

**EUROPLAN**

European Project for Rare Diseases National Plans Development  
Coordinated by the Italian National Centre for Rare Diseases  
Italian National Institute of Health

2012-2015



# RUSSIA

## EUROPLAN NATIONAL CONFERENCE

### FINAL REPORT

28 February 2013, Moscow



# FOREWORD

**The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.**

These national plans and strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN National conferences are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

**EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines.** They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;
2. Definition, codification and inventorying of RD; Information and Training;
3. Research on RD;
4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;
5. Orphan Drugs;
6. Social Services for RD.

The themes “Patient Empowerment”, “Gathering expertise at the European level” and “Sustainability” are transversal along the conference.

## I. General information

<b>Country</b>	<b>RUSSIA</b>
<b>Date &amp; place of the National Conference</b>	February 28, 2013 Moscow, Hotel "Arbat", 12, Plotnikov lane
<b>Website</b>	<a href="http://www.nacgenetic.ru/">http://www.nacgenetic.ru/</a> ; <a href="http://www.rare-diseases.ru">http://www.rare-diseases.ru</a>
<b>Organisers</b>	- <b>National Association of Organizations of Patients with Rare Diseases “Genetics”</b> - <b>Russian Patients Union (Rare Diseases Working Group)</b>
<b>Members of the Steering Committee</b>	<b>Irina Andreeva</b> – Deputy Minister of Health of the Russian Federation <b>Igor Seleznev</b> – Health Care Committee of the State Duma, Chairperson <b>Petr Novikov</b> – Professor, Chief non-staff specialist on the medical genetics of the Ministry of Health of the Russian Federation <b>Oleg Kvlividze</b> – EURORDIS Advisor in the frame of EUROPLAN 2 project <b>Svetlana Karimova</b> - National Association of Organizations of Patients with Rare Diseases “Genetics”, President; National Charity Foundation of the Assistance to People with Rare Diseases "Rares", President <b>Ian Vlasov</b> – Russian Patients Union, Co-chairperson <b>Jury Zhulev</b> - Russian Patients Union, Co-chairperson <b>Irina Myasnikova</b> - Russian Patients Union, Rare Diseases Working Group, Chairperson
<b>Names and list of Workshops</b>	<b>PLENARY SESSION 1: RARE DISEASES IN RUSSIA – ACHIEVEMENTS AND CHALLENGES</b> <b>PLENARY SESSION 2: WRAP-UP SESSION;</b> <b>WORKSHOP 1: METHODOLOGY AND GOVERNANCE OF A NATIONAL PLAN</b> <b>WORKSHOP 2: DEFINITION, CODIFICATION AND INVENTORYING OF RD; INFORMATION AND TRAINING</b> <b>WORKSHOP 3: RESEARCH ON RD</b> <b>WORKSHOP 4: CARE - CENTRES OF EXPERTISE / EUROPEAN REFERENCE NETWORKS</b> <b>WORKSHOP 5: ORPHAN DRUGS</b> <b>WORKSHOP 6: SOCIAL SERVICES FOR RD</b>

<p><b>Workshop Chairs</b></p>	<p><b>CONFERENCE CO-PRESIDENTS:</b>  <b>Svetlana Karimova</b>, National Association of Organizations of Patients with Rare Diseases “Genetics  <b>Irina Myasnikova</b>, Russian Patients Union, Rare Diseases Working Group  <b>Jury Zhulev</b>, Russian Patients Union  <b>Ian Vlasov</b>, Russian Patients Union</p> <p><b>PLENARY SESSIONS AND WORKSHOPS CHAIRS:</b>  <b>Irina Andreeva</b> – Ministry of Health of the Russian Federation  <b>Prof Petr Novikov</b> –Ministry of Health of the Russian Federation  <b>Prof Rumen Stefanov</b>, Plovdiv Medical University  <b>Svetlana Karimova</b>, National Association of Organizations of Patients with Rare Diseases “Genetics  <b>Irina Myasnikova</b>, Russian Patients Union, Rare Diseases Working Group  <b>Prof Valentina Larionova</b>, Saint Petersburg State Medical University  <b>Prof Aleksander Rumiantsev</b>, Director of FSBI «FSCC of Children’s Haematology, Oncology and Immunology</p>
<p><b>Attachments</b></p>	<p>Programme; Photos; documents</p>

## II. Main Report

<p><b>Plenary Report – Opening Session</b></p> <ul style="list-style-type: none"> <li>• The <b>Country status</b> was presented by representatives of MoH/Irina Andreeva, Federal Service on Surveillance in Healthcare/Georgi Petruchenkov; medical professional/researcher/Prof Petr Novikov.</li> <li>• <b>European policy and guidelines</b> (e.g. EU document presentations and discussion, presentation of EUROPLAN, EUCERD and EUCERD Joint Action) The opening Plenary Session was partially focused on the presentation of the European documents (2008 EU Communication; 2009 EU Recommendation; EUCERD recommendations, EUROPLAN recommendations and indicators. Please see detailed information attached (Strategy)</li> <li>• <b>Other plenary</b> (N/A)</li> </ul>
<p><b>Report of Workshops</b></p> <p><b>Theme 1 - Methodology, Governance and Monitoring of the National Plan</b> (the themes listed below were presented by 3 speaker: Prof. Rumen Stefanov, Jury Zhulev and Svetlana Karimova)</p> <p><b>Sub-Themes:</b></p> <ol style="list-style-type: none"> <li>1.1 Mapping policies and resources (Yes)</li> <li>1.2 Development of a National Plan /Strategy (Yes)</li> <li>1.3 Structure of a National Plan /Strategy (Yes)</li> <li>1.4 Governance of a National Plan (Yes)</li> <li>1.5 Dissemination and communication on the National Plan (Yes)</li> </ol>

1.6 Monitoring and evaluation of the National Plan (Yes)

1.7 Sustainability of the National Plan (Yes)

Participants developed the main objectives in the field of **Methodology, Governance and Monitoring of the National Plan**:

- Identification of the priority areas of development within the National Plan: education and training among medical professionals and patients; information awareness among civil society; improving of the early diagnostics and prevention of RD; stimulation of research and adoption of the innovating technologies, particularly, in treatment;
- Development of the coordinating and control mechanism on creation and implementation of the National Plan (incl. all stakeholders, particularly patients' organizations) at regional, interregional, and national levels;
- Active participation of regional representatives in the creation of the National Plan; advocacy on adequate involvement of all territorial subjects of the Russian Federation in the activities for the main directions of its development;
- Development of the international cooperation. Participation in the international projects (particularly, in EUROPLAN).

**Theme 2 - Definition, codification and inventorying of RD; Information and training** (the themes listed below were presented by 2 speakers: Prof Igor Lisukov, Prof Valentina Larionova)

**Sub-Themes:**

2.1 Definition of RD (Yes)

2.2 Codification of RD and traceability in national health system (Yes)

2.3 Registries and databases (Yes)

2.4 Information on available care for RDs in general, for different audiences (Yes)

2.5 Help Lines (Yes)

2.6 Training healthcare professionals to recognise and code RD (No)

2.7. Training healthcare professionals (Yes)

Participants developed the main priorities in the field of **Definition, codification and inventorying of RD; Information and training**:

- Legislatively approve a new definition of a rare disease (instead of existing 1:10000), that will be more closer to the existing definition adopted in the European Union (1:2000);
- Intensification of work on creation of registries and registers in the field of rare diseases. Integration into the international programs and projects working on the creation of registers for rare diseases (EpiRare);
- Extension of the educational programs in the field of rare diseases as for students and residents, as well as for GPs in the capital and regions;
- Extension of information and awareness programs on existing schemes (in particular, care schemes) addressed to parents of patients with rare diseases;
- Improvement of existing and creation of new RD online information resources for medical professionals and patients. Finding the alternative ways to transfer information on RD.

**Theme 3 - Research on RD** (the themes listed below were presented by 1 speaker: Dr Ekaterina Zakharova)

**Sub-Themes:**

3.1 Mapping of existing research resources, infrastructures and programmes for RDs (Yes)

3.2 Dedicated RD research programmes and governance of RD research funds (Yes)

3.3 Sustainability of research programmes on RD (No)

3.4 Needs and priorities for research in the field of RDs (Yes)

- 3.5 Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects (Yes)
- 3.6 RD research infrastructures and registries (Yes)
- 3.7 EU and international collaboration on research on RD (Yes)

Participants developed the main priorities in the field of **Research on RD:**

- Studying of the epidemiology, incidence and prevalence of RD in the country;
- Creation of the biological material banks of RD;
- Creation of the scientific preventive programs on RD;
- Stimulating the pharmaco-economic researches of RD;
- Supporting existing registries and creation of new registries for particular forms of diseases;
- Optimization of the regulatory framework for the implementation of registries and information security;
- Promoting the inclusion of Russian patients into international and national research programs;
- Expansion of researches in the field of molecular diagnostics;
- Integration into the international research projects on RD.

**Theme 4 – Care for RDs - Centres of Expertise and European Reference Networks for Rare Diseases**  
(the themes listed below were presented by 2 speakers: Prof Vera Ijevskaya and Prof Aliy Asanov)

**Sub-Themes:**

- 4.1 Designation and evaluation of CE (Yes)
- 4.2 Scope and functioning of CEs (Yes)
- 4.3 Multidisciplinarity, healthcare pathways & continuity of care (Yes)
- 4.4 Access to information (Yes)
- 4.5 Research in CEs – How to integrate research on RDs and provision of care (No)
- 4.6 Good practice guidelines (No)
- 4.7 Diagnostic and genetic testing (Yes)
- 4.8 Screening policies (No)
- 4.9 European and international collaboration – Cross-border healthcare and ERNs (European Reference Networks) (Yes)
- 4.10 Sustainability of CEs (Yes)

Participants developed the main priorities in the field of **Care for RDs - Centres of Expertise and European Reference Networks for Rare Diseases:**

- Creation of the Expertise Centres with specialization on the management of rare pathologies on the basis of the federal scientific and clinical centers in a number of the large cities/regions of Russia;
- Delivering to patients available diagnostics of rare diseases in such centers through the introduction of modern methods;
- Implementation of the training programs in the field of the rare diseases for medical professionals on the basis of such centers;
- Promoting for giving to such centers the status of National Centers of Expertise/Reference Center on Rare Diseases;
- Establishment of new and expansion of existing communication avenues with the Centers of Expertise on rare diseases worldwide. Integration into European Reference Networks.

**Theme 5 – Orphan Medicinal Products** (the themes indicated below were presented by 3 speakers: Veniamin Chernov, Elena Chadova and Elena Nazarenko)

**Sub-Themes:**

- 5.1 Support to Orphan Drug (OD) development (Yes)
- 5.2. Access to treatments (Yes)
- 5.3. Compassionate use programmes (Partially)
- 5.4. Off label use of medicinal products (Yes)
- 5.5. Pharmacovigilance (Yes)

Participants developed the main priorities in the field of **Orphan Medical Products:**

- Optimization of the existing regulations in the field of import, registration, and realization of the orphan drugs;
- Development and launch of incentive mechanisms for Russian pharmaceutical industry to promote the production of quality orphan medical product;
- Advocacy in the optimization of existing and creation of new programs on treatment of rare diseases.

**Theme 6 –Social Services for Rare Diseases** (the themes indicated below were presented by 2 speakers: Irina Myasnikova and Anna Soykina)

**Sub-Themes**

- 6.1. Social resources for people with disabilities (Yes)
- 6.2. Specialised social services for rare diseases (Yes)
- 6.3. Policies to integrate people living with rare diseases into daily life (Yes)
- 6.4. International–supranational dimension (No)

Participants developed the main priorities in the field of **Social Services for Rare Diseases:**

- Intensification of the policy on social support of representatives of RD community both from the state and from socially focused public organizations at regional, interregional and federal levels;
- Advocacy to ensure an equal and free access to drug therapy and rehabilitation for patients with rare diseases;
- Development of a system of palliative care for patients with rare diseases;
- Promote and support activities and initiatives aimed to deliver social assistance services to the families of patients with rare diseases;
- Development of home care support services for families with heavy patients;
- Introduction of good practices developed at the European level in the field of social services for RD.

**Report of the Closing Session – Conclusions**

During the **Closing Session** the participants analyzed, and interpreted information provided during each of the workshops. They have carefully considered national realities and the European experience for the creation of a National Plan and optimal management of rare diseases.

- **Overall assessment of the usefulness of the European guidelines and policy recommendations:** the usefulness of EU guidelines and policy recommendations and necessity to be guided by them were approved by participants of the Conference.
- **Identification of specific gaps, challenges and needs across all Themes:** the gaps and the global challenges seem to mainly relate to the lack of proper information in professional and civil

societies. Early diagnosis and prevention of rare diseases and the shortage of the governmental financing were identified as basic needs.

- A **final resolution** was adopted.

### III. Participants

197 registered participants in total.

- Academic/Researcher – **16**
- Clinician/GP – **62**
- Healthcare Professionals (other than clinician or GP; geneticists and other laboratory workers) – **14**
- Industry – **24**
- Medical /Learned society – **12**
- Patient representatives – **52**
- Politicians – **5**
- Public administration – **12**

### IV. Programme

#### NATIONAL CONFERENCE

**“DEVELOPMENT OF THE STRATEGY OF RUSSIA IN THE FIELD OF RARE (ORPHAN) DISEASES FOR 2013–2016”**

**February 28, 2013**

**Moscow, Hotel "Arbat", 12, Plotnikov lane**

#### **WITH SUPPORT OF:**

Ministry of Health of the Russian Federation

EURORDIS - European Organization of Rare Diseases

Public and professional organizations of the Russian Federation

#### **PRESIDENTS OF THE CONFERENCE:**

**Zhulev Jury** - Russian Patients Union, Co-chairperson

**Vlasov Ian** – Russian Patients Union, Co-chairperson

**Karimova Svetlana** - National Association of Organizations of Patients with Rare Diseases “Genetics”, President; National Charity Foundation of the Assistance to People with Rare Diseases "Rares", President; Eurasian Alliance for Rare Diseases, Secretary General.

**Myasnikova Irina** - Russian Patients Union, Rare Diseases Working Group, Chairperson



**ORGANIZING COMMITTEE**

- **Andrusova Katerina** – IPO “Velikan”, Chairperson
- **Makaeva Albina** - National Association of Organizations of Patients with Rare Diseases “Genetics”, Vice-President
- **Khrolenok Oksana** – National Association of Organizations of Patients with Rare Diseases “Genetics”
- **Zakharova Ekaterina** – Russian Union of Rare (orphan) Diseases, Chairperson
- **Rumyantsev Aleksander**, Director of FSBI «FSCC of Children’s Haematology, Oncology and Immunology named after D.Rogachyov», Corresponding member of RAMS, Professor, Academician of RAMS
- **Terekhova Marina** – Interregional Public Organization for support to people disabled since the childhood, suffering from Gaucher's disease, and their families, Chairperson
- **Petr Novikov**, MD, PhD, Professor, Chief non-staff specialist on the medical genetics of the Ministry of Health of the Russian Federation
- **Shkolnikova Marina** - MD, PhD, Center of the Heart Rhythm Disturbances, Chief; RPO «Russian Association of the Paediatricians-cardiologists”, President

<b>February 28, 2013 Programme</b>		
<b>9.00 – 11.00</b>	<b>Registration Welcome coffee</b>	
10.00 – 11.00	Press-conference, dedicated to the Sixth International Day of Rare Diseases, February 28, 2013	
11.00 – 11.30	<b>Opening of the Conference</b> Welcome speech - Conference organizers Welcome speech - Oleg Kvlividze, EURORDIS representative, Professor	
11.30 – 11.40	Welcome speech of the representative of the Federation Council of the Federal Assembly of the Russian Federation	
11.40 – 11.50	Welcome speech - Igor Seleznev, Representative of the State Duma of the Russian Federation, Chief of the Staff of the Committee on Health	
11.50 – 12.10	<b>Irina Andreeva</b> , Deputy Minister of Health of the Russian Federation	Rare diseases – progress and problems
12.10 – 12.30	<b>George Petrochenkov</b> , Deputy Chief of the Department of Monitoring of State programs realization of Federal Service on Surveillance in Healthcare	State programs on the rare diseases in Russia
12.30 – 12.50	<b>Petr Novikov</b> , MD, PhD, Professor, Chief non-staff specialist on the medical genetics of the Ministry of Health of the Russian Federation	Prospects on the solution of problems of rare (orphan) diseases in the Russian Federation - EU recommendations
12.40 – 13.00	<b>Rumen Stephanov</b> , Professor of the Public Health; