



**REPORT OF THE CONFERENCE  
ICORD 2013  
Building Global Bridges  
for Rare Diseases Research  
and Orphan Product Development  
The 8th International Conference  
on  
Rare Diseases and Orphan Drugs  
October 31st - November 2nd, 2013  
Saint Petersburg, Russia**

**Organizers:** ICORD and The National Association of Organizations of Patients with Rare Diseases "Genetics"

**Co-organizers:** Geiser Foundation (Argentina), CTSI (Clinical and Translational Science Institute) (USA), ICRDOD (Bulgaria), NZORD (New Zealand), JPA (Japan), Eurordis (France), «Center of the Innovation and integration programs and technologies» (Russia).

**Conference language :** English, Russian.

**Websites:** [www.icord.se](http://www.icord.se) , [www.icord2013.ru](http://www.icord2013.ru)

**Country participants :** Russia, Brazil, Canada, Germany, Italy, Japan, Nigeria, Sweden, Switzerland, Turkey, United Kingdom, United States, Argentina, Belgium, Bulgaria, Spain, Georgia, Kazakhstan, Colombia, Lithuania, New Zealand, China, Serbia, Ukraine, France, Gambia, Bosnia and Herzegovina

**The conference was attended by** 234 participants.

**The conference included:** Meeting of the Board ICORD, Round table of the «National Council of Experts on Rare Diseases», Satellite Symposium of Pharmaceutical Industry, 9 sessions, 2 Hot Topic Conferences and 4 Working Groups, which were the leading experts in the field of rare diseases from different countries. 41 presentations was heard.

## OPENING



**The First Deputy Chairman of the Federation Council of the Russian Federation A.P.Torshin** has welcomed the participants of ICORD 2013 on behalf of the Federal Assembly of the Russian Federation. In his speech he has admitted this is an authoritative discussion platform which held a substantive, thoughtful discussion on key issues in the field of medical affairs rare diseases and orphan drugs. A.P.Torshin also emphasized that debatable discussion of the topical and sensitive issues will contribute for developing common solutions, and expressed confidence, that this meeting will give a new impulse for developing national health care and will help to solve actual points in the theory and practice of medical treatment of rare diseases. He has full comprehension about real existence of this problem in Russia where are 83 subjects of the federation. **He told to the participants of the conference about work of the Senator from Penza region, founder of BIOTECK B.I.Shpigel. And also gave words of encouragement from the Minister of Health V.I.Skvortsova.** Alexander Porfirievich expressed his gratitude to the National Association of Organizations of Patients with Rare Diseases "Genetics" for the fact that it gives hope for the people with rare diseases, "fights to the last" for the each life.



**Vice Governor of the Government of St.-Petersburg O.A. Kazanskaya** has welcomed the participants of ICORD 2013 on behalf of the Government St-Petersburg and on behalf the Governor of St.-Petersburg. In her presentation O.A. Kazanskaya has noted that the main goal of the Conference is developing of the constructive international partnership which lead to the global improvement of the life of patients with rare diseases and developing of the transboundary cooperation in the treatment of rare diseases, development of the orphan drugs and rarely used medical technologies. The Conference ICORD 2013 gathered world-class experts, physicians, researchers, representatives from Pharm industry, public authorities, Patients movement from many countries, and people, connected by desire to improve quality of life of patients with rare disease and their families conducting new researches, developing new technologies and increasing their knowledge. The forum is striking confirmation, that problem of rare diseases and laws about people suffering from it, unites all stakeholders and knows no national boundaries. She also emphasized this is a honor for Saint-Petersburg to be a place for holding the Conference because this city has advanced achievements of the science, technology and medicine. In Saint-Petersburg there are more than 470 people including 166 children suffering from rare diseases. This is important to build the common system which provide favorable conditions for treatment and life of patients and which will connect all stakeholders: health and social services, community organizations, representatives of executive and legislative power of the city, charitable foundations.



**Deputy Chief of headquarters of interfractional deputy association "Eurasia" in the State Duma of the Federal Assembly of the Russian Federation N.N.Starikov** expressed his sincere gratitude to all the participants who came to the conference from different countries who devote their attention and energy to help people with rare diseases and their families. In his speech he said that today it is important that the international community is paying special attention to rare diseases. And Russia in this work is one of the main participants.

