combination of medication is also now used in Boston.

A “Family Project” to fight Usher, a rare disease leading to deaf-blindness
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It was 1994 when physicians told us that our two sons - deaf from birth - will lose their eye sight also to Usher, a rare genetic disease. With inspiring support from motivated scientists, private equity, National- and European Agencies we moved from fragmented basic research in identifying the gene, the molecular mechanisms of the protein and the specific mutation to shared objectives in disease specific research for a therapy. We founded a Family-Foundation to finance our activities, a medical device company to develop a retina implant, a chip for the eye and worked in EU- and US organisations to bring science ahead, e.g. the Foundation Fighting Blindness (FFB), Fondation Voir & Entendre, Pro
Retina Foundation. We have already identified a cell-based treatment by a drug which, down the road, has the potential to put on hold the progression of our son’s disease. Therefore we have focused our activity with a view to strengthening research with the European TREATRUSH project within FP-7 Health, participated in networks for fund-raising and pursued tests to see, if the drug (PTC 124) will stop the degeneration process in the retina with the assumption that what works in Duchenne disease will also work in Usher 1C, and raise awareness of companies to invest in therapy with PTC 124 for patients with nonsense mutations in retinal degenerative diseases including Usher 1C. Unlike the early days we see a clear road to a therapy and are planning clinical trials with interdisciplinary teams. There is also a critical mass of expertise and substantial management by a biotech company for the benefit of all with Usher 1C. Yes, we can.

O11
Very rare disorders - organisation of care
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Care for patients and their families with rare disorders is gradually recognised as being essential both on an individual basis and on a population scale. Attention is usually paid to those with disorders with prevalence between one in 2,000 to one in 100,000. Not many official bodies realise that the majority of patients with rare disorders have in fact disorders that are much rarer, with a prevalence of one in 1,000,000 to one in 100,000,000. This has consequences in providing the best care as the creation of support groups for such rare disorders is much more problematic, and centralisation of care cannot be established on a national level but only on a European level. This has wide-spread consequences, for instance regarding language, insurance, transportation, and research. The use of the internet to establish contacts, create Wikis around such rare entities, and to allow for e-mail consultations with medical experts elsewhere in Europe is emphasised and demonstrated using the entity Marshall-Smith syndrome (30 patients known in Europe) as an example. The establishment of large centres of excellence for patients with very rare disorders has been suggested, either one in each country or one for around populations of 30,000,000 Europeans, to provide essential care and form a basis for further research.

O12
The Epidermolysis bullosa house in Salzburg
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With a reported prevalence of 4.6 per million, Epidermolysis bullosa (EB) is a rare disease comprising a clinically as well as genetically heterogeneous group of inherited mechno-bullous disorders. Minor trauma leads to blisters and erosions on skin and mucous membranes. Based on mutations in genes encoding for structural proteins of basal keratinocytes or within muco-cutaneous basement membranes numerous extra-cutaneous manifestations may complicate this disease, causing multi-systemic involvement with significant morbidity and even mortality that necessitates multidisciplinary care.

The EB House Austria was founded in 2005 and forms part of the Department of Dermatology of the Paracelsus Medical University, Salzburg, which has mainly been financed by the patient organisation DEBRA Austria and a one-time subsidy granted by the federal government. As an interdisciplinary clinical unit for diagnosis, medical care, academic affairs and research related to EB, the EB House Austria fulfils all criteria for Centres of Expertise according to the “Final Report on European Centres of Expertise (ECZ) from RDTF expert group” in 2005. Besides treating patients from Austria, EB suffers from 15 other countries have been visiting the EB House up to now. In the light of the proposed Directive for European Networks of Centres of Expertise, pros and cons of cross border healthcare like liaisons with the referring physicians, post-treatment care, training opportunities for foreign clinicians or reimbursement are discussed. For example for some of the border EC countries, reimbursement is well organised. For others individual solutions have been found so far. All the specialists of the EB House Austria collaborate closely with other major EB groups, both in Europe and worldwide, and are willing to give all EB patients a home and hope for the future.

O13
Advancing diagnosis, care and treatment for people with neuromuscular diseases around the world: a network of excellence to catalyse research infrastructure globally
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The TREAT-NMD (Translational Research Europe: Assessment and Treatment of Neuromuscular Diseases) network was initiated in January 2007. With 21 partners encompassing clinical and research centres of excellence in the field together with industrial and advocacy partners, the NoE was launched in response to the need for infrastructure to support a growing translational agenda in the field of neuromuscular diseases (NMD), supported by a strong group of advocacy organisations. From the outset, TREAT-NMD has embraced the need to work on a truly international stage by developing partnerships with groups across the world. This approach has led to international collaborations on infrastructure projects such as the definition of animal models for preclinical studies in NMD, patient registries and biobanks, a care and trial site registry, harmonised standards of care, standardised outcome measures and assessments and ethical and regulatory interactions. The network has also initiated a new committee to advise on the multifaceted issues facing drug development in NMD- the TREAT-NMD advisory committee on therapeutics or TACT. From an initial focus on Duchenne muscular dystrophy and spinal muscular atrophy, the utility of the tools developed through the network is now recognised across a wide range of NMD and the “toolkits” generated are also relevant to other rare diseases with the need to address the pathway to therapy delivery.

EU investment in the network has provided a model which has added enormous value to the field. This is reflected in industrial investment in the tools of the network as well as the ability to generate additional grant income through the utilisation of the newly available infrastructure. Guaranteeing the sustainability of such successful infrastructure projects is an important consideration for the future.

O14
DYSCERNE: a European Network of Centres of Expertise for Dysmorphology
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There are over 2,500 identifiable dysmorphic conditions, which individually are rare but together form a significant proportion of referrals to a genetic service. The rarity of these diseases means that even in centres of Expertise established in many EU countries, experience can be limited, resulting in delay or uncertainty of diagnosis.

To improve diagnosis of dysmorphic syndromes across the EU, a formal European Network for Dysmorphology was created within the EU-funded DYSCERNE project (2007-2010) coordinated by the UK (University of Manchester). It links a total of 85 centres, including 32 centres of expertise with the remaining centres acting as case submission nodes for
a web-based electronic Dysmorphology Diagnostic System (DDS). Making a correct diagnosis is essential for patient management and for providing accurate information and counselling. The DDS allows rapid access for clinicians from across Europe to expert opinions increasing accuracy of diagnosis. It will also facilitate definition and classification of rare dysmorphic disorders and promote further research. Linked to DDS, educational tools in a modern, on-line format were created aimed to guide and educate clinicians throughout Europe on key aspects of clinical dysmorphology (http://www.dyscerne.org). One of the principal activities of the DYSCERNE network was developing best practice management guidelines which use a robust methodology. Management protocols for four selected conditions: Angelman, Noonan, Kabuki and Williams Syndromes will be available on the DYSCERNE website soon.

There is a need to continue activities initiated by DYSCERNE, in particular to sustain European networking, which helps to make a correct diagnosis and continue development of management guidelines for further rare diseases. DYSCERNE is funded by the European Commission Public Health Executive Agency (DG Sanco) Project 2006122 - A European Network of Centres of Expertise for Dysmorphology, and is supported by the Manchester Academic Health Sciences Centre (MAHSC) and the NIHR Biomedical Research Centre.


SCIENCE FROM THE BENCH TO THE BEDSIDE

O15 The European research area network - E-Rare
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Rare diseases represent an important public-health issue, affecting 26-30 million persons across Europe, and a major challenge for research. The fragmentation of resources and knowledge for the 6000-8000 rare diseases and the lack of efficient treatments for many of them necessitate a coordinated European approach to unravel the underlying molecular defects and pathophysiological mechanisms. The low number of affected patients requires transnational collaboration with multidisciplinary approaches to map prevalences, build patient registries, identify biomarkers, develop new diagnostics and finally perform clinical studies for the development of treatments. To this end, 8 main European research funding organisations have gathered into the FP6-funded EC ERA-Net on rare diseases (E-Rare) (2006-2010) and developed a number of joint activities regarding systematic exchange of information and best practises, definition of strategic priorities, and, most importantly, joint funding activities through the launch and completion of two fully fledged joint transnational calls for research projects on rare diseases (2007 and 2009). This exemplary joint funding activity has attested the need of, and the acknowledgment from, the research community for transnational funding of collaborative, multidisciplinary and ambitious projects on rare diseases. It has leveraged funding for rare disease research in countries that do not have specific programmes for rare diseases and thus enabled the participation of researchers in these countries to transnational projects. A new E-Rare project (E-Rare-2) (2010-2014) aims at deepening and extending the cooperation among the E-Rare-1 and four new partner countries by systematic exchange of information, yearly launched joint calls, thorough assessment of the funding mechanisms and results of the funded research projects and, finally, strategic activities aiming at a sustainable development and extension of the network. Special attention will be given to the outreach and knowledge exchange with new Member States, countries outside of the European Union and key stakeholders/initiatives important for rare diseases. E-Rare-2 activities will thus further contribute to reducing fragmentation of research and resources through the enhanced coordination and transnational funding of excellent research on rare diseases, thereby shaping the European Research Area for rare diseases.

