ICORD

International Conference on
Rare Diseases & Orphan Drugs

CONFERENCE REPORT

ICORD 2014 - the IXth Annual Conference

"The Societal Value of Prevention, Diagnosis and Treatment of Rare Diseases"

7-9 October 2014, Ede, The Netherlands

Report compiled by Annika Larsson, ICORD Secretariat

Conference Summary

The IXth ICORD meeting was held 7-9 October 2014, in the ReeHorst Hotel, Ede, The Netherlands. The conference was locally supported by ZonMw and took place in conjunction with the FIGON Dutch Medicine Days (FIGON-DMD), which had around 600 participants. ICORD wishes to express its sincere gratitude to ZonMw for inviting ICORD to the FIGON-DMD and to the local organising committee for organising this meeting together with ICORD.

The conference was attended by participants representing many different stakeholders, such as patient organisations, researchers, clinicians, industry and governmental authorities from 23 different countries in Asia-Pacific, Europe, Latin America and North America. The conference programme included sessions with invited speakers and authors from a selection of submitted abstracts from many countries and continents, all devoted to the field of rare diseases and orphan drugs, as well as joint sessions with representatives from different institutions in The Netherlands. The programme encompassed ten plenary sessions with 36 oral presentations, a poster exhibition (46 academic and institutional posters) and a working group session. All approved abstracts have been published in the journal Rare Diseases and Orphan Drugs (Vol 1, No 3, 2014).

During the meeting in Ede the ICORD General Assembly was held and the new ICORD board was elected. Unlike other organisations in the rare disease community, ICORD is unique in that it is a society for all rare disease stakeholders from all over the world. The new ICORD board represents various stakeholders from Asia, Europe, North America, Oceania and South America. After two successful years Virginia Llera (Geiser, Argentina) handed over the Presidency to John Forman (NZORD, New Zealand). One objective of ICORD is to build and provide bridges between all rare disease stakeholders as it is ICORD's vision that the rare diseases challenges can only be conquered by joint efforts of all parties. The ICORD annual meeting in Ede was an important event to achieve this objective.

The IXth ICORD meeting in The Netherlands made it evident that a lot of hard work is currently ongoing to improve the lives of people with rare diseases around the globe. However, the work takes time and still access to diagnostics and treatments are limited. New working models and structures are needed in order to progress faster. During the conference there were several presentations on novel approaches trying to overcome the many obstacles in the rare disease field, mainly by including all stakeholders in collaborations. A constant topic in rare disease meetings is data sharing and this was also discussed in several sessions of this meeting, including presentations introducing new platforms of data sharing. The attendees got an interesting insight into the Dutch rare disease status, the current debate and the challenges for the future in the Netherlands, in Europe and elsewhere. Patients and patient-advocacy organisations do immense work for the benefit of rare disease patients and great examples of this were presented along with other inspiring topics and sessions.

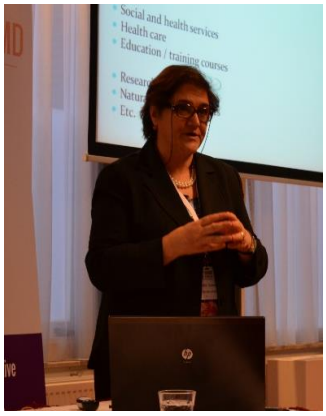
ICORD was delighted to welcome representatives of the local organising committee of the ICORD 2015 to the Netherlands. The Xth annual ICORD meeting will be celebrated in Mexico in October 2015 and delegates in Ede included representatives from FEMEXER (the Mexican umbrella organisation for rare disease patients), the Institute of health security in Mexico (ISSSTE) and the Federal Commission for the Protection against Sanitary Risks in Mexico (COFEPRIS). In 2010 the VIth annual ICORD conference took place in Buenos Aires, Argentina. After ICORD's presence in the country and involvement with the ministry of health, the Argentinean regulation has been changed for the benefit of rare disease patients. As a continuum of the work started in 2010 ICORD will now return to Latin America. ICORD looks forward to a fruitful 2015 Meeting, with the overall aim to contribute to an improved situation for rare diseases patients in Latin America but also world-wide. ICORD warmly welcomes you to Mexico City 15-17 October 2015!

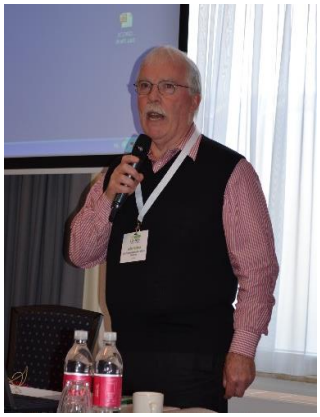
Content

Opening

ICORD's capabilities, position and challenges

The IXth ICORD annual meeting was officially inaugurated by opening remarks by Virginia Llera (ICORD President), Domenica Taruscio (ICORD Past-President) and John Forman (ICORD President-Elect). During their presentations they gave the audience an historical perspective of ICORD and explained its role and significance in the rare disease field. ICORD is a society for all individuals active in the field of rare diseases and orphan drugs around the globe with the ultimate aim of improving the welfare of patients with rare diseases and their families world-wide through better knowledge, research, care, information, education and awareness. ICORD's position statement, also called the Yukiwariso Declaration, was published in Acta Paediatrica 2012 (Aug; 101(8): 805-7) and can be found on the ICORD website (www.icord.se). ICORD is an inclusive society where all stakeholders participate on an equal level independent of origin. It functions as a bridge, by connecting experts and by linking countries and regions, and empowers local initiatives, also initiatives from regions in the less developed world. ICORD stimulates networking and sharing of best-practices, something that is essential in the world of rare diseases where knowledge and resources are scarce and scattered. Issues for rare diseases go well beyond drug discovery, regulation and reimbursement. They include primary prevention, screening programmes, early and accurate diagnosis, best clinical care, and social and educational support. Rare diseases need a womb-to-tomb approach. ICORD's challenge for the future is to bring the agenda and people together to make real progress on these issues.