At the opening ceremony **S.I.Karimova the President of the National Association of Organizations of Patients with Rare Diseases "Genetics"** addressed to the participants of the conference with a welcoming speech:

"We hope that the conference will serve as a convenient platform for global dialogue International and Russian experts, will determine the vector further exchange experiences and develop common approaches to sustainable global development in an assistance to patients with rare diseases . For us it is important that Russia is now the part of a developing global work that makes ICORD for many years. We expect that the proposals which will be in result of the conference, the Russian Federation and the international community will consider and implement in practice in the near future. " **S.I.Karimova has read out welcoming letter on behalf of Federal Council of the Russian Federation, State Duma and Health Committee, directly from the Chairman of the Committee S.V.Kalashnikov.** In his letter S.V.Kalashnikov claimed that nowadays before the medical scientific community faces serious challenges, the main objective - improving the accessibility and quality of the medical care. And he expressed his gratitude to the all those present for indifferent attitude to the present and future of people with rare diseases.





**Virginia A.Llera President of ICORD, Founder and President of "GEISER"** appealed to the participants and guests of the event with parting words.

She noted how great the difference today in different regions of the world for rare diseases and the importance of global cooperation to develop the best methods of care for patients with rare diseases. Virginia A.Llera emphasized that holding of the conference ICORD is an annual and gradually leads towards integration of excellence in research, high technology combined with the development of projects and the state policy in the field of rare diseases



## 31 October

Before the main working days of the conference was a meeting of board members of ICORD. At a board meeting it was decided to adopt a member organization which will represent Russia in the ICORD. This honor was given to the National Association of Organizations of Patients with Rare Diseases "Genetics". Our Association will participate in conferences in Amsterdam in 2014 and 2015 Mexico City has as a full member of ICORD.



## Round table of the National Council «National Council of Experts on Rare Diseases»,

On a round - table gathered board members of advisory council and experts in the field of rare diseases (main experts, academics, representatives of patient communities, businesses) for solving the further development directions of the Board, the main provisions of possible proposals to the authorities, as well as was considered "Strategy for the development of state policy in the field of rare diseases by 2025 ." Led roundtable Asanov A. - President of NC "Council of Experts in the field of rare diseases ." A report on the situation with rare diseases was made by chief geneticist MoH RF Novikov P.V and he has led the discussion. It was decided to improve the strategy all the expert community and discussed priorities for the work of the Council.



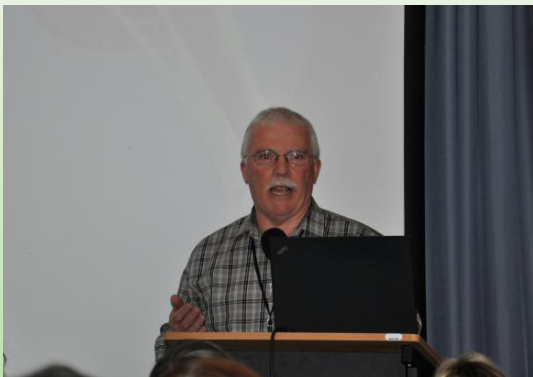
## Satellite Symposium of Pharmaceutical Industry

In the afternoon, a symposium of pharmaceutical industry was held, which was attended by more than 70 delegates of pharmaceutical industry. The main theme of the symposium was "Including industry needs in spending up global orphan drug accessibility."



During the meeting, a report on "Rare Diseases and Orphan Drugs priorities as seen from the Academia" was delivered by Professor Karolinska Institute and Director of Research and Education, Karolinska University Hospital in Stockholm, **Jan-Inge Henter**.

Research Project coordinator of “Sistemas Genomicos” Ph.D **Rebeca Minambres Herraiz** told about new genomic technologies. The application of Next Generation Sequencing to the development of new drugs has potential advantages: a) benefits for pharma industries: saving money and time in the stages to achieve new more effective drugs , b)benefits for society and rare diseases patients: reducing the process to have personalized treatment at their disposal and increase of new orphan drugs. It's becoming a real business for innovative pharmas: developing the “biomarker and drug” pack.