O16 Determinants for research on rare diseases
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The term rare diseases (RD) includes a wide heterogeneous group of disorders that can involve any human organ or system. In the last decade RD have been incorporated into the agenda of health-care providers and public-health authorities; however, many problems still remain to be solved and even addressed properly. Some of these issues refer to patient care and management such as early diagnosis, quality of the information, access to treatment and rehabilitation, and appropriate multidisciplinary healthcare. Research is also necessary as it is the way to learn about mechanisms of diseases, improve diagnosis techniques and develop new therapeutic approaches. Research in psychological and social aspects of RD is also needed. Determinants for biomedical research in RD and orphan medicines affect basic science, clinical investigation, translational approaches, and transfer activities at the pharmaceutical and biotech companies, making the patient the focus of the triangle made up of the academy, hospitals and industry. These determinants can be summarised as follows: i) The human factor: human resources for research need to incorporate new research teams into the field of RD and especially young scientists and physicians; ii) the financial factor: funds from public agencies, national and international, investment from the industry, and participation of charities and private foundations; iii) the training factor: interest in RD research has to be encouraged in young people at the graduate school of medicine, biological sciences, public health, and healthcare professionals to promote the professional and research interest; iv) core facilities: patients’ registries and biobanks; v) the healthcare system: reorganisation of health resources and creation of centres of expertise to facilitate research into the clinical practice; vi) the social factor: promotion and empowerment of patients and their role as participants in clinical studies or clinical trials; vii) the political factor: RD as a main topic for public research.

O17 Health technology assessment: oncology drugs with orphan designation as an example
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Background: Since (limited) health care resources have to be invested efficiently, HTA/ health technology assessment is applied even more often in many health care systems for ‘rational decision-making’. Around 40% to 50% of all drugs with orphan designation are high-priced oncology drugs. Within EUnetHTA (an EU-supported project) a work package deals with the pre-coverage exchange of early assessments among European countries and the collaboration on projects for further generation of evidence.

Methods: Description of methodology of the early assessment of oncologic drugs and possible mechanisms of exchange of pre-coverage knowledge within Europe.

Results: A temporary coverage/funding of drugs with orphan status often requires additional collection of data on safety, effectiveness, cost-effectiveness, and the appropriate use of the drug. Many of the oncology drugs show little (or marginal effectiveness) at time of approval and reimbursement agencies demand further data before deciding whether to cover the new drug. Pragmatic clinical trials, patient access schemes and standard data requirements on patient relevant outcomes in registries across Europe are some of the approaches to generate further evidence and to fill the gap between knowledge on efficacy at time of approval and demanded knowledge on effectiveness for coverage decisions. EUnetHTA provides the necessary structures for coordinated efforts.
Conclusion: Exchanging information on and developing tools to facilitate evidence generation and collaboration on the assessment of new costly technologies, many of them drugs with orphan designation and a reduction of duplication of assessments is the intention of EUnetHTA.

Predictors of orphan drug approval

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Objective: To encourage the development of drugs for rare diseases, an orphan drug legislation has been introduced in the USA (1983) and in the EU (2000). Recent literature provides criticism on the slow development of orphan drugs in the EU. This study aims at identifying predictors for successful marketing authorisation of potential orphan drugs in the EU and the US.

Methods: Using publicly available data, the number of orphan designations and orphan drugs in the EU and the US was determined. A comparison between randomly selected authorised and a matched sample of not yet authorised orphan drug designations in the EU has been performed. Determinants in the study included characteristics of the indication, of the product and of the sponsor.

Results: More orphan drugs were developed in the US during the first ten years of the US Orphan Drug Act (1983-1992, N=73) and during the first ten years of the EU Regulation on Orphan Medicinal products (2000-2009, N=112) than in the EU (2000-2009, N=59). Orphan drug approval was strongly associated with previous experiences of the sponsor in obtaining approval for another orphan drug (OR=17.3, 95% CI=5.6-53.1). Furthermore, existing synthetic entities compared to biotechnology products tended to have a higher likelihood of reaching approval status (OR=3.9, 95% CI=0.9-16.6).

Conclusion: This study showed that the experience of a company in developing orphan drugs is an important predictor for subsequent authorisation of other orphan drugs. The same applies for existing (synthetic) molecules, for which more knowledge is available. Companies or institutions wishing to develop an orphan drug should therefore seek experienced assistance and engage in dialogue with the regulatory authorities.

Cross-border genetic testing

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As genetic testing (GT) is now available for many rare disorders, no GT laboratory can be expert enough to provide every test. Samples are therefore sent between laboratories for testing, often across borders. Such cross-border testing raises some questions:

The 2003 OECD survey of 827 molecular GT laboratories in 18 countries found that 64% of laboratories had received samples from abroad. This percentage is likely to have increased in the intervening years, but no data are available. Samples are usually sent abroad for specialist testing, but factors such as patents on genes and the centralisation of testing by private pathology companies also contribute to the traffic.

The main issue identified in the OECD report was the wide range of quality frameworks operating in different countries, leading to uncertainty about the quality of GT results obtained when samples are sent across borders. EuroGentest, with Orphanet, has established a quality assurance database of 1500 laboratories providing genetic tests in Europe. The database uniquely includes validated information on accreditation status and EQA participation for each laboratory and each test. For the public, this database facilitates an informed choice of laboratories; for genetic services, it allows the selection of partners for referral of tests based on their commitment to quality; for the laboratories, it valorises their efforts and investment in quality assurance.

While the in Vitro Diagnostic (IVD) Directive regulates the quality of diagnostic devices in the EU, most GT is exempt from the Directive as tests are manufactured and used in the same institution. EuroGentest has proposed that this exemption from CE-marking should be retained in the revised IVD Directive but it should be restricted to laboratories accredited to ISO 15189 or equivalent. This would protect the availability of rare disease testing, but ensure that it was carried out in laboratories with a robust quality system.

Comparative demographics of the European Cystic Fibrosis population: does EU membership confer an advantage?

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Background: Country-specific patient rare disease registries are rarely used to make international comparisons because of protocol discrepancies in data collation. Here, we attempt to overcome this limitation by using the inherited disease Cystic Fibrosis (CF) as a paradigm. CF provides a good example because its common form (homozygous F508del-CFTR) occurs across all European social strata appearing frequently but randomly thus providing an opportunity to measure health outcomes.

Methods: Country-specific CF Registries were combined cross-sectionally using a common data protocol (http://www.eurocarecf.eu) to compare patient demographics between the European Union (EU) and non-EU countries using EU membership in 2003 as a reference base. We tested the hypothesis that the nine-fold higher resources within the EU would translate into better outcomes.

Findings: Data were collected on age, age at diagnosis and CF genotype from 29,025 CF patients registered in 35 European countries. Median age was 16.3 years but was ~4.9 years older in EU countries (17.0 years) than non-EU countries (12.1 years; p<0.001). CF for the difference was 4.5-11 years, a significant difference (OR 2.4, 95% CI 1.9 - 3.0). Under-ascertainment was unlikely because the relative paucity of F508del-homozygous patients outside the EU was also significant (95% present clinically in childhood). We estimate that the current CF population of non-EU countries would rise by 84% if they had a CF demographic profile comparable to those of the EU countries who were already EU members in 2003.

Interpretation: Given that neither the CF carrier frequency nor the relative territorial population size is significantly different between the EU and non-EU participants, the reasons for this apparent deficit in CF patients of a common genotype in non-EU countries require explanation. It may be that under diagnosis and premature childhood mortality are the main drivers of the relative paucity of CF in non EU states.

Classification of rare diseases: a worldwide effort to contribute to the International Classification of Diseases

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Most rare diseases are absent in the International Classification of Diseases (ICD10) and those with a specific code are often misclassified. As
a consequence, morbidity and mortality due to rare diseases is invisible in health information systems. To overcome this difficulty, Orphanet (http://www.orpha.net) has established a partnership with WHO to ensure a fair representation of rare diseases in general. Orphanet has collected all published expert classifications and established a database of phenotypes indexed with ICD10 codes, MIM codes, genes, mode of inheritance, age of onset and class of prevalence. Phenotypes are assigned to as many classification systems as necessary to represent them. The Orphanet nomenclature of rare diseases is a stable one, directly exploitable by information systems and available on request. A WHO Topic Advisory Group on rare diseases has been established to manage the revision process. The first revised chapters currently circulating among experts and expert groups for review are Haematology, Endocrinology, Nutrition, Metabolism, Immunology, Neurology and Malformations. Revised chapters follow a primarily clinical approach, only secondarily an aetiological one up to the gene level. When several possible names are available for a disease, descriptive names formed in accordance with a clinical approach are preferred. Every entity is assigned a unique identification number. Rare diseases affecting several body systems are included in every relevant chapter, as ICD11 will be poly-axial, but a main code is proposed to allow for linearisation, according to the most severe involvement and/or the specialist most likely to be relied on for the management of the disease. The rare disease community is invited to take an active part as the results will condition the visibility of all activities in the field. All the revised chapters open for comments are available on http://www.eucred.eu

O22
Scope and management of Patient Registries for orphan rare disease (ORD) meeting the demands of all involved stakeholders
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Once a product for an ORD is approved; conditional or non conditional, there are still unanswered questions on long-term safety and effectiveness. And there is often limited knowledge on natural progression of the disease and optimal disease management. These questions could be answered by prospectively collecting real world data into patient registries. The scope of the registry should be in line with demands from registry stakeholders such as regulatory agencies, payers, patients, treating physicians and the pharmaceutical industry. Fabry and Hunter Outcome Surveys (FOS and HOS) supported by Shire Human Genetic Therapies have been designed and managed for almost 10 years in close collaboration with stakeholders. Patient registries do meet challenges related to data completeness and quality. Treating physicians are not mandated to provide real world data to patient registries. Over the years, global guidance on good practices for design, conduct, analysis and reporting of patient registry data have been developed to increase the quality and facilitate the usage of patient registry data. In addition, specific initiatives from FOS and HOS Governance bodies have been undertaken to increase the robustness of the collected data. FOS and HOS have contributed to increased disease knowledge and improved disease management. Examples on findings are increased knowledge of the natural progression of the diseases, disease involvement in patient populations not eligible for inclusion into the initial clinical trials as well as long term clinical benefit of treatment. However there are still questions to be addressed and answered especially to support pricing and reimbursement decisions in some European countries. This is to insure patients gets access to equal treatment on approved treatment independent of where they live in Europe. Questions that need to be more closely addressed and evaluated are beyond clinical effectiveness and related to patients capacity to work and implications on public spending for these ORD.