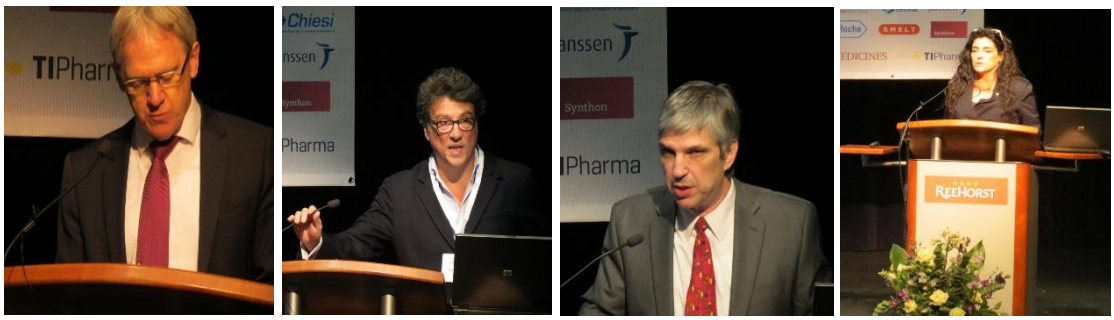





Domenica Taruscio (ICORD Past-President, Italy), John Forman (ICORD President-Elect, New Zealand) and Virginia Llera (ICORD President, Argentina) during the conference in Ede.

Opening plenary lectures

The opening session, moderated by the local organisers Sonja van Weely (ZonMw, The Netherlands) and Remco de Vrueh (TI Pharma, The Netherlands), was continued by plenary lectures with the overall title "Main steps for recognising the societal value of diagnosis, prevention and treatment of rare diseases". A Dutch national perspective was given by Fred Krapels (Dutch Ministry of Health, The Netherlands), broad European actions driven by EURORDIS was addressed by Yann Le Cam (EURORDIS, France) and the national initiatives and regulatory actions from the leading Latin American country Mexico were exposed by Mario Alanis Garza (COFEPRIS, Mexico). Virginia Llera concluded the session by explaining the methodologies of ICORD and the process used to promote cultural changes into different regions and countries in order to overcome rare disease challenges and drive the development forward.



Fred Krapels (Dutch Ministry of Health, The Netherlands), Yann Le Cam (EURORDIS, France), Mario Alanis Garza (COFEPRIS, Mexico) and Virginia Llera (ICORD President, Argentina) discussed different perspectives in the opening session on "Main steps for recognising the societal value of diagnosis, prevention and treatment of rare diseases".

New cooperative approaches on the way

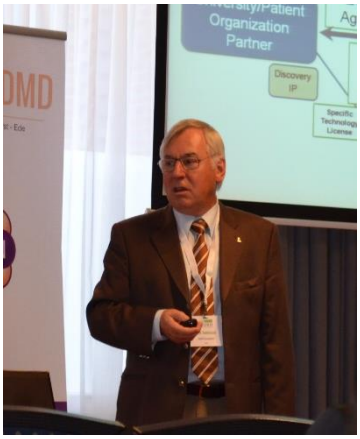
Despite the avalanche of discoveries and innovations in medicine in the last decades many rare disease patients still lack diagnosis and treatment. In order to progress faster there is a need for new working models and approaches to overcome the challenges in the rare disease field. Several speakers with different background (patient organisation, academia and industry) in different sessions (session III, V, VIII, IX and X) introduced new methodologies to move forward.

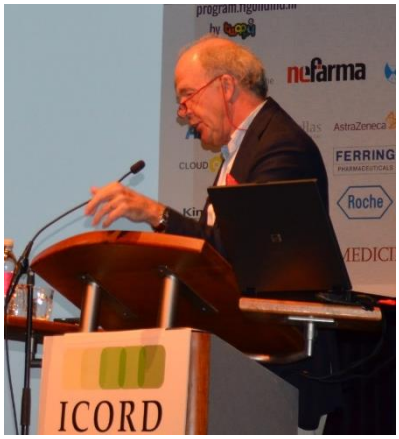
The Biopontis Alliance for Rare Diseases (BARD), presented by Erik Tambuyzer (Belgium), is a newly started public-private-philanthropic partnership network with the aim of speeding up the development of new therapies for rare diseases. None of the today's players in the rare diseases field, e.g. the industry, academia or regulators, have all necessary knowledge and therefore a multidisciplinary collaboration is needed to minimize the work and the cost of orphan drug development. The BARD presents a new model including scientific and economic partnerships, where the gap between basic research and clinical trial is bridged. BARD consists of a network of scientific, technical, legal, regulatory, commercial and negotiation experts. The network is currently looking for projects, partners and funds across the globe to find treatments for diseases where there currently is no therapy.