**John Forman**, executive director of the Organization for Rare Diseases in New Zealand, touched on the main aspects that must be changed on the existing tendency, refer to the interests of patients without proper attention.

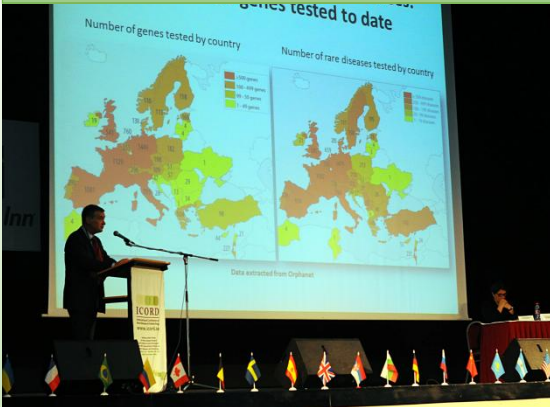
In continuation of the meeting was made by **Howard Yuwen**, executive director of Alexion Pharmaceuticals, a report entitled "Improving communication and collaborations among stakeholders in rare disease community to expedite drug development."





In the first work day of the Conference (Nov.1) were held 4 sessions, plenary conference, Hot Topic Conference and session of the four Working Groups.

On the Plenary Conference with report on theme «**IRDiRC**: an opportunity for worldwide rare disease research networking» made by Chair of the Executive Committee of the International Rare Diseases Research Consortium **Paul Lasko (Canada)**.



**Paul Ledger**, Head of Commercialization for EMAP Rare Disease for GSK (UK), behalf of A. Lemoine said about innovative collaborations models to fund discovery stage rare disease research. GSK has been applying creative approaches to find and fund innovative early stage rare diseases science in academic laboratories through: 1. Indirect project financing: participation in a venture capital fund. 2. Direct financing: creative partnership with academic institutions through dedicated alliance management unit. The main goal – build integration collaboration to convert innovational researches in medicine for help patients.



The main theme of **Session 3** became “Primary Prevention, Modern Diagnostic and Screening for Rare Diseases”.

The Session was opened by **Ann Nordgren**, Senior Consultant of Karolinska University Hospital (Sweden), with presentation “Next Generation Sequencing for diagnosis of Rare Diseases in clinical practice”



**Alberto Mantovani**, Director of Food and Veterinary Toxicology Unit at the ISS (Italy), said about primary prevention of congenital anomalies and paid particular attention to safety precautions. In his report Alberto Mantovani gave a good example regarding foods and environment namely a ban European Unit of chemical agent bisphenol A which was used for manufacture plastic for baby bottles.



**Tom Pulles**, Global Medical Director for Shire's MPS program (Switzerland) summed up the Session 3 with report "Challenges and opportunities of screening initiatives in rare diseases". Although screening may lead to an earlier diagnosis, not all screening tests have been shown to benefit the person being screened; over diagnosis, misdiagnosis, and creating a false sense of security are some potential adverse effects of screening. For these reasons, a test used in a screening program, especially for a disease with low incidence, must have good sensitivity in addition to acceptable specificity. T.Pulles picked out three main methods of screening: **Mass/universal screening** involves screening of all individuals in a certain category (for example, all children of a certain age ; **High risk or selective screening** is conducted among (high) risk populations only (hereditary diseases); **Multiphasic screening** application two or more screening test to a large population at one time instead of carrying out separate screening tests for single disease.





Holding meetings of the **Hot Topic Conference** held in parallel in the Congress Hall "Moscow" and in the hall "Dayneko."