O23
Ageing in rare, chronic diseases
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With more and more treatments becoming available for people with rare diseases, we encounter the issue of ageing more often. A number of patients with rare, chronic diseases have experienced a number of remarkable transitions in their lives. The most important transition is the one from no treatment at all to treatment becoming available, like in haemophilia about 45 years ago. Another transition is the one that during their lives they go from one disease to a number of diseases (co-morbidity) through the natural process of getting older or as a consequence of the side-effects of treatment. These transitions can cause a number of problems. The first problem could be the lack of co-ordination between medical specialists and paramedical staff. The second is polypharmacy - the use of multiple medications and as a consequence of these first two problems a third problem occurs which I prefer to address as the ‘fear’ factor. The ‘fear’ factor can be described as a lack of control when you are not able to influence or check the treatment you receive. When more and more older persons with rare, chronic diseases will gradually transit to special care institutions for the elderly, more knowledge should be available in these institutions to deal with these new groups of patients. In this way, these patients will differ a lot from the group of elderly persons who develop co-morbidity at a much older age. Some of these issues will be addressed in this presentation, based on the personal experience of the speaker, who is also the (co-)author of several recent books on getting older with chronic diseases, like haemophilia and HIV.

O24
EMP's first steps in the field of clinical trials
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The development process of clinical trials has for long been the territory of the researchers, healthcare providers and regulators. Patients were only involved as "subjects" and had a passive role. However, in the last decennium, patients and their advocates have been moving more towards an active participation in the development of clinical trials. Many stakeholders realise that by actively involving patients, a patient tailored system of care can be achieved. However, it can be quite a challenge for patient organisations, who often work with volunteers and limited manpower to act as an equal partner in this specialised area where deadlines, standards and certain work principles have to be met. Moreover, patient representatives need to acquire enough “literacy” in this highly scientific environment. This document presents the case of a young patient organisation EMP and the way EMP handled this challenge. It explains how a patient organisation can proceed step by step to meet the exigent criteria and how literacy can be gained through different training programmes. The document further explains how EMP started active involvement in the development of clinical trials, by initially only focusing on activities that were within realistic reach of the group. These first prudent steps gave the patient representatives further knowledge and research skills; it gave them confidence and it created a relationship of trust with the other stakeholders. These are probably strong foundations for more extensive collaboration in the field of clinical trials in the future.
INFORMATION AND MEDICAL EDUCATION

O25 Contribution of rare disease patient organisations to medical education
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Orphanet Journal of Rare Diseases 2010, 5(Suppl 1):O25

Faced with about 7000 rare diseases, the limited knowledge of physicians is obvious, and deficits in diagnosis and therapy in consequence as well. Rare disease patients however, being usually confronted with only one disease or syndrome during their life time learn to cope with the symptoms, course and specificities of their disease and acquire expertise in diagnostics and treatments, as well as a feeling for a limited competence even of specialists. As disease-specific patient organisations provide a platform for the exchange of these experiences and for the provision of specialised information and/or services, the question arises: How is the knowledge of patients and the infrastructure of patient organisations utilised not only for the benefit of patients but also for the continuous education of doctors?

A Charité research project in cooperation with ACHSE (German Rare Disease Umbrella Organization), funded by the German Ministry of Health, among other aspects investigates the “Contribution of patient organisations to medical education”, by

- implementing a survey of rare disease patient organisations (members of ACHSE),
- providing the documentation, analysis and support of activities of rare disease patient organisations for the training and continuous education of doctors.

As the project is still ongoing at the time of the ECRD, only a few interim results and preliminary recommendations can be presented so far.

Method of data collection: - E-mail questionnaire survey of 90 member organisations of ACHSE with short introductory paper attached; - Good response rate of 58%, i.e. 52 member organisations responded by end March 2010

Preliminary overall results: - Most RD patient organisations are actively and successfully involved in numerous activities for and by doctors, researchers and clinicians specialised in the field of their respective diseases; - Priority is placed on the organisations of annual or biannual conferences, research colloquia and patient days followed by patient-oriented presentations, articles and publications in specialised professional meetings, journals, books etc.; - Training of medical students by patient organisations is far less important than education of doctors although some innovative activities exist.

Major preliminary recommendation: The regular observation, collection of data and support of educational activities by RD patient organisations for doctors and medical professionals is recommended, given their specific learning impact and benefit of interaction not only for professionals but also for patients, patient representatives, researchers and the wider public.

O26 Medical education: the role of patients
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Orphanet Journal of Rare Diseases 2010, 5(Suppl 1):O26

Health care professionals cannot be taught or know about 6000 to 8000 rare diseases. Though research, care and treatments progress every year, rare diseases are chronic, severe, complex and disabling conditions affecting patients and their families 24 hours a day. The national plans in France organise a 2 hours course for medical students to raise awareness and provide tools. 2 mothers of children living with rare diseases participated in a 20 hour pilot course in a medical school in Paris. During this course, they present the outcomes of the EurodisCare studies (12 000 questionnaires to patients and families throughout Europe), discuss their daily experiences with students and present the complexity of a very necessary comprehensive approach for rare diseases: medical, educative, social, from childhood to adulthood, extended to parents and siblings.

O27 The Swedish rare disease information database and the Swedish information centre for rare diseases
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The Swedish National Board of Health and Welfare database of rare diseases contains detailed documents describing over 250 rare diseases, and new texts are constantly being produced. There are currently more than 500,000 Swedish and international visitors every year, and the figure is constantly rising. People with rare diseases and their families, parent and patient organisations, professionals, researchers and public authorities are all regular users. The database is freely and easily accessible to all at http://www.socialstyrelsen.se/ovanligadagnoser. The Swedish Information Centre for Rare Diseases is the organisation commissioned by the Board of Health and Welfare to produce this material. The Information Centre is in regular contact with the most prominent Swedish specialists with expertise in rare diseases and works closely with them to produce the texts, ensuring that the documents are as clear and easy-to-read as possible. Patient and parent organisations supplement this information and a scientific advisory board reviews all documents before they are published. The texts are continually updated and revised. The information produced is available on-line but also in the form of printed pamphlets on specific diseases.

The Information Centre also serves as a helpline and a source of guidance to those directly or indirectly affected by rare diseases. It aims to increase awareness and understanding of these disorders not only by offering descriptions of the diseases, their symptoms, causes and treatment, but also by providing advice on habilitation, and information on relevant psychological, social and educational implications.

The Swedish Information Centre for Rare Diseases is run under the auspices of the Sahlgrenska Academy at the University of Gothenburg. It is funded by the Swedish National Board of Health and Welfare.

THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

O28 The European Union Committee of Experts on Rare Diseases (EUCERD): a new committee to help the European Commission advance in the field of rare disease policy
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The European Union Committee of Experts on Rare Diseases was formally established via the European Commission decision of 30 November 2009 (2009/872/EC). The EUCERD will aid the European Commission (EC) with the promotion and implementation of Community activities in the field of rare diseases, in cooperation and consultation with the specialised bodies in Member States, the relevant European authorities in the fields of research and public health action and other relevant stakeholders acting in the field.

The EUCERD replaces the EC’s Rare Diseases Task Force. Members of the EUCERD include representatives of: each Member State, patient organisations, the pharmaceutical industry, ongoing/past Community projects in the field of RD, ongoing/past RD projects financed by Community
Framework Programmes for Research and Technological Development, DG Sanco, DG Research, DG Enterprise, Eurostat, and the ECDC. The EUCERD will foster exchanges of relevant experience, policies and practices between these parties and is charged with the following responsibilities: assisting the EC in the monitoring, evaluating and disseminating the results of measures taken at Community and national level in the field of rare diseases; contributing to the implementation and improvement of Community actions in the field; contributing to the preparation of EC reports on the implementation of the Commission Communication and the Council Recommendation; delivering opinions, recommendations or submitting reports to the EC either at the latter’s request or on its own initiative; assisting the EC in international cooperation on matters relating to rare diseases; assisting the EC in drawing up guidelines, recommendations and any other action defined in the Commission Communication and in the Council Recommendation; providing an annual report of its activities to the EC. The EUCERD may establish temporary Working Groups including external experts for specific missions.

The Scientific Secretariat of the EUCERD is supported by an EC Joint Action.