Sharon Terry (Genetic Alliance, USA) described the EspeRare Foundation, which uses a similar approach as BARD by including several stakeholders in the drug development process. The EspeRare Foundation makes patient groups, academia, medical reference centres and industry work together to uncover the potential of existing drugs to address severe therapeutic unmet needs in neurological and immunological rare diseases (so-called repurposing of existing drugs). By using already existing drugs the research costs and the time to access of treatment can be decreased significantly.

Prices of orphan drugs are generally very high and stay high even after patents expire. With the increased development of personalised medicines a new approach is needed to contain future costs. Hans Büller (ErasmusMC, The Netherlands) presented the "fair medicine approach" for orphan drug development. Fair medicine involves trust, a shared responsibility and shared financing. From day one patients, doctors, researchers, investors and companies need to be involved in the drug development process. The approach includes an increased risk for all stakeholders but also a shared financial benefit. The primary focus should be societal need and not shareholder value. With this approach transparency will increase and result in acceptable profit margins. Today there is no transparency of prices, information which is crucial for performance of proper health technology assessments.

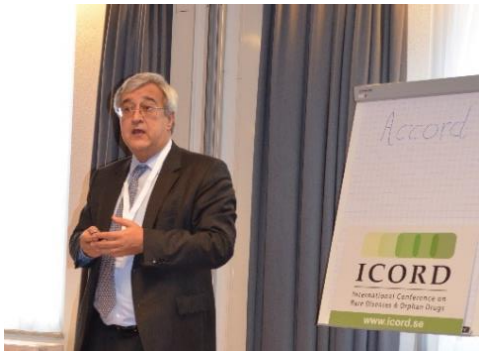
Also the industry has changed its way of thinking and is developing new working models. Barbara Wuebbles (Audentes Therapeutics, USA) has a long experience from work in clinical trials for the industry. She described the benefits of including patients already in the planning of research projects and how true partnership has evolved between industry and patient groups the last years. She still sees a big need of other groups, such as academia and regulatory expertise, to join the partnership in order to proceed faster.





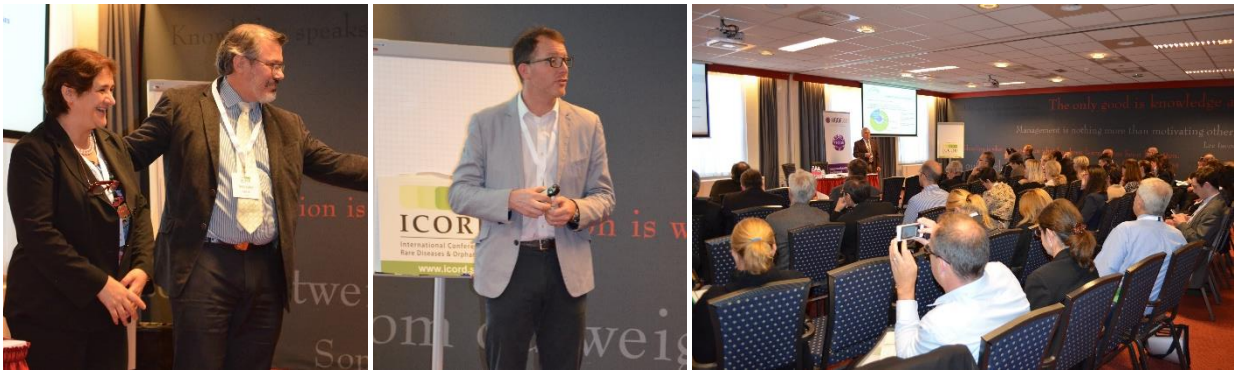
Erik Tambuyzer (BARD, Belgium), Barbara Wuebbels (Audentes Therapeutics, USA) and Hans Büller (ErasmusMC, The Netherlands) all described the necessity to involve all stakeholders in the development of new treatments for rare diseases.

Fernando Royo (Fundación Genzyme, Spain) described how the cooperation of a biopharmaceutical company with other rare diseases' stakeholders can be developed to include other initiatives beyond provision of orphan drugs. Fundación Genzyme is a Spanish non-for-profit entity which focus its activities on research support, continued medical education, patient empowerment & social awareness programmes.



Paul Ledger (GSK, United Kingdom) presented novel strategies at GSK to adapt to the rare disease challenges, especially in the less developed world. To improve access, GSK has employed a novel funding model with the use of a flexible pricing approach based on a country's wealth and ability to pay. With this model GSK has succeeded to introduce orphan drugs to the least developed countries. "Adaptive Licensing" is another approach that GSK is engaged in to find treatments for amyloidoses (a group of protein mis-folding disorders). The Adapting Licensing Initiative is a pilot programme launched early 2014 by the European Medicine Agency (EMA) with the aim to improve timely access for patients to new medicines by issue of an initial license earlier in the development process and by including all stakeholders with a role in determining patient access, such as EMA, industry, health

technology assessment bodies, associations developing clinical treatment guidelines and patient organisations. Also during session VII, Orphan drugs and Personalised medicine, Marjon Pasmooij (Dutch Medicines Evaluation Board) informed about adaptive licensing.



Domenica Taruscio (ISS, Italy), Emilio Roldàn (SLADIMER, Argentina, session moderator), Paul Ledger (GSK, UK) and Erik Tambuyzer (BARD, Belgium) during the session V on International Collaboration.