In the Congress Hall "Moscow" **Klaus Mittmann** presented the report behalf of **Malcolm Allison**. In his speech he highlighted the problem of CTEPH treatment and reimbursement in Europe. He emphasized the importance that patients It is essential that patients, who are suspected of having PH, are sent to Specialist Centers for assessment. CTEPH is curable by surgery in more than half the cases, so surgery should always be considered as the first option. For patients, for whom surgery is not an option, or for whom surgery has not resulted in complete restoration, Riociguat is the first agent to show successful outcomes. Riociguat was generally well tolerated with a good safety profile in patients with CTEPH

Professor and Doctor of Medicine, Director of the Center OncoHematology and Transplantation Northwestern State Medical University named Mechnikov , **Igor Lisukov** made a presentation on the role and impact on decision-making in access to innovative treatment on the example of rare diseases in hematology . The main thesis of the report is the statement that officially registered orphan drugs are often defined as "necessary for the use ( treatment) " , they will not actually "available " until they are legally only "claimed" , but approved in the established order of the state system health within the effective model compensation in case of relevant medical evidence.







**Rosa Yagudina**, Head of the organization of drug supply and pharmacoconomics First Moscow State Medical University named Sechenov, spoke about the contradictory method "analysis of the impact on the budget" for a decision on funding of orphan drugs and selecting criteria for inclusion in the public drug program. On an example of the treatment of multiple myeloma drug bortezomib and lenalidomide.

On the situation on rare diseases in Russia told **Aly Asanov**, President of the "National Council of Experts in the field of rare diseases", Head of the First MGMU named Sechenov. In his report, Professor A. Asanov drew attention of participants to the statistics of patients with rare diseases in Russia: in the moment at least 1 million people in the Russian Federation patients with rare diseases.



According to Professor **A.Asanov** the Main RD patients Medical Care problem in Russia are:

- No Diagnosis or difficulties in diagnosis
- Boundedness of treatment options even if Diagnosis presents
- Treatment exists, but Orphan Drugs are not yet registered in Russia
- Orphan Drugs has been registered, but the cost is so high that they cannot be used.
- Orphan Drugs are available BUT:
- No experience usage (especially in province);
- There are only separate standards and patient treatment protocols;
- Very few hospital agreed to use Orphan Drugs;
- No therapy monitoring system (i.e.the feedback)



In the hall " Dayneko " Head Global Market Access Rare Diseases «Shire AG», **Jose Fernando Gomez Martinez** made a report "Prices of medicines for very rare diseases: Balancing, Value, Risk, Cost and Innovation", on the basis of inside information of "Shire ".

In his speech, speaker revealed such components in drug costs, as the cost of research expenses, clinical trials and insurance associated with treatment "orphan" drugs risks. In particular, it was shown that the company's revenue reinvestment in research activities at certain times were up to 44% of total revenues .

Within the **Session 4** "Regional Scenario (Russia, Eastern Europe/and Southern Caucasus country, and/or Central Asia country)" their presentation represented:

Executive Director of NPO GeRAD, Eurodis Advisor in Russia, Ukraine and Georgia in the frame of the EUROPLAN project **Oleg Kvlividze** made a presentation "The collaboration in the field of RD in EE countries: today experience and tomorrow prospects" he said about the history of activity in the field of rare diseases in Georgia, and emphasized the cooperation of the former Soviet Union and Eastern Europe countries. he mentioned the main problems in his country in the RED sphere

Absence of a special law to regulate the situation of RD

Absence of global support from the State at the level of diagnostic and treatment programs

Low activity in the donor organization in the field of RD

Poor integration of the scientists, physicians and organizations working in the field of RD into the international institutions and programs

Lack of qualitative information available about rare diseases.



**Ugur Ozbek**, Director of the Institute of Experimental Medicine at the University of Istanbul, focused his presentation on the activities and practices in the field of rare diseases in Turkey. He stressed the importance of the availability of prenatal screening, if there is a genetic or medical requirement, and the need to support the activities of patient organizations.



Director Public Affairs Genzyme EMEA, a SANIFI company **Vinciane Pirard** in her report noted that:

All kind of needs and scenarios but all have to consider the long term (eg: policies targeted to children need to take in account they will grow in adulthood)

Do not focus only on access to treatment - Optimize the process of care .

New policies are inspired by existing experience: Political willingness and Funding mechanisms





## On the first working day of the conference, a special sessions of the working groups took place

In the congress hall “Moscow” was held a meeting of the Working Group 1 (Moderator S.Karimova/A.Makaeva) “The legislative basis for state regulation in the Russian Federation in the field of rare diseases”



The meeting discussed the problems that were identified during 2013 in the Russian Federation, such as problems with the provision of drugs and problems in rehabilitation.