RARE DISEASES IN CENTRAL/EASTERN EUROPE

O29 Cross-border healthcare? The Polish experience
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Patients with rare diseases are spread all over large distances, resulting in difficulty to find proper medical experts and centres and use new modern therapies. Cross-border healthcare enables equality in availability of medical services and the best medical care all over the world for affected patients regardless of their place of residence. In our centre, the cross-border healthcare is based mainly on informal personal cooperation in taking care of the patients with inborn errors of metabolism. We consult foreign patients from Eastern European countries; we diagnose the Polish patients through analysis done in materials sent abroad, take care of foreign affected children during their stay in Poland and continue the medical care of migrating patients. Some examples include, when a doctor visits a patient and consultations are performed by a metabolic team from our centre, in Riga and Vilnius, where among thirty two and twelve patients with suspicion of rare diseases, five and two diagnoses, respectively, were finally established. Moreover one of our patients diagnosed as hyperammonemia type II in the newborn period, at the age of thirty days was transferred by plane from our hospital to Heidelberg for liver cell infusions (the new modern procedure), which was successfully done with subsequent liver transplantation and a good outcome. In order to improve access to the cross-border healthcare, the following are needed: formal regulations among Member States, right for reimbursement for services provided abroad, establishing of European networks of reference centres for rare diseases, collecting data concerning availability and quality of medical management in order to enable comparable monitoring of cross-border healthcare and also developing e-health by including information technology in healthcare.

O30 The present situation of Rare Diseases in Central/Eastern Europe? The role of patient organisations
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Our goal was to introduce the limited available comparative data to characterise the situation in Central/Eastern Europe in the field of rare diseases. We collected the most important observations of current international surveys which were performed with or without the involvement of member associations of EURORDIS.

Results: 1. At the level of medical and social services: It is still "Incidental" to get to the appropriate expert or centre for diagnosis or treatment. It is difficult to find even the services, because of the lack of suitable "pathways" and referral. There are long delays in obtaining the first appointment, resulting in defencelessness and regional irregularity. The overall consequence is the lack of access to medical and social services. We also have difficulties with the supply of orphan medication and with the long duration of hospitalisations. 2. At the level of patient organisations: financial scarcity and uncertainty is typical and combined with inappropriate infrastructural background and human resources.

Conclusions: We need to continue our efforts for the formation of Centres of Expertise and their Networks together with the development of our National Plans. We also want to improve habilitation and rehabilitation services to compensate the social disadvantages. The relative low organisation level of patient organisations and the unsatisfactory level of their cooperation is also characteristic. More efficient, professional operations associated with strategic approaches, with the help of national and international cooperation is necessary!

O31 Cross-border health care represents a key issue in the field of rare diseases
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The Veneto Region, north-east of Italy, 4.7 million inhabitants, is interested by this phenomenon, both for its geographical position, a hinge between Eastern and Western European countries, and for the presence of specialised health care services, most of which are specifically addressed to RD patients.

The aim of the study is to analyse patients’ mobility in Veneto Region, especially from Eastern European countries, describe the established RD care network, and how it was successfully extended to other Italian Regions and its possible scalability to other neighbouring countries. In 2009, 17,769 RD patients were cared for in our Region, 16% coming from other Italian Regions. Foreign RD patients were 766, 3.3% of all the patients coming from abroad and 4.3% of all RD patients followed in the area. Comparing these patients to Italian RD patients, the paediatric component is more represented (44% vs 30.3%). Congenital malformations are the most frequent diagnoses (16.5%), followed by metabolic diseases (9.2%). Foreign RD patients move to undergo very specialised interventions, i.e. heart surgery (n=52), organ transplantations (n=36), or to obtain second opinions. 67.5% come from European countries, in particular 49.2% from Eastern countries, both EU and non-EU. These patients are referred to Centres of Expertise, officially identified, which are part of a wider interregional network, based on the collaboration between specialised Centres and primary care services. The continuity of the care process is supported by the use of shareable patients’ electronic records in neighbouring Regions and by the elaboration of common management protocols for specific RD. RD patients’ mobility should lead to the development of common policies and services addressed to these patients involving neighbouring Regions/ countries. Preliminary intercourses already occurred with Slovenia in order to share best practices in the complex field of RD patients’ care.

O32 Patient involvement and empowerment through the NPRD (Eastern Europe)
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Orphanet Journal of Rare Diseases 2010, 5(Suppl 1):O32

Patient involvement and empowerment through the National Plans for Rare Disorders (NPRD) (Eastern Europe)
We present the current situation of the NPRD from Bulgaria, Romania and Hungary: three different countries confronted with the same problems and limits, and most probably not so different realities.

Bulgaria: the NPRD was officially launched on 1 January 2009. The draft for rare diseases and orphan drugs programme was prepared by BAPES and proposed to the Bulgarian Ministry of Health (MoH) in November 2004. The national plan includes 9 priorities (total budget 11 294 515 Euro).

Romania: RPWA established RONARD – and organised the first working groups on a National Plan for RD on 9 August 2007. 1-2 November, RONARD organised the first National Conference on RD. 29 February 2008 – signed a partnership agreement with MoH – NPRD with 6 priorities. The first national programme on RD started in June 2008. Still there is no NPRD in the National Strategy for Health. Hungary: there are several improvements but still before designing the Plan. HUFERDIS and enthusiastic professionals pushed it, but they have four running national health plans, and there is no more money for a new one.

In all three countries, Patient Organisations and medical professionals are the main engine for drafting and lobbying for a national strategy on RD. They organised information campaigns with stronger and stronger impact, help lines, contribute in designing social and medical health care services, initiate therapies, respiratory care, involve in research and clinical trials, etc. They organised working groups with members of the national alliances, networks and advocates at national level. The Plan should be promoted both to parliament members and Ministry of Health. More “actors” have to be involved because RD patients need varied social assistance as much as medical care. EURORDIS offered specific tools for lobby and advocacy actions toPOs. Recommendations and Communications of the EC give the best tool to promote RD as a public health and research priority.

POSTER PRESENTATIONS

SERVICES TO PATIENTS, FAMILIES AND CARERS

P1

WHO International Classification of Diseases (ICD) Revision Process: incorporating rare diseases into the classification scheme: state of art

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World Health Organisation has established various Topic Advisory Groups to serve as planning and coordinating advisory bodies in the update and revision process for specific areas of the ICD. A Revision Steering Group oversees the overall revision process. Working groups organised by the Topic Advisory Groups (TAG) review the proposals. A TAG for rare diseases was established in April 2007 as rare diseases should now be traceable in mortality and morbidity information systems. The production of the basic information needed to establish an Alpha draft of the classification of rare diseases has been assigned to Orphane and may serve as a template for the whole revision process, as rare diseases are present in all areas of medicine. Currently, the Orphane database includes over 6,000 distinct phenotypes which are classified according to published classifications. These classification systems are mainly based on scientific grounds (aetiology and mechanism). To complement these classifications, Orphane has developed a strictly clinical in-house classification to meet the needs of clinicians: they can be viewed on the Orphane website. They serve to elaborate a proposal for the ICD revision. The first revised chapters currently circulating among experts and expert groups for review are Haematology, Endocrinology, Nutrition, Metabolism and Immunology. The next chapters to be considered are Neurology, Malformation and Multi-systemic diseases. Input from the Rare Disease Community is expected. It is the responsibility of TAG members to contact experts from their region of the world to ensure the widest possible consultation. The alpha draft of the chapters which have already been revised will be published in April 2010 and the beta draft, for field testing, is planned for 2011. The budget of the working group on coding and classification of rare diseases is currently provided by a grant of the European Commission supporting the activities Rare Disease Task Force’s Scientific Secretariat.

P2

European Project for Rare Diseases National Plans Development (EUROPLAN)

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EUROPLAN (http://www.europplanproject.eu) is a three year EU funded project (2008-2011) coordinated by the Italian National Centre for Rare Diseases (Istituto Superiore di Sanità, Italy), which involves 30 Countries and EURORDIS. The Council Recommendations on European Action in the field of Rare Diseases (RD) which was adopted by the EU Council in June 2009 calls upon Member States to adopt National Plans for RD, before the end of 2013. In line with this, EUROPLAN is an operational measure within the European strategy in the field of RD.

The project aims at a) elaborating recommendations on the different steps to develop a national plan or strategy; they will include priority areas and actions of intervention supporting the harmonisation of public health strategies on RD throughout Europe; b) select indicators for monitoring the implementation and evaluating the impact of national plans or strategies.

In order to turn the Council Recommendation into concrete actions in favour of RD patients at national level, several rare disease patient alliances, coordinated by EURORDIS in conjunction with national authorities, are organising National Conferences to be held in 16 Countries in 2010. During the National Conferences local stakeholders will discuss the EUROPLAN recommendations and the main elements of the European strategy on RD, with the aim to assess their transferability in their own Country.

In conclusion, EUROPLAN is collecting information on EU Member States initiatives contributing to share experiences, data and effective strategies to address RD; is promoting the development of RD plan or strategy trough recommendations and indicators; is increasing awareness on RD and the recommendations will also serve as an important advocacy instrument at policy level. The EUROPLAN outcomes will support and encourage EU MS in developing national health policies to ensure equal access and availability of prevention, diagnosis and treatment for citizens with RD.

P3

EURORDIS Summer School for patient advocates in clinical trials and drug development

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EURORDIS, the European Organisation for Rare Diseases launched this new initiative in 2008 with the support of the Drug Information Association (DIA) Philanthropy programme, the Association Française contre les Myopathies (AFM), Fundacío Dr. Robert and the European Agency for Health and Consumers (EAHC). Many of EURORDIS’ members are extensively involved in EU decision-making processes in the European Medicines Agency (EMA), including as experts, patients’ representatives and/or members of scientific committees and working parties.
The aim of the EUORDIS Summer School is to assist these and future patient advocates to i) better promote drug development, ii) be involved in regulatory affairs, and to iii) guarantee access to treatments and iv) improve the quality of orphan drug information to patients.

The 4-day programme consists of:

1) a description of the drug development process from clinical trials to regulatory stages and includes statistics, ethics and post-marketing aspects.

2) a description of the committees and working parties at the EMA with particular emphasis on the role of patients’ representatives and

3) the role of EUORDIS and in particular of its “Task Forces” whose volunteers can also be involved in the above activities.

The aim of the Summer School is to:

1. to establish a forum for exchange of information, education and training for these high-level selected patient advocates from a wide range of rare diseases across Europe;

2. to enhance the experience they have gained and their leadership capacities.

As patient advocates are increasingly involved in all aspects of drug development (from research to economic aspects), there is a growing need to provide support and training in these areas. The EUORDIS Summer School not only addresses these needs but also provides the means for advocates to interact with each other as well as with regulators, academic partners and industry. In addition to the learning experience, the Summer School provides an excellent forum for in-depth exchange of information and experiences, all in the beautiful city of Barcelona.

P4

Genetic testing in Europe: transborder testing is a necessity

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Genetic tests are now offered internationally, through both public and private sector genetic testing services. Physicians prescribing these tests and biologists receiving the samples need to know which tests are available, where they are performed and whether the identified laboratories meet quality standards. To fulfil this need, http://www.orpha.net was launched thirteen years ago to set up a database of clinical laboratories in the field of rare diseases. Data was collected in 1 country in 1997, 15 in 2003, 26 in 2006 and 38 in 2010. This major effort was made possible thanks to resources from the EC DG for Public Health. In collaboration with the EuroGentest Network of Excellence, information on quality management has been added to the Orphanet database over the past four years. To obtain information on genetic testing in Orphanet, it is possible to search by disease name or by gene (symbol or name in English) in addition to the traditional search by name of laboratory or professional. The information provided on laboratories includes data on quality management. Currently, 956 laboratories offering tests for 1,559 genes are registered in the Orphanet database. The test offer differs greatly from one large country to another: Germany (1,141 genes), France (874 genes), Italy (625 genes), Spain (582 genes), UK (414 genes), Medium and small-sized countries have a test offer ranging from 1 to 233 genes. This situation explains the large cross-border flow of specimens and underlines the need to provide access to services in other countries when necessary, especially for very rare diseases. Testing for Cystic fibrosis is the only service which is provided by every country. The distribution of this test offer will be presented.

P5

Rare diseases research in Europe: an overview based on data from the Orphanet database

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Orphanet compiles 6500 research projects. This information has been analysed to identify areas in need of collaborative research projects and to target future calls for proposals. The analysis of the distribution of number of diseases by number of treatments in development showed that most RD have no more than 3 orphan designations, whereas 53 RD have over three orphan designations. Similar results were obtained when clinical trials, marketed drugs, patient registries and preclinical/epidemiological/basic research were analysed. Some of the diseases which were over-represented upstream in the process of R&D (with a treatment on the market or drugs in development) are also well represented regarding ongoing research, like Cystic fibrosis, pulmonary arterial hypertension and some rare cancers. Diseases with a higher prevalence are anticipated to have more treatments in development: this assumption is not backed up by our data analysis. The best represented medical domain in terms of percentage of diseases with MA/OD is rare tumours, followed by Systemic and Rheumatologic diseases, Respiratory diseases, Immunological diseases, Metabolic diseases and Haematological diseases. It seems that the most mature fields keep on investing in research and are also the strongest ones regarding the products in development, and even in basic research for some of them. Other fields, however, like Neurology, seem to be essentially in development, since the percentage of diseases with MA is low compared to other domains and to the percentage of neurologic diseases with OD, clinical trials and research. The absence of orphan designations for some medical domains, like Cardiology, could be explained by the fact that the Cardiology rare diseases benefit from treatments already available for these diseases’ common forms. This work was supported by the RareDiseasePlatform contract (RDPlatform), a three-year project which began in May 2008, financed by the European Union’s Seventh Framework Programme (HEALTH-F2-2008-2012).

P6

Cystic Fibrosis in Europe - remote measurement of outcome

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Background: Cystic Fibrosis (CF) occurs randomly in children across all European social strata providing an opportunity to sample their underpinning health care provision for this rare disease in an unbiased manner. We developed a 35 country European cystic fibrosis (CF) demographic registry to compare CF outcomes through an FP6 programme of the EU called EuroCareCF (http://www.eurocarecf.eu).

Methods: We applied methods (http://www.cystic-fibrosis.org.uk) previously used to create country-specific registries after inviting participation through the European CF Society and CF patient organisations using a double hub (Dundee and Prague) and spoke model supplemented by conferences, workshops and telephone support using a single data collection system. Implementing this common data collection platform, we collated demographic and genotype data in around 30,000 patients scattered from Iceland to the Black Sea.

Results: Amongst the ~30,000 CF patients in our Registry, a widely different country-specific prevalence of childhood CF exists that cannot be explained by differences in population size, underlying heterozygote CF gene frequency or under-ascertainment. In particular, we do not believe that the lattermost can explain our findings.
because in late childhood, we observe a significant paucity of the clinically severe homozygous F508-del form of CF that is of early childhood onset in 90% of cases and is widely dispersed across mainland Europe.

Conclusions: It is likely that an excess premature CF mortality in childhood still occurs across many parts of Europe, a mortality that has largely disappeared in countries such as the UK, France Germany and other wealthy nations. We suggest that much of (better resourced) Western Europe now has a vanishingly low mortality for the severely commonly occurring F508del homozygous CF in childhood that is not replicated in our study in many European countries. The reasons require investigation.

P7
How a motivation programme can affect complex treatment compliance in a rare disease? Results of a questionnaire-based, self-reported study to evaluate “Life Club CF”, a programme intended for patients with cystic fibrosis
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Orphanet Journal of Rare Diseases 2010, 5(Suppl 1):P7

Background: “Life Club CF” is a motivation programme for patients with cystic fibrosis (CF) in Poland, initiated and coordinated by “MATIO” Foundation (Krakow, Poland). Participants collect points for regular specialist outpatient follow-up, adequate nutrition, conducting physical exercise, regular drug intake and reduction in the number of exacerbations. These factors play a major role in complex treatment of CF. Patients may exchange collected points for rewards, such as toys, digital cameras, computers and others. The program was evaluated in a questionnaire-based study.

Aim: The programme was created to increase treatment compliance through promoting responsible activities among patients with CF and encouraging regular, multidisciplinary outpatient follow-up. Questionnaire-based study was conducted to evaluate how the programme accomplishes these objectives.

Methods: A questionnaire consisting of two parts was used: general evaluation (2 questions) and activity before and after having signed up in the programme (7 single-choice questions: rhDNAse intake, body mass control, physical activity and attending outpatient consultations – CF physician/pulmonary specialist, physiotherapist, dietician and psychologist). It was completed either by patients or parents/caregivers. Chi-square and Fisher-Freeman-Halton tests in StatsDirect software package were used to analyse the contingency tables of answers for statistical significance.

Results: 89 questionnaires were returned (53 women and 36 men). The studied group consisted mostly of children with CF; 86.5% were aged 18 and less (mean age: 13.7 ± 5.6). Significant differences in frequency of dietician (once a year: 9% vs. 21.3% / once in 6 months: 31.5% vs. 48.3%; p=0.0009), physiotherapist (once in 6 months: 19.1% vs. 20.2% / once in 3 months and more often: 21.3% vs. 39.3%; p=0.025) and psychologist consultations (once a year: 12.4% vs. 16.9% / once in 6 months: 18% vs. 34.8%; p=0.027) before and after participating in the programme were detected. Furthermore, significantly more CF patients focus on increasing body weight (57% vs. 75%; p=0.013) and regularly perform physical exercise (19% vs. 31%; p=0.044). No significant differences in rhDNAase intake and frequency of CF clinic consultations were detected.

Conclusion: Motivation programme “Life Club CF” is a novel and effective method in increasing treatment compliance in important aspects of complex CF care, particularly multidisciplinary outpatient follow-up, physiotherapy, nutrition and physical activity.