Research is crucial in order to drive the development of diagnostics and therapies further. However, in the field of rare diseases, where patients are few and the knowledge is scarce, executing a successful research project is almost impossible without collaboration. The International Rare Diseases Research Consortium (IRDiRC) is a recent example showing that joint international research funding strategies and research cooperation are needed to proceed faster. Ségolène Aymé (IRDiRC, France) explained IRDiRC, its structure and its ambitious aim of developing 200 new treatments and means to diagnose most rare diseases by 2020. IRDiRC was an initiative by the National Institute of Health in the USA and the European Commission, but the consortium has now expanded to include many organisations in the USA, Europe, Asia and Australia as well. These include patient organisations, foundations, research funding organisations and industry. IRDiRC has three scientific committees; the diagnostics committee, the interdisciplinary committee and the therapies committee. The Therapies Scientific Committee (TSC) was presented by Gert-Jan van Ommen (Leiden University, The Netherlands). TSC has set up recommendations for IRDiRC's funding organisations including criteria and priorities for funding of clinical and non-clinical projects in order to reach IRDiRC's aim of 200 new therapies in 2020.

Gert-Jan van Ommen (Leiden University, the Netherlands), Ségolène Aymé (INSERM, France), Sonja van Weely (ZonMw, The Netherlands) and Anthony Brookes (University of Leicester, UK) just after the session X about IRDiRC, see text for details (picture kindly provided by Ségolène Aymé).

Sharing of data, patient registers and biobanks

Rare diseases patients are few and therefore data sharing is particularly needed in the field of rare diseases. This message was repeated many times during the conference in Ede. However, sharing of data is difficult when different terminologies are used. Ségolène Aymé (INSERM, France) presented in session X the immense work that she and the Orphanet team have been involved in, trying to align the various terminologies used, such as ICD, MeSH, SnoMED CT and MedRA. In order to transform all terminologies into one common terminology and ontology (representation of knowledge understandable by computers) the International Consortium for Human Phenotype Terminologies (ICHPT) was formed and it has now adopted a core set of more than 2300 terms common to all terminologies. In January of 2015 the ICHPT website (http://ichpt.org) will open and the conference participants were urged to promote this new terminology for the benefit of rare disease patients. The WHO will use this terminology for the ICD-11.

In the EU there are more than 600 rare disease registries. However, registries are only set up for a small fraction of the diseases and there are multiple registries for the same disease. Furthermore, there is no uniform standard for storing and managing the data which makes data sharing and comparability challenging. Domenica Taruscio (ISS, Italy) presented in session IX the EPIRARE project, which aims to build consensus and synergies for registration of rare disease patients in the European Union. Examples of a national rare disease registry was given by Manuel Posada (Institute of Rare Diseases Research, Spain) who coordinates the Spanish Rare Diseases Registries Research Network, which is based on the interaction of two methods, i.e. population-based registries and patient research outcomes registries. The ultimate objective of the Spanish registry is to improve prevention, diagnosis, prognosis (at different levels), treatment and quality of life for patients and their families using high quality information. The registry is connected to a national rare disease biobank (BioNER), which was described by Veronica Alonso (Institute of Rare Diseases Research, Spain). The biobank holds biological samples (DNA, plasma, serum and cells) for 120 different rare diseases. This biobank is part of the Eurobiobank, which is the first European consortium of biobanks for rare diseases and provides access to 440,000 biological patient samples. In the discussion the difficult items for registries like privacy of the patient, sharing of data and sustainability of registries were mentioned.

New approaches of data sharing were presented in session X by Anthony Brookes (University of Leicester, United Kingdom) who is chair of the IRDiRC Working Group on Genome/Phenome and member of the working group of Data Sharing and Bioinformatics. He has been involved in developing a platform, Café Variome (http://www.cafevariome.org/), which shares the “existence” of data rather than “substance” of data, which might be sensitive. Once the existence of data have been discovered it is up to the owner of the data to decide if that data should be shared or not. Another method to accelerate data collection and sharing is the Community-Engaged Network for All (CENA) project initiated by Genetic Alliance. Sharon Terry presented in session XI the network, which has nine disease-specific advocacy organisations (DAO) and academic partners. Online registers have been customised for each condition represented by the DAOs using a Platform for Engaging Everyone Responsibility (PEER), developed by Genetic Alliance. The PEER is highly intuitive, uses a game-like interface with survey questions and each participant decides with whom and for what purposes his or her data may be used (dynamic consent).

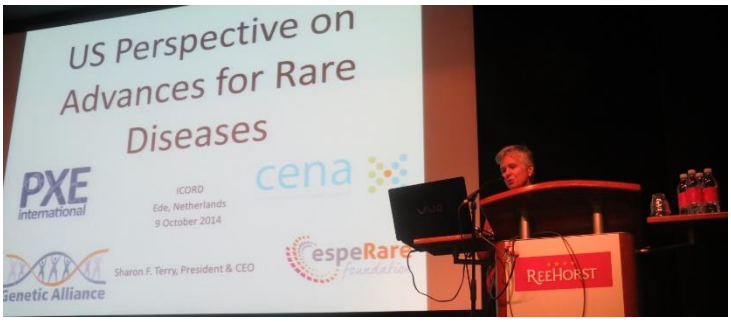
Patients’ impact

Although rare disease patients are few, patients can make a difference and together they have proven strong and very influential, especially in the field of rare diseases. Yann Le Cam (EURORDIS,