***Participants concluded that the development of the Strategy of the Russian Federation in the field of rare diseases, the "road map", the amendments to the Federal Law, as well as help the pharmaceutical industry to provide technical support for the establishment of registers and offer new schemes for provision of medicines can solve these problems.***



Also were held 3 meetings of the **Working Group «Seed Accelerators»**.

Results of the meeting **Working group A (Facilitator: Timothy R. Cote)**, the main theme of discussion was «Discuss Building and International Network; Rare Disease Patient Care and Regulatory Action»:

Under the category of regulatory harmonization, it was decided that the “easiest” or most achievable goal was the establishment of a universal criteria for orphan designation

- It was also discussed that universal harmonization in other related areas (such as patient registries, data collection standardization, healthcare system and infrastructure development plans, etc...) would benefit developing countries and prevent “reinvention of the wheel”
- Successful examples from which these infrastructures could be modeled include Children’s Oncology Group, the European reference networks (groups of leading hospitals and organizations), and the European clinical trials directive (collective IRB approval)
- The mechanism by which these systems could implemented include driving organizations such as ICORD, WHO, Pan American Health Organization



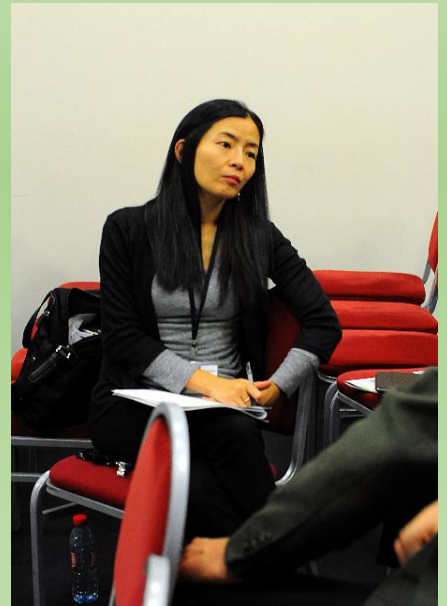
Participations of the **Working Group B (Facilitators: Emilio Roldan, Maja Stojiljkovic)** discussed issues «Research collaboration in rare diseases; epidemiology, basic research and clinical trials».

Within the ideas discussed during the session the proposal of pre-planned studies, that is the make-up of guide-lines for conducting clinical studies with small sample-sizes but appropriate for further prospective meta-analysis will increase the validity of data.

International collaboration in rare diseases needs to be organized, including studies, which are not typically made for registration of therapeutics, but essential for the daily work of practitioners as long-term phase IV trials. Epidemiology trials of rare diseases are highly requested because very few data is available.



Guidelines for patients consent in order to exchange valid data from multiple centers were also discussed. Finally, can ICORD become a bridging tool for worldwide researches? Guidelines, meetings, publications, interactions with other institutions, can be duly promoted by the organization, specially having the opportunity of introducing the ideas within the regions less developed in the field. Big organizations, as well as small initiatives can be supported from ICORD and linked to other driving means.



Participants of **Working Group C (Facilitator: Yukiko Nishimura)** discussed issues, connected with quality life of patient with rare disease and family members: joint efforts for better understanding regional differences and building maps.

On the second day of the conference - **November 2**, work continued in the Sessions and in the framework of the Hot Topic Conference.



**Session 5** («Clinical Trials of Rare Diseases and Orphan Drugs and Repurposing of Existing Drugs»), begun from report of **Simon Day**, Director of "Clinical Trials Consulting&Training Limited" «Статическое планирование и сбор достоверных данных». Свою работу он посвятил исследованиям и клиническим испытаниям.