P8
Cell therapies for Duchenne muscular dystrophy: some ethical issues for personalised medicines
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Duchenne muscular dystrophy (DMD) is a chronic, complex, genetic childhood disease affecting boys, where sufferers typically die in their 20s. DMD is caused by mutations in the dystrophin gene and trials are currently underway with exon skipping, a therapy which works by patching the mutation thereby allowing production of a functional gene. The dystrophin gene contains 79 exons and mutations can be found in one or more exons. Treatment must be targeted to the specific exon which is at fault and each one therefore requires a different exon skipping chemistry. The success of trials with exon skipping will present patients and physicians with the possibility of a personalised medicine and with that, a number of ethical issues. Firstly, equity and harm by omission. Exons for the most common mutations will be tested first and researchers assert that conforming to the existing regulatory pathway for every exon, it will not be cost effective. This raises the real possibility of those children with rarer exons being denied a potentially lifesaving treatment. Secondly, if therapies are developed for the rarer mutations, risk and uncertainty become more difficult to assess. In some cases a boy may be his own control in a RCT and the boundaries between experiment and therapy become blurred. Thirdly, justice and minimum entitlement: given that minimum entitlement in this case is likely to be very excessive (the comparable myozyme therapy costs $300,000 per person, per year), the distribution of limited health resources throughout a population is once again under scrutiny. This paper aims to anticipate these ethical issues with a view to helping researchers, clinicians, regulators and patient organisations to find ways to ensure that the development of these life-saving therapies progresses.
Promotion of research on all aspects of OI, in cooperation with an international OI-registry based in the US
Collection and publication of information about OI
Support of member-societies by the exchange of information and experience
Promotion of public awareness of OI
Education for doctors and other professionals by the organisation of international conferences, topical meetings and workshops
Support and coordination for international Student exchange for young OI people

P10
Primary Immunodeficiencies (PID): driving diagnosis for optimal care in Europe
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Primary Immunodeficiencies (PID) are defects of the immune system that cause increased susceptibility to infections that are chronic, persistent, recurring, debilitating, and often fatal. There are more than 150 PID diseases, which affect more than 10 million people worldwide. PID are often manageable, if properly evaluated and treated. However, awareness of these diseases is low amongst both primary care physicians and the general public. As a result, many patients are left undiagnosed. Delayed diagnosis and insufficient treatment lead to increased morbidity, mortality, and inflated medical costs, in addition to a life of chronic illness and suffering. The Jeffrey Modell Foundation (JMF) was established in 1987 in memory of our son, Jeffrey, who died at the age of 15 after struggling with a PID. JMF’s mission is to assure early and precise diagnosis, meaningful treatments, and ultimately, cures of the ever increasing known PID diseases. JMF initiated a Physician Education and Public Awareness Campaign to create greater awareness and encourage the earliest possible diagnosis and precise treatment. This global campaign has had extraordinary results. Expert physicians from the Jeffrey Modell Centres Network of 72 Diagnostic and Research Centres and more than 196 Referral Centres worldwide have reported annual increases of more than 25% in the numbers of patients referred, patients followed, and patients identified with a specific PID. To date, more than 414 physicians from 176 teaching hospitals in 51 countries participate in the Jeffrey Modell Centres Network. Nearly half of the Jeffrey Modell Diagnostic, Research and Referral Centres are located in Western, Central, and Eastern Europe. This European Network of Diagnostic Centres promotes improvements in the standard of care for patients with PID by fostering collaboration, measuring outcomes, sharing data, and serving the patients, families and caregivers of those affected by PID, and serves as a successful model for other diseases.

P11
Orphandev, French Clinical Trials Network dedicated to Orphan drugs and therapeutics development for rare diseases
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Orphandev is a French Clinical Trials Network based on a strong collaboration principle with all actors involved (academics, industries and patients) dedicated to orphan drugs development. It was created by academics to help academics but also other actors involved in rare diseases’ research. The Network’ skills have already been dedicated to orphan drugs’ trials: Charcot Marie Tooth (2004), Ret (2006) and Progeria (2008). Orphandev has intervened according to the mutualisation and translational research concepts from the experimental phase (in vitro and animals’ tests) to the results’ valorisation with every single actor involved in the study. With the experience gained and the successful results, we have developed an organisational concept in order to capitalise on the lessons learnt and optimise the trials process.

However during the last decade, France led several initiatives to improve the burden of rare diseases. Centres of Expertise were identified, ability centres have been appointed and a National Plan for rare diseases was developed. Nevertheless, in spite of the great dynamic created by France, development and availability of orphan therapeutics remain problematic regarding rare diseases specificities. In this context, it is important to gather skills and strengths to make patients benefit from fundamental research’s results and accelerate clinical trials. Orphandev is a French Clinical Trials Network based on a strong collaboration principle with all actors involved (academics, industries and patients) dedicated to orphan drugs development. It was created by academics to help academics but also other actors involved in rare diseases’ research. The Network’ skills have already been dedicated to orphan drugs’ trials: Charcot Marie Tooth (2004), Ret (2006) and Progeria (2008). Orphandev has intervened according to the mutualisation and translational research concepts from the experimental phase (in vitro and animals’ tests) to the results’ valorisation with every single actor involved in the study. With the experience gained and the successful results, we have developed an organisational concept in order to capitalise on the lessons learnt and optimise the trials process.

In a time of great therapeutics development with solutions coming from gene breakthroughs but not only, Orphandev allows for further improvement of the interface between fundamental research, clinical research and drug developments in rare diseases in a more operational way.

P12
Registry of Outcome Measures (ROM): tools supporting review and selection of outcome measures (OMs) for studies and trials
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Background: Selecting the right OMs for clinical trials/studies is critical to success. Unless it is done it can be a major barrier to translational research. The choice is best made by systematically reviewing existing OMs to identify suitable measures and inform decisions about adapting existing OMs or creating new ones. ROM helps this effort by offering information on an expanding number of potentially suitable OMs. We have added web-based tools to support the review and selection process.

Method: ROM incorporates:
1) a ‘Tree of OMs’ - allows the reviewer(s) to record OMs by category as being considered for a specific study or trial
2) a search engine that enables investigators to find potential OMs in ROM
3) a comparison table that displays information about multiple OMs to aid selection
4) a document in progress which will evolve into a Manual for the review and selection of OMs

These web based tools are easily accessible to collaborative groups. They can be open access so that all investigators can see work in progress, avoid duplication of effort, and contribute their views.

Results: These tools have led to the publication of more OM records in ROM and are appreciated by investigators.

Conclusion: These new tools on http://www.researchom.com will play an important part in helping translational research.

P13
E-learning for carers
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The Centre for Rare Disorders is an interdisciplinary, nationwide competence centre which offers information, counselling and seminars on a selected range of rare disorders.
The purpose of the e-learning programme in question is to improve quality of life for patients with Huntington’s disease by increasing the carer’s knowledge. E-learning and interactive sharing of information can be an effective and secure way of providing professionals with new knowledge. E-learning also functions as a tool for network-building.

Methods/techniques: The e-learning programme is organised as a module based compilation of knowledge and competence gathered by the Centre for Rare Disorders and its collaborators over the last 15 years. In addition to the modules, the users have shared information in on-line discussions. Their level of knowledge was evaluated through several tests.

Security: The participants received a password and username for logging on to the programme.

P14
Social profiles - a dialogue tool
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Rare disease patients and their families often navigate in unknown territory. Even if medical guidelines for their disease exist, they normally do not mention social and educational matters and the public services in these areas are often inadequate. Because the disease is rare, social workers, teachers and care takers have no knowledge of the consequences and necessary support.

Rare Disorders Denmark has developed a tool, Social Profiles, for the dialogue between the rare disease patients and professionals, that - gives a short description of the diagnosis and its characteristics in lay terms - provides a check list for relevant support in a life time perspective, for instance:

- Necessary considerations in general, e.g. assistive technology and interior special design
- Special needs of the small child, the taller child and the youngster, e.g. needs for extra resources in day care, special resources at school and special vocational guidance
- Special needs of adults, e.g. special programmes to obtain labour market contact or special housing
- Contact with the relevant patient society

The Social Profiles have been developed in a process involving rare disease patient societies and exist for 15 rare diagnosis, with more to come.

The Social Profiles consist of verified facts only and are published at the website http://www.sjaeldenborger.dk (meaning: http://www.rarecitizen.dk).

The Social Profiles have been developed within a state funded project of 420,000 € over a 4-year period, also containing - Upgrading of skills for patient society advisers - Virtual tool kit for rare patient societies - Raising awareness about rare disease patients in order to avoid stigmatisation

P15
Abilities of development support in children with genetic syndromes.
Experiences from annual international meetings
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Recognition of a genetic syndrome, as a course of psychomotor development retardation, requires genetic counselling where a prognosis of development for children with different syndromes is given. Recent knowledge according respective genetic syndrome defines: how to diagnose them, describes the child’s limits in comparison to coevals, usually do not obtain any data according to the child’s abilities, which we may use in our stimulation advance with children during their individual developmental profile. The pessimistic vision of child’s development leads towards social isolation and even isolation of the whole family. Looking for a solution against social isolation of children with genetic disorders with mental handicap and deprivation of their families motivated us to organise conferences, which would integrate clinical geneticists, other doctors, psychologists, pedagogues, therapists and families associated with support groups. This idea allowed for the building of a dialog platform between specialists, presenting the newest discoveries of native and European scientists and parents taking part in the conference together with children, as specific experts of their child. The meetings were a special form because individual and group medical and pedagogical consultations of patients and/or their families with medical students and young physician were included. The aim of respective conferences about such syndromes as Down s., Rett s., Prader-Willi s., Angelman s., Wolf-Hirschhorn s., Cat cry s., Russell-Silver s., Neurofibromatosis type I, Cornelia de Lange s. and Williams s. was to point out the necessity of a collaboration among different groups of specialists taking care of children with genetic disorders together with parental support groups. This kind of collaboration is important for the recognition of the important role which parents play in the development of knowledge about genetic disorders and of public awareness.

P16
European Porphyria Network (EPNET) for information, epidemiological data, quality and equity of service
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Porphyrias are metabolic diseases that affect about 1 in 20,000 of the EU population. Most healthcare professionals rarely encounter these disorders and there is disparity between countries in the levels of support provided for patients and their carers.