France) gave a presentation in the opening plenary lecture on EURORDIS, the European umbrella patient organisation for rare diseases. It was formed in 1997 and now has 633 patient organisations as members from 59 countries (including 26 EU countries), representing about 30 million people and over 4000 rare diseases. EURORDIS is involved in many of the rare diseases activities taking place in Europe and elsewhere. Major advocacy achievements include patient participation in COMP from the beginning (EMA's Committee for Orphan Medicinal Products) and nowadays also in other committees of the European Medicines Agency (EMA), the Council Recommendation on an Action in the Field of Rare Diseases (8 June, 2009), the EU Directive on Patients' Rights in Cross-border Healthcare and recently the Revision of the EU Directive on Clinical Trials. EURORDIS is very much involved in assisting European countries in shaping national plans for rare diseases and is member of the Commission Expert Group on Rare Diseases (former EUCERD). A recent initiative is Rare Diseases International, which aims to create a global alliance representing patients and families of all nationalities across all rare diseases and across the world.

Sharon Terry (Genetic Alliance, USA) has done impressive work, as mentioned above. Her "career" in the rare diseases field started when she became a mother of two children with pseudoxanthoma elasticum (PXE). Although being laypersons, she and her husband initiated and got involved in activities normally performed by medical researchers, such as searching for the gene involved (and found it), setting up a PXE biobank, developing a diagnostic test for the disease and initiating a clinical trial.

Another example of the important role that patients may have in improving the situation of people with a rare disease was given by Cees Smit (Dutch Genetic Alliance, The Netherlands), who is a hemophilia patient. One of his policy activities as patient representative (amongst many others) is the membership of the Appraisal Committee of the National Health Insurance Board (ZINL) that advises ZINL on the content of the legally insured care package from societal point of view (e.g. reimbursement of medicines, medical devices, etc.). The decisions of the Minister of Health are based on the advices of ZINL.



Sharon Terry (Genetic Alliance, USA) during the session during session XI, "Patient perspectives on the societal value of rare diseases and orphan drugs".

Prevention and education

One important objective of the ICORD annual meetings is to spread knowledge and awareness of rare diseases as well as sharing of best-practices.

Session VI dealt with prevention of rare diseases. Amanda Julie Neville (Universitaria di Ferrara, Italy) described EUROCAT, a European network for epidemiologic surveillance of congenital anomalies. EUROCAT currently includes 1.7 million births or 31% of the yearly births in Europe. Many of the congenital anomalies are preventable by simple measures, such as folic acid supplementation or fortification of food to avoid neural tube defects (NTDs). Critics of fortification programmes claim

that further studies should be undertaken before widespread fortification of food occurs, arguing instead for supplementation use by individuals, because of concerns of possible adverse side effects, such as cancer. However, after analysing multiple folic acid studies Derrick Bennett (University of Oxford, United Kingdom) and his colleagues could not verify a correlation between folic acid supplementation and any presumed problems. A further presentation planned to be given by Stefania Ruggeri (University of Rome, Italy) was reported on her behalf by Amanda Julie Neville with emphasis on registry figures showing that supplementation policies had failed to have any significant impact on the incidence of neural tube defects, and offering a challenge to all, regarding the best way to reduce these cases. Lieven Bauwens (International Federation for Spina Bifida and Hydrocephalus, IFSBH, Belgium) explained the mission of his organisation: reduction of the incidence of both neural tube defects by primary prevention, and improvement of the quality of life of people with SBH and their families through human rights education, political advocacy, research and community building. The IFSBH started in 1979 and is a global umbrella organisation of 52 national/regional members in 49 countries. John Forman (NZORD, New Zealand) gave an overview of the societal benefits, in terms of economic impact and decreased suffering, of folic acid fortification programmes. Despite the proven decrease in neural tube defects, countries around the globe have been restrictive about implementing fortification programmes. He called for more consideration of the ethical responsibilities of decision-makers when faced with firm evidence of benefits but very weak data about possible harms. Public health decision-making frameworks should be consciously considered as the major guide to decisions regarding folic acid.

Education is one important factor in prevention. However, education is scarce and scattered in the field of rare diseases. To increase the knowledge and in order to increase the capacity of public health intervention Domenica Taruscio (Istituto Superiore di Sanità, Italy) has initiated an International School of Public Health for Rare Diseases (http://www.iss.it/cnmr/news/index.php?lang=2&tipo=41). The courses are tailored training courses for all stakeholders and could address many topics such as epidemiology, prevention, genetic counselling, health care guidelines, diagnostics etc. The courses are endorsed by ICORD and are also provided through internet for those participants that cannot travel to Italy.