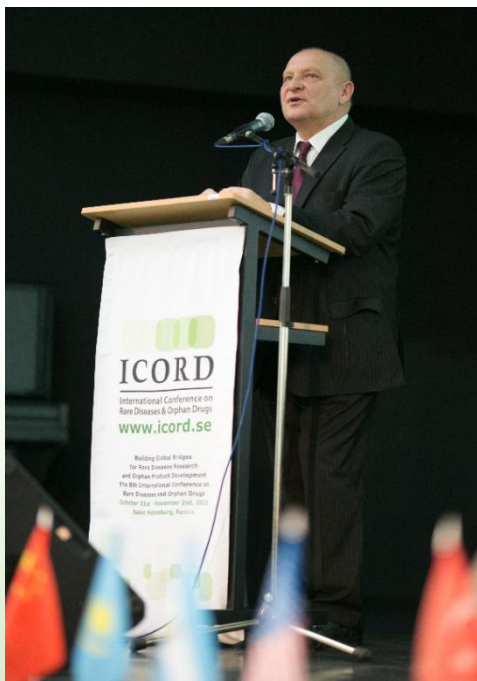
On behalf of **Stephen Groft** made **Marlene Haffner**. She spoke about the work of the National Center for Health in the U.S., research programs, and resources to establish a clinical research network in the field of rare diseases.



Professor regulatory practice at the Institute Claremont, California, **Timothy Cote** in his report paid attention to this problem as the judicious use of existing drugs in the treatment of diseases. He talked about repurposing use of drugs, when the drug for the treatment of rare diseases can be used in the treatment of common diseases and vice versa. Of his report follows, that this approach has great clinical potential, and this may require a change in local politics.







Professor of Department of Hematology, Oncology and Radiotherapy RSMU n.a. Pirogov **Veniamin Chernov** devoted his report to experiences on running clinical trials on Rare Diseases in Russia.

The main problem in the study of rare diseases - a set number of patients needed to obtain correct conclusions. Joint efforts are needed to address the diagnosis and treatment of rare diseases. In our country the opportunity to develop , manufacture and use of orphan drugs. Effectiveness of the treatment of rare diseases can be demonstrated in multicenter cooperative studies.



The main topic of the **Session 6** became «Big ideas for small populations: incentives, regulatory flexibility and harmonization».

Vice President – Business Development of Dohmen **Penny Bemus** said about “Value-Driven Supply Chain Models to meet the Needs of Individual Patients”. In her report she compared historical development two models: B2B and B2C in the USA and Europe. She admitted that healthcare supply chain is inefficient and does not meet the needs of individual patients.







**Howard Yuwen**, Executive Director of Alexion Pharmaceuticals, spoke about the 30-year development of orphan drugs on the example of the United States. Were marked success in this industry (public awareness and attention to rare diseases, the lives of many patients with rare diseases are saved or changed to the better) and identifies goals for further development.



E-Rare Project Coordinator **Daria Julkowska** made her report about Fostering transnational cooperation for rare diseases research funding on the example E-RARE project. The aim was to provide to promising independent investigators the opportunity to build transnational collaborations in the field of rare diseases research. The E-RARE Group of Funders recently joined the IRDiRC that teams up researchers and organizations investing in rare diseases research in order to achieve two main objectives by the year 2020; to deliver 200 new therapies and the means to diagnose most rare diseases.



Science Policy Director at Pfizer **Adam Heathfield** made his report on the topic of EU Policy developments on HTA-Regulatory interface and implications for global harmonization. His summary is:

1. EU essentially looking to bridge the gap in evidence requirements between regulators and payers and shift to earlier approval:

- Challenging even for the EU with major differences in national health systems

2. EU/US efforts on orphan designation process should be unaffected

3. Parallel scientific advice may be rendered more complex:

What decision point(s) are we aiming for with adaptive licensing? Same as US?

- Can we address all perspectives (FDA plus various EU payers and HTA bodies)?
- 4. Need to harmonise to drive efficiency and to encourage dialogue between all stakeholders, but must create something tractable
- 5. Harmonised ambition to get safe and effective new medicines to patients as rapidly as possible

Do we still agree, or are the economics becoming dominant and divisive?



Results of **6 session** summed report of Associate Professor of Faculty Therapy. Academician A.I Nesterov Medical University RNIMU n.a. NI Pirogov Alesya Klimenko on behalf of N.Shostak.

She told conference participants about the new methods of treatment of rare diseases, such as Chronic thromboembolic pulmonary hypertension(CTEPH). Pathogenesis of the disease has not been fully elucidated, and factors contributing to its development remain poorly defined. Concerning the use of new oral anticoagulant (rivaroxaban) in patients with CTEPH, we should be aware of drug interactions with Prostacyclin, ERA PDE5i.

**Session 7** “Clinical Guidelines and Best Practices” was opened by **Domenica Taruscio**.



**Domenica Taruscio** spoke about the four-year project RARE-Best Practices, which aims to improve the clinical management of patients with rare diseases and narrow the existing gap in quality of care between countries, creating a suitable platform to collect, evaluate and disseminate high-quality treatment and updating information on rare diseases.

In his report, Professor **Jan-Inge Henter** spoke about the positive aspects of international cooperation on the example of rare diseases with a low survival rate and children cancer:

We can now cure around 75% of all children with cancer.

This is mainly the result of collaborative studies among academic researchers.

We mainly use chemotherapy, surgery and irradiation.

Note: The drugs we use are old drugs, used in novel combinations.

Almost all these studies have been academia driven, and usually international efforts (as within SIOP)



**Nickolas Ah Mew**, Doctor-consultant with the Department of Genetics at children's National Medical Center in Washington, in his report "A Multi-Institutional Research Network in Rare Diseases" marked, that thanks to Multi-Institutional Research Network, three new drugs, have been approved, two drugs under clinical trials and new equipment developed.

**Alexey Sokolov**, professor of internal medicine and nephrology SZGMU Mechnikov, reported a rare technologies for the treatment on the example of therapeutic apheresis. From his report that technology Therapeutic apheresis (TA) are effective in the treatment of some rare diseases (RD), when no other treatment is not open. But to date, high medical technology can not compete with the pharmaceutical industry because of the size of the market, so these technologies need more support.





## Holding meetings of the Hot Topic Conference was held in the Congress Hall "Moscow".

The keynote speaker of the session was **Evelina Paberze**, representative of Orphan Europe. The theme of her report was "Advancing Knowledge in Rare Diseases." On the example of Orphan Europe: to emphasize cooperation in the field of rare diseases, awareness and education, the steady expansion of "live" and e-learning, training activities; dialogue with the scientific community, international networks.



**Session 8** «Health Policy and National Plans for Rare Diseases» started with report of **Yukiko Nishimura**, Chief Secretariat International Relations «Japan Patients Association». Prof. Nishimura Y. told about the current situation with policy NANBYO in Japan. The JPA is the largest organization in association of NANBYO. **Target disease** of NANBYO policy: 1.Etiology is unknown 2.Frequency is low (less than 50,000) - rare disease 3.Therapy is not established -hardly curable 4.Economical, psychological and physical burdens - progressive course / necessity of heavy care. **Target area** of NANBYO policy: Research Study, Medical, Welfare



**Rumen Stefanov**, Professor of Public Health and Dean of the Faculty of Public Health at the Medical University of Plovdiv (Bulgaria), made a presentation on "Rare Diseases National Plan in Eastern Europe." Professor Stefanov R. spoke about Setting the scene for rare disease health policy in Eastern Europe. He marked positive and negative moments in the Information and awareness, Diagnosis and screening, Management and rehabilitation, Patient organizations and National Plans in the Eastern European countries.



**Albina V. Makaeva**, Master of Business Administration, Master of Public Administration, Vice President of the National Association of organizations of patients with rare diseases "Genetics", spoke on "The development strategy of the state policy in the field of rare diseases by 2025." Was reviewed by three objectives, namely: information, social, financial - with which it is possible to successfully generate the implementation strategy. Also been proposed subsequent steps and mechanisms for the successful development and implementation of this project.



At the end of the session **Manuel Posada De La Paz**, director of the Institute for Rare Diseases (IIER) spoke on "Social Economic Burden and Health-related Quality of Life in Patients with Rare Diseases in Europe(BURQOL-RD)». Manuel Posada told the conference participants on the activities BURQOL-RD, and its main benefits: The BURQOL-RD model will provide an integrated and harmonized means to assess the impact of new public health policies, interventions and treatments for RD "in" and "among" EU Member States. Moreover, the associated dissemination activities undertaken by BURQOL-RD improve RD awareness and literacy among European citizens.



**Session 9** «Patient Priorities in Primary Prevention, Diagnosis and Clinical Care of Rare Diseases» was opened by President of the “National Association of Organizations of Patients with Rare Diseases "Genetics" **S.I.Karimova**

The report examined priorities for primary prevention and diagnosis of rare diseases in Russia. In her speech, **S.I. Karimova** noted that treatment of patients with rare diseases is one of the biggest problems of the health system, and not only in Russia. Diagnostics of such diseases is often difficult or unavailable, treatment is ineffective because of the lack of appropriate medicines, treatments. The result of these factors are unfavorable forecasts for patients. The most important priorities are : Epidemiological Registry , monitoring the situation throughout Russia; educational programs for physicians and pediatricians who are the first to identify the symptoms of rare diseases patients, implementation of national programs for genetic testing to determine the risk of disease before symptoms appear, the introduction of territorial program for each region - promoting a healthy lifestyle and attachment “roadmap” patient. In the implementation of these programs will be achieved the goal of reducing morbidity and mortality population based on a complex problem solving prevention and diagnosis of treatment and rehabilitation , as well as prolongation of the quality of life of patients with rare diseases .



**Svetlana Karimova** drew the particular attention to the work of the Resource Information Center for Rare Diseases, which is established by the National Association of organizations of patients with rare diseases "Genetics" . She stressed that today it is the only existing Russian information center that fully supports patients with rare diseases. In the Resource Information Centre for Rare Diseases received calls from all regions of the Russian Federation, where patients get the information about possible methods of rehabilitation, treatment, prevention, assistance in increasing the efficiency between patients, health professionals, public authorities, representatives of science and business, the distribution of legislative and legal acts , providing regional public authorities information about the state of the situation on rare diseases in their regions.



Professor at the National Children's Medical **Center Pablo Cure** spoke on "Rare diseases in Latin America: Identifying the Needs and Finding Appropriate solutions." He stressed the need to create awareness about rare diseases at the local, national and regional levels, as this is a key aspect in achieving the fact that rare diseases - a real problem in Latin America.



**John Forman**, Executive Director of the New Zealand Organization for Rare Disorders, in his oral presentation he claimed that many birth defects are preventable and most are rare diseases. In his opinion two-thirds of cases are preventable by fortification of a staple food with Folic Acid. He believed that cancellation of mandatory fortification of bread in New Zealand is damage more harder than damage from thalidomide.



***The results of the conference ICORD 2013*** mentioned the high level of the cooperation among all stakeholders: Patient community, professionals, researchers, government, and that is very important - civil society. Necessary is raise awareness of rare diseases, promotion of cooperation and the continued development of constructive international partnerships in all directions care of patients with rare diseases, knowledge sharing, both within Russia and within the international community. Need for new legislative and policy initiatives, as well as the completion of the legal framework in the field of rare diseases.

Conference proceedings will be published in a special supplement to the international magazine «RARE Journal», which will be devoted to the conference's ICORD 2013. Get more information about the conference, check out a program and materials are on the conference website: [www.icord2013.ru](http://www.icord2013.ru) , [www.icord.se](http://www.icord.se). Also will be posted on the websites of the photo report on the conference and links to videos of the plenary lectures.

### ***ICORD Conference 2014 :***

The next ICORD conference will be held in the capital of Netherlands, Amsterdam, in the fall of 2014.

Recently, in Western Europe, appeared a large number of initiatives and projects, related to rare diseases , successful experience which must be borrowed for the international community, and not be limited to the inside of the Western European region. Discussion of this experience will be a key issue in the upcoming conference .

### ***ICORD Conference 2015 :***

The Annual Conference ICORD 2015 held in Mexico and will be a large-scale platform for discussion of the differences, needs and priorities of the organization for rare diseases from around the world .

**genzyme**  
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**sobi**  
SWEDISH ORPHAN BIOVITRUM

**ALEXION**

**Shire**

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