EPNET is a DG Sanco funded project (1/4/2007-31/3/2010) with 15 participating countries that aims to establish a network of specialist porphyria centres, each working to agreed quality criteria. Work achieved: 1. Information on porphyria for patients in 10 languages, and for healthcare professionals in English, is available at http://www.porphyria-europe.org with easily downloadable pdf files.

2. Data on the safety of drugs in acute porphyria has been collected from participating countries (2001 drug reports on 616 drugs). Information is disseminated on http://www.drugs-porphyria.org

3. European Quality Assessment (EQA) schemes have been established for specialist laboratories (24 participants in 17 countries) and data on laboratory performance collected annually. Variations between centres are being addressed and diagnostic protocols developed.

4. The EPNET registry now contains data on 371 patients (320 new cases; 51 with long term complications) that is being used to calculate the incidence of each porphyria in participating countries and the prevalence of long term complications, The incidence of new cases of acute intermittent porphyria is around 0.15/million/year in most countries.

The initial purposes of the EPNET project have been achieved; we have established a rich data resource, a network for improving quality of care and a European platform for expert exchange.

EPNET has been funded by the European Commission through its Public Health and Consumer Protection Directorate (DG SANCO), PHEA programme.
The aim of the study is to provide information about aging and the need for services among persons with Marfan syndrome. Health and welfare service providers need research based knowledge to offer this new category of elderly multidisciplinary help and counselling along the life course. There is a lack of knowledge about aging among people with a rare diagnosis and how they manage their disability in daily life during the life course. Advances in medical treatment have greatly improved the outcomes for persons with rare diagnoses, among others persons with Marfan syndrome. Many now reach an advanced age.

Marfan syndrome may not cause any recognisable impairment, but aging and increasing health problems create challenging situations in daily life. Information about the diagnosis and its combination with special competence is important for daily life, health condition, self image and identity. Norway has a special model with 16 national centres for rare diseases, supporting better quality of life. But there is little knowledge about the ageing processes.

Methods: Qualitative group interview with eight persons who have Marfan syndrome and qualitative in-depth interviews with ten persons having Marfan syndrome (≥40 years of age) are transcribed. Life course experiences are analysed with a focus on the need for information, the impact of participation in patient organisations and support from the centres.

Results: The findings show the importance of having a «normal» life course, work and activities. For persons with Marfan syndrome biological and physiological changes which originate from the disorder play together with physical changes connected to ageing. The patient organisation and the national centre help the person managing challenges in daily life and the health situation in many ways. The study reveals the difficult balancing needed to function «normally» and at the same time accepting the life course consequences.

Evidence-based information guides to rare chromosome disorders for families and professionals
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The purpose of this project is to develop reliable, relevant, accurate leaflets for affected families and health (and other) professionals that fill an information gap about rare chromosome disorders. In 2003 Unique surveyed information materials published in the UK about specific rare chromosome disorders: for over 93% of members, no accessible disorder-specific information was available. Unique asked families what they most wanted to know at diagnosis and what questions remained unanswered. Unique prioritised 66 disorders according to frequency on its database (7,140 member families at February 2010) and absence of existing information accessible to families. Information was compiled from the medical literature, from Unique’s database and from detailed surveys sent to member families. Draft texts were reviewed for accuracy by Unique’s medical adviser and by medical and genetics professional experts in the specific disorders. Photographically illustrated draft leaflets were vetted for content by families. By early 2010, leaflets have been developed on 113 rare chromosome disorders including numerical and structural disorders, subtelomere deletions, mosaic disorders, emerging microdeletion and microduplication syndromes and a broad range of less common diagnoses. Twenty-one leaflets have been translated into at least one European language. Many more leaflets are in preparation or planned. Leaflets are available free to families and the professionals who work with them either in print format or online from Unique’s website at http://www.rarechromo.org. The leaflets improve families’ understanding and acceptance of a rare chromosome disorder and help diminish the acute stress and anxiety associated with diagnosis. They are also proving to be a useful resource for professionals, including health professionals in clinic.

EU Clinical trial regulation in the environment of rare diseases: time for a change
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The clinical trial directive 2001/20/EC is out for public consultation. This session will draw the attention of the challenges faced by Small and Medium Enterprises (SME) companies when developing new products for the rare diseases that affect a very limited number of patients and discuss possible options to overcome the challenges. Indeed, with only a few patients in each country, clinical trials need to be conducted in many countries in order to enrol the number of patients required to demonstrate benefit/risk. The management of these clinical trials is associated with several issues that slow down the overall process of drug development: administrative hurdles associated with non harmonised regulatory authorisation process as well as cost pressure linked to the need to ensure local activities to several consultants to prepare and manage submission to health authorities and ethic committees.

Today, Europe must streamline its system and all stakeholders should raise their voice to propose specific approaches in order to facilitate the development of new drugs for orphan drugs in a timely manner.

DYSCERNE: developing clinical management guidelines for selected dystrophic syndromes
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The DYSCERNE Network of Centres of Expertise for dystrophomorphology (http://www.dyscerne.org), is developing clinical management guidelines for Williams (WS), Angelman (AS), Noonan (NS) and Kabuki (KS) syndromes. An initial scoping exercise identified these conditions as rare, complex, multi-system disorders, for which it was felt that affected patients, families and health/social care professionals would benefit from access to up-to-date evidence based management guidelines. Published evidence from which to develop management recommendations for these conditions is very limited, and devising a systematic and robust methodology has been challenging. Our approach is based on the Scottish Intercollegiate Guidelines Network (SIGN) method, which we modified placing more emphasis on expert opinion
and consensus, whilst maintaining systematic rigour and transparency of processes.

The development process includes:
- Identification of key management issues by guideline group leaders.
- Targeted, systematic literature searches using PubMed.
- Review, identification and grading of results by panel of invited experts.
- Consensus meetings at which experts present, discuss and agree recommendations.
- Initial drafting of guideline document which is circulated amongst experts and stakeholders for comments.
- Amendments incorporated and guidelines finalised.
- International pilot testing.

The process has involved 49 experts from 8 countries reviewing between them, over 1000 papers. The WS guidelines are currently being piloted in 20 centres, by paediatricians and geneticists. The first draft of AS guidelines has been circulated for feedback. Consensus meetings for NS and KS will be held very shortly. The finished guidelines will be available from the OYSCEEN website.

P21
Wilson France: a national database for Wilson’s disease
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Introduction: Wilson’s disease (WD) is a rare inherited disease with an efficient treatment if initiated early. Improving the knowledge of this disease is a priority of the French national Centre of Expertise for a better access to diagnosis and treatment. This national organisation created a database.

Objective: Improve the knowledge of WD by an epidemiological study on the French database.

Methods: We registered all patients followed by all the French centres working with the national Centre of Expertise.

Results: Since 2006, 281 patients (1-73 year old) were included in the Wilson France database (sex ratio: 1). Mean age at diagnosis was 19 years. First symptoms were neurological for 36% of the patients, hepatic for 38%, renal, psychiatric or hematologic for 11%. Fifteen percent were diagnosed after familial screening. At time of diagnosis, Kayser-Fleischer ring was observed in 95% of patients with neurological symptoms, in 55% of hepatic presentations and in 26% of the presymptomatic forms. Mean coeruloplasminemia was low (0.08 g/L) but 5% of patients had normal values (>0.2 g/L). Mean urinary copper was increased in 96% of the patients. Genetic investigation was not conclusive in 15.9 % of the families (only one or no mutation found). First treatment was D-Penicillamine in 85% of the cases and after a mean follow up of 15 years, the treatment was D-Penicillamine for 44.4% of the patients, Trientine for 14.4%, Zinc for 26.7%, association of chelator and zinc for 5.6 %; 5.6 % of the patients had liver transplantation.

Discussion: The database included approximately 1/3 of the Wilson disease patients in France. In order to improve the recruitment of Wilson’s disease patients, coordination of all health professionals with a multidisciplinary approach is necessary. This work is realised in collaboration with Eurowilson database.

P22
International registry: genetic and phenotypic characteristics of a heterogenous group of disorders
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Severe congenital neutropenia (CN) stands for a group of disorders characterised by extremely low neutrophil counts (ANC < 0.5x109), early stage maturation arrest of myelopoiesis and recurrent bacterial infections. In general more than 90% of CN patients respond to daily G-CSF treatment with a sustained neutrophil increase resulting in significantly reduced infections and an improved quality of life. Besides neutropaenia, the differences in treatment response and the presence of various concomitant clinical features in subpopulations of patients in conjunction with an increased risk of leukaemia transformation in about 10% of all CN patients strongly suggested to search for new sub diagnoses to identify patients at risk of leukaemia.

Within Europe the SCNIR has collected longitudinal clinical data on more than 493 patients with various causes of CN (289 congenital, 64 cyclic, 132 (idiopathic and 8 others) from 22 countries. This unique resource of data was used to identify new genes, classify patients by genetic subtypes of CN, estimate their relative frequency and correlate genetic subtypes with prognosis and outcome.

To date, more than 10 disease causing gene mutations have been identified in congenital neutropaenia patients. In approximately 50-60% of all patients autosomal dominant mutations in the ELANE gene are present. Initial genotype-phenotype correlation identified a group of different genetic defects sharing a high risk of leukaemia transformation in contrast to others with no increased risk of leukaemia.

The identification of new CN subtypes, their distinctive risk of malignant transformation and the response to treatment has contributed substantially to our general understanding of neutropaenia. New risk adapted strategies for diagnosis and treatment have to be implemented in the management of CN patients.

P23
Psychological aspects of living with rare disease: development of psychological skills of rare disease patients. How to improve patients’ quality of life by developing psychological skills necessary to cope with the disease
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Based on the example of psycho-education project for PKU (Phenylketonuria), patients and carers in Pomerania region (Poland), could draw general conclusion: for the improvement of patients well-being, not only medical care, but also psychological knowledge and skills are very important. Some psychological skills are crucial to cope efficiently with the disease and patients and carers should be trained.

The project of psycho-education for PKU patients and carers in Pomerania includes workshops designed especially for 3 target groups:

1. Parents of small children with PKU (age 0-3)
2. Teenagers with PKU (age 14-18)
3. Young women with PKU (age 18-25)

Each of these groups has specific needs concerning psychological skills, which depend on the group’s age and stage of life:

1. Parents of small children: building psychical strength and developing new attitudes.
2. Teenagers: getting ready for independent life.
3. Young women: preparing to be a mother.

To cope efficiently with the disease all patients and carers need both:

a) Basic medical information about the disease and its treatment
b) Basic psychological information about the mechanisms connected with the disease and psychological skills necessary to cope with it.

Most of the support projects usually concentrate on medical aspects of PKU, the treatment and the diet. But for the improvement of patients’ well-being, psychological aspects are also very important. That is why the workshops include both:

- short medical education module
- psycho-education workshop, aiming at delivering basic knowledge and training skills such as self-motivation, self-discipline, self-reliance, resistance to stress, optimism, communication skills etc.
Although there are some psychological skills specific for different groups of patients and careers, the above mentioned skills are quite universal and can be helpful for many rare disease patients and careers.

P24
The patients’ organisations of children with primary immunodeficiency in Poland
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Primary immunodeficiency are rare diseases, approximately 1 per 10 000 children are affected. Since 1980, the Department of Immunology at the Children’s Memorial Health Institute (CMHI) is the reference centre for PIDs for the whole of Poland. Up to now, 1,098 children with PID have been diagnosed. Since 2010, the Department of Immunology CMHI has been nominated as a Jeffrey Modell Foundation Diagnostic Centre. Thanks to the initiative of the Department of Immunology the first patients’ organisation named the Association of Friends for Children with Immunological System Deficiencies was founded in 1987. The original members are patients and their families and friends, doctors, nurses and other medical staff. The Association co-operated with similar organisations such as ESID, IPIOI, INGID up to 1999, when they finished their activity. The other patients’ organisations have been founded, which focused on specific immune diseases (e.g. for DiGeorge syndrome, Nijmegen Breakage Syndrome etc.). These organisations are small and not very active, besides Ataxia-Telangiectasia Foundation which collected large group of patients with AT. In 2007, a new patients’ organisation named IMMUNOPROTECT was set up which gathered parents of patients with primary antibody deficiencies mainly. The main aim of setting up this unit has encompassed the support of children and adults with primary immuno-deficiencies. Basic activity comprises development of an educational programme for patients and families with primary immunodeficiencies and information initiatives for doctors and parents concerning primary immunodeficiency disorders. IMMUNOPROTECT started to put together other small groups of patients as a reasonable way to build up one powerful, effective organisation. These organisations aim at providing better care and access to sufficient treatment.

P25
New functionalities in Orphanet for orphan drugs, R&D and marketing authorisations to better serve the rare diseases community
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The data contained in the Orphanet “Orphan Drugs” database is extracted from official sources. This data includes a list of all substances which have been granted an orphan designation for a disease(s) considered as rare in Europe, whether further developed to become drugs with marketing authorisation (MA) or not. The Orphanet database also includes drugs without an orphan designation as long as they have been granted a MA with a specific indication for a rare disease. Orphanet also publishes a quarterly report (“Orphanet Report Series”) listing orphan drugs on the European market with or without prior orphan designation. In order to improve access to Orphanet’s rich database of information and resources, the search engine has been recalibrated to render data more accessible. In addition to existing search options (by drug or disease), four new sub-tabs improve the visibility of information pertaining to orphan drugs, allowing users to search by a wider range of criteria. Several alphabetical lists of designated products, orphan-designated products with MA, substances and drug trade names are now available. New advanced search options allow users to refine their search by sponsor, MA holder and ATC (Anatomic, Therapeutic, Chemical) category. Substances are now clearly separated from trade names in the results pages: trade names are used solely for products granted MA, whereas substances with orphan designation status (prior to MA) are referred to by their active molecule. Additionally, each substance or trade name is linked to the “Clinical trials” sub-tab of the “Research and trials” tab. Users can retrieve clinical trial(s) that are (or have been) performed for a particular drug. These can also be searched by a wider range of criteria (disease concerned, principal investigator by country, sponsor or clinical trial category). These features are available in all five languages of the Orphanet website.

P26
Evaluation of population newborn screening practices for rare disorders in member states of the European Union
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As a result of a tender launched by the European Commission, an activity has started with the aims of identifying and evaluating all aspects deemed relevant to the implementation of a public health action in newborn screening (NBS), taking into consideration the views of professionals, patients and health authorities. As a result, newborn screening practices and policies will be mapped in the whole European Union and analysed on the basis of current expert methodologies and stakeholders’ views. Within the perspective that NBS is implemented as a public health initiative, a range of aspects will be considered, which ensure the feasibility and sustainability of the screening programme and its efficacy in improving population health, as well as patient care and quality of life. Moreover, challenges and opportunities resulting from NBS implementation will be identified and solutions proposed, accompanied with the information necessary to let national authorities make their own free but informed choices. Finally, the feasibility of supporting actions at the Community level will be explored in order to identify the strategies which the European Commission can adopt to promote the establishment and improvement of NBS programmes in the EU.

The expected deliverables are:

1) Report on the practices of NBS for rare disorders implemented in all the Member States
2) Expert opinion, including a decision-making matrix, on the development of European policies in the field of newborn screening for rare diseases.
3) A European Union Network of Experts on Newborn Screening (EUNENBS)
4) European Experts Consensus Workshop on Newborn Screening

At the European Conference on Rare Diseases, the questionnaire set up to collect information on NBS in the EU and the criteria for inclusion of experts in EUNENBS will be presented.

P27
APTC: a social network to improve the quality of life of members of patients’ associations
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The APTIC social network has been developed by PSINET research group at the Universitat Oberta de Catalunya together with Hospital de St. Joan de Déu and Fesalud Foundation, and has been partially funded by Tic Salut Foundation (health department of the Catalan government). Our social network aims at enabling the individual members of patients’
associations (mostly parents of children with chronic and rare diseases) to share experiences, information, advice and, ultimately, to offer them online tools to improve the quality of life of the entire family. Participation in specialised social networks like APTIC offers a great opportunity to use technology to improve quality of life with low cost and with a large impact on health. From the health psychology perspective, variables such as the perception of self-efficacy, empowerment and social support are key to improving quality of life. For this reason, like health psychologists we are interested in the analysis of these variables and in their impact on the quality of life of members of patients’ associations. We are examining these variables and network usage in order to establish whether social networks are indeed useful for parents.

Our social network has been built on open source software and through collaborative work between organisations (the hospital itself, patients’ associations...) and individual users.

We will offer our first results and some thoughts about the work with patients and families through social networks. Our goal is to enhance their positive effects, for instance using them as a tool to overcome the fragmentation of knowledge in the field of rare diseases.

**P28**

**Issues of management of Epidermolysis bullosa in Georgia**

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As of today there are no radical methods of treatment of epidermolysis bullosa (EB) as well as other genetically determined diseases. This accounts for the acuteness of the problem of providing adequate care for such patients.

The Georgian Foundation for Genetic and Rare Diseases (GeRaD), a self-help organisation of people personally affected by rare diseases in Georgia, observed 3 patients, age 1.5, 2, and 3.5 years. The first 2 patients were diagnosed with dystrophic polydysplastic form of EB, while the third one had a simple generalised epydermolytic form. All patients were received in various clinics and with various level of success some pathogenetic (steroids, erythromycin, tocopherol acetate, retinoids), symptomatic, and local treatment. All of them had reached periods of remission of various lengths. It should be noted that the longest remission was achieved, contrary to the opinion of many authoritative dermatologists, as a result of treatment with Phenytoin. The families of the patients, despite notable therapeutic success, faced care and nutrition related problems caused by the lack of information about the disease that affected their children. We tried to solve these problems by addressing the Institute of Dermatology and Venereology. During 6 months in the frame of the activities of the nursing for patients with severe dermatoses the doctors and nurses of the Institute provided free treatment for our patients and information for their parents about actual issues of living with EB (care, nutrition, prevention of opportunistic infections, etc.). According to the parents’ evaluations, the quality of life of their children significantly improved due to our activities. This example of successful collaboration between a State medical institution and an NGO in the sphere of providing care for patients with rare diseases may serve as one of the preconditions for creation of the optimal model of management of rare diseases in Georgia.

Cite abstracts in this supplement using the relevant abstract number, e.g.: Kvlividze et al. *Issues of management of Epidermolysis bullosa in Georgia*. *Orphanet Journal of Rare Diseases* 2010, 5(Suppl 1):P28