Orphan Drugs and Personalised Medicines

Individualised treatment of common diseases, e.g. for some cancers, is imminent. The discussions on the so-called personalised medicines have similarities with the discussions on orphan drugs for rare diseases, like how many patients have to be included in clinical trials to show statistically significant results, costs of development of treatment for a low number of individuals etc. Session VII, organised together with the Dutch Clinical Trial Foundation and moderated by Manuel Posada (Institute of Rare Diseases Research, Spain) on behalf of ICORD, focused on orphan drugs and personalised medicines. Angela Brand (Maastricht University, The Netherlands) gave a background to the development of personalised medicine and the current status and initiatives in Europe. She encourages collaboration between actors in the rare diseases and personalised medicine fields in order to proceed faster and learn from each other. Marjon Pasmooij (Dutch Medicines Evaluation Board, MEB) explained the MEB strategy plan, which is centred around 1) patient-oriented evaluation of medicinal products, 2) promoting the proper use of medicinal products and 3) promoting the innovation of medicinal products. Two innovative approaches to increase the number of treatments on the market were presented, “n-of-1 trials” and “adaptive licensing”. In n-of-1 trials a single patient is the entire trial. In this single-case study a patient is followed before and during treatment and in this way the patient is its own reference. Methodology considerations include the heterogeneity of the effect seen between

patients, how the treatment effect could be generalised to the population intended and the importance of patient registries for future patients. The goal of adaptive licensing is to get effective medicines to patients as soon as possible. The approach will be piloted for medical conditions for which there is a high unmet medical need. After an initial license is given patient data are collected and evaluated before a full licence is given. During normal procedure treatment experience does not contribute to evidence generation, which means that more data need to be presented before a licence can be issued. Adaptive licensing also includes bringing all stakeholders together early in the process to face the different challenges as a team. Paul Ledger indicated in session V that GSK is involved in a pilot project of adaptive licensing.

Access to treatments in the Netherlands

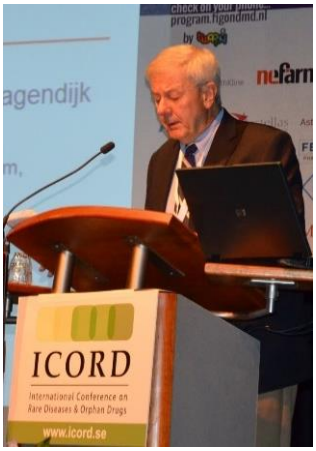
The conference delegates were offered an insight into the state of rare diseases of the host country, The Netherlands. Fred Krapels from the Dutch Ministry of Health provided a summary of the Dutch priorities and activities within the rare disease field. He stressed the importance of sharing expertise and skills on the diagnosis and treatment of rare diseases and said that the ICORD conferences strongly contribute to this aim by gathering many different stakeholders from all over the world.

A topic that has caused a lot of controversy in the country the last years was the leak of a preliminary advice of the advisory body ZINL to the Dutch Ministry of Health in 2012 to stop reimbursement of enzyme replacement therapy for Fabry and Pompe diseases in the Netherlands with the argument of low cost-effectiveness/low efficacy. This gave rise to an intensive public debate and many questions were asked concerning the value of life, transparency on the high price of orphan drugs, the need for cost-effectiveness studies on orphan drugs and the responsibility for the development of these treatments (industry or government). In the end the ministry decided to continue the reimbursement of the two diseases, at least for another 2-3 year period and started discussions with the pharmaceutical companies on price cuts. For Fabry disease there are two treatments available from two different companies. The cost is approximately 200 000 EUR per patient and year. Currently both companies have developed their own Fabry registry but there is no collaboration between them. Carla Hollak (Academic Medical Center, The Netherlands) requested more cooperation as the number of Fabry disease patients is too low to work individually. In order to evaluate the drugs properly and to move forward data sharing is essential.

Leona Hakkaart (Erasmus University Rotterdam/Institute for Medical Technology Assessment, The Netherlands) explained more details on the assessment for reimbursement of (orphan) drugs in the Netherlands. Several factors play a role in any reimbursement decision: effectiveness of the drug, cost-effectiveness, necessity and feasibility. For orphan drugs, other factors might be included, or might replace ‘common’ factors, such as rarity, target population and equity/solidarity. She showed that reimbursement of orphan drugs is often a black box with differences both within and between countries.

A political perspective was given by Rob Hagendijk (University of Amsterdam, The Netherlands). He is studying the politics of rare diseases and orphan drugs. In the Netherlands and Europe there is a fear of increasing orphan drug costs as they may be unsustainable. However, rare diseases is an area where Europeans see an added value of collaboration, even those that normally are against the EU. After analysing the Pompe and Fabry controversy he believes that short term pragmatism is needed in treatments of rare diseases but should be associated with discussions on adjustment of institutional frameworks in European and national health care and research.

Cees Smit (Dutch Genetic Alliance, The Netherlands) described in the session (session XI) on patient's impact the hard but successful work of Dutch patients to influence advisory bodies to continue the reimbursement of orphan drugs for Fabry and Pompe disease patients.



Stephen Groft (NIH, USA) moderated session III where Rob Hagendijk (University of Amsterdam, The Netherlands) and Carla Hollak (Academic Medical Center, The Netherlands), among others, gave perspectives on the Fabry and Pompe disease controversy in the Netherlands.

ICORD 2015 in Mexico

ICORD will celebrate its Xth annual meeting in Mexico City, Mexico, in the week of October 12-16, 2015. ICORD was honoured to have the local organising committee attending Ede, including representatives from FEMEXER (the Mexican umbrella organisation for rare disease patients) and the Institute of health security in Mexico (ISSSTE). The Federal Commission for the Protection against Sanitary Risks in Mexico (COFEPRIS) supports the organisation of ICORD 2015 in Mexico. Mario Alanís Garza, representing COFEPRIS, presented the COFEPRIS and the regulatory actions taken to improve access to medicines and health. Before 2010 Mexico did not officially recognise the existence of orphan drugs and consequently no authorization was given prior to that year. A new regulation is currently under discussion, but since 2010 forty-eight authorizations for orphan drugs have been granted.

Conclusions of the working groups

The ICORD working groups offer an opportunity for the conference delegates to meet and discuss matters of importance with other delegates with similar interests. The aim of the working groups is to improve the international collaboration and to find topics that could be worked on between the annual meetings. But above all, the session provides a forum where conference attendees can network, learn from each other and bring new ideas back home. The working groups are open to everyone and their contents are decided by the participating delegates. Each working group has a coordinator who functions as a facilitator to drive the discussion and continued work of the group.

Policies and citizen perspectives around the world: working together for ICORD 2015

The working group on "Policies and citizen perspectives around the world: working together for ICORD 2015" was coordinated by Virgina Llera (GEISER, Argentina) and Sharon Terry (Genetic Alliance, USA). Topics put on the table included improved access to information, provision of transparent educational tools and different aspects of the societal impact and human cost of rare diseases. ICORD is a global society and despite the big discrepancies that exist between different

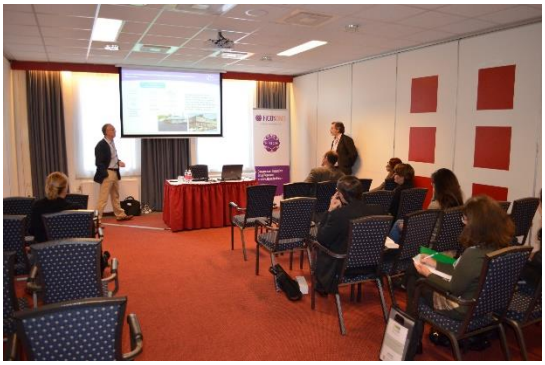
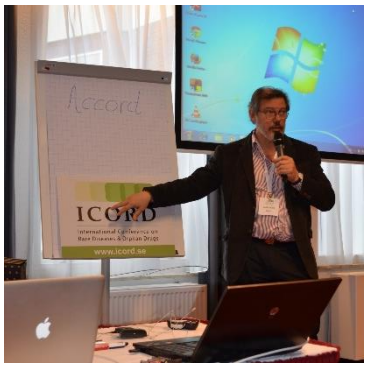
geographical regions and cultures it was stressed that strategies of ICORD must include a global dimension. Also, during strategy development (of ICORD but also of other rare diseases initiatives) the real life situation of rare disease patients should always be assessed. To ensure that the initiatives taken really improve the quality of life patients should be the core of all rare diseases actions. Access to education is one of the basic elements required for the rare disease community to reach similar opportunities globally. Virginia Llera volunteered to develop an educational project for this purpose. ICORD's position statement, the Yukiwariso Declaration, was considered important. However, it needs increased promotion and distribution to a wider audience.



Sharon Terry (Genetic Alliance, USA) and Virginia Llera (GEISER, Argentina) coordinated the "Working Group of Policies and citizen perspectives around the world: working together for ICORD 2015".

Research working group

Maja Stojiljkovic (Belgrade University, Serbia) and Emilio Roldàn (SLADIMER, Argentina) coordinated the Research working group. An example of an integrative model of rare diseases management in a tertiary hospital was presented by Joan Comella and Eduardo Tizzano from the Vall d'Hebron University Hospital, Spain (Rare J. 2014, 1(3):138). Furthermore, the potential of ICORD as a main bridging tool for researchers worldwide was discussed and how that could be partly achieved through the development of a specialised rare disease research website. The next activity of the ICORD research working group will be the arrangement of the "1st Accord with Academy – Translational Research in Rare Skeletal Diseases" at Leiden University, The Netherlands, in 2015. The objective of the one-day meeting is to understand the needs of rare disease researchers, to examine points of mutual interests and to develop collaborations. The outcome of the meeting will support ICORD in its priorities for future activities.

Eduardo Tizzano (Vall d'Hebron University Hospital, Spain), left picture, gave a presentation during the "Research Working Group", coordinated by Emilio Roldàn (SLADIMER, Argentina), right picture.

Poster session

The conference poster session had 46 outstanding posters, 29 of those were academic posters and 17 institutional posters.

During the conference the best poster award was given to the CRESim group at Lyon University Hospital in France. The title of the poster was “Mathematical model of T-cell lymphoblastic lymphoma: disease, treatment, cure or relapse of a virtual cohort of patients” by Nathalie Eymard *et al*. The CRESim (Child-Rare-Euro-Simulation) project has the objective to develop “in-silico” randomized controlled clinical trials in order to choose the most appropriate clinical trial for real patients, thereby decreasing the number of patients in clinical trials and increasing the pace of orphan drug development. Their study, using mathematical modelling, suggests that treatment duration of T-cell lymphoblastic lymphoma in children can be shortened without increasing the number of relapses.

About the organisers

ICORD

International Conference for Rare Diseases and Orphan Drugs (ICORD) is an international society for all individuals active in rare diseases and/or orphan drugs, including health care, research, academic, industry, patient organisations, regulatory authorities, health authorities, and public policy professionals. ICORD's mission is to improve the welfare of patients with rare diseases and their families world-wide through better knowledge, research, care, information, education and awareness. ICORD's position statement, also called the Yukiwariso Declaration, was published in Acta Paediatrica 2012 (Aug; 101(8): 805-7). One major objective of ICORD is the organisation of the ICORD annual meetings, where everyone within the rare disease field can meet, network and exchange ideas and best practices with other stakeholders around the globe. For more information on ICORD see www.icord.se.

ZonMw and FIGON

ZonMw, The Netherlands Organisation for Health Research and Development (www.zonmw.nl/en/), funds health research and stimulates the actual use of the knowledge developed to help improve health and healthcare in the Netherlands. ZonMw's main commissioning organisations are the Ministry of Health, Welfare and Sport and The Netherlands Organisation for Scientific Research. ZonMw collaborates with many organisations on national and international level. ZonMw is one of the partners of the platform FIGON, The Netherlands Federation for Innovative Drug Research (www.figon.nl), which consists of several Dutch parties involved in drug research, such as scientific and professional associations, universities, business and government. FIGON enhances existing initiatives in the area of drug research and signalizes new developments in this area. An important activity of FIGON is its annual meeting each October, the so-called FIGON Dutch Medicines Days, to which ICORD was invited for its IXth annual meeting in 2014.

Conference Organisation

Strategic and Planning Committee

Conference Chair

Virginia A. Llera, MD (ICORD President, GEISER Foundation Latin America president, Argentina)

Members

Dr. Domenica Taruscio (ICORD Past-President, Director National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy)
John Forman (ICORD President-Elect, President NZORD, New Zealand)
Dr. Manuel Posada (ICORD Secretary, Director Institute of Rare Disease Research (IIER), ISCIII, Spain)
Marlene Haffner (ICORD Treasurer, MD, President and CEO Haffner Associates LLC, Founder Orphan Solutions, Former FDA Director Orphan Products Development, USA)
Dr. Simon Day (ICORD Board Member, Director of Clinical Trials Consulting & Training Limited, UK)
Dr. Stephen Groft (ICORD Board Member and former ICORD President, former Director at NIH Office of Rare Diseases Research, USA)
Prof. Jan-Inge Henter (ICORD Board Member and Founder, MD, PhD; Director of Research and Education at Karolinska University Hospital, Sweden)
Dr. Yukiko Nishimura (ICORD Board Member, Department of Intellectual Property and Social Application of Technology, University of Tokyo, Japan)
Dr. Rumen Stefanov (ICORD Board Member, Head, Department of Social Medicine and Public Health, Medical University of Plovdiv, Bulgaria)
Dr. Désirée Gavhed (ICORD Member, PhD, Karolinska Institute, Karolinska University Hospital, Sweden)
Dr. Annika Larsson (ICORD Member, PhD, Karolinska University Hospital, Karolinska Institute, Sweden)

Advisors

Dr. Emilio Roldán (ICORD Member; MD, PhD; Scientific Director GADOR S.A, Argentina)
Georgi Iskrov (MBA, Head of HTA Unit Institute for Rare Diseases, Bulgaria)

Local Organising Committee

Dr. Sonja van Weely (ICORD Member; Scientific officer at The Netherlands Organisation for Health Research and Development ZonMw, The Netherlands)
Dr. Remco de Vrueh (Programme Manager at Top Institute Pharma & Founder Rare Disease Matters, The Netherlands)
Gijs Nyst (Project Manager TCMan, The Netherlands)

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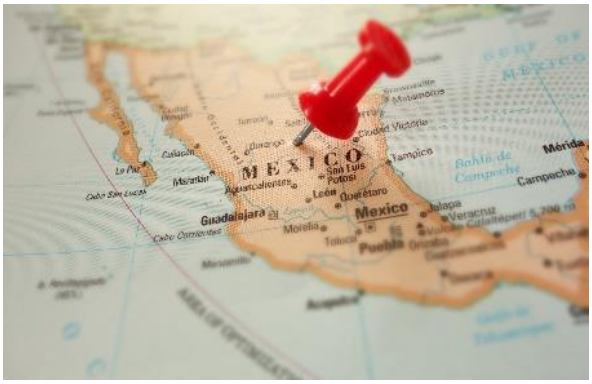
ICORD 2015 Annual Conference

The Xth ICORD annual meeting will take place October 15-16, 2015, in México FD (México), in association with the Mexican Federation of Rare Diseases FEMEXER. The event will be preceded by the 5th Latin American meeting of Rare Diseases ER2015LA (GEISER Foundation and FEMEXER) October 12 and the World Congress of Discoveries and Innovations in Orphan Diagnoses and Drugs October 13-14. The events will be developed in a coordinated format incorporating all the main topics of interest within the rare disease field. For proposals or more information contact the local secretariats at proyecto.pideundeseo.mexico@gmail.com (FEMEXER), info@fundaciongeiser.org (GEISER) or the ICORD secretariat at icord@karolinska.se.



Future ICORD meetings

Any legally constituted not-for-profit organisation involved in the rare disease field (e.g. patient organisation, NGO, health research institute, university or governmental agency) may apply to host an ICORD annual meeting by submitting a letter of intent for a given year. The ICORD Board will invite selected applicants to submit full proposals to be evaluated in detail. Final decision about hosting of an annual meeting is made by the ICORD Board and approved by the ICORD General Assembly, which may consider both the quality and location of proposals as well as the strategic interests and priorities of the society.

Any member of ICORD can suggest the inclusion of topics and/or speakers in the programmes of the annual meetings. The respective Strategic and Planning Committee will consider it. Deadline for proposals will close 9 months before the meeting date (consult the ICORD website, www.icord.se).

For more information and questions please contact the ICORD Secretariat (icord@karolinska.se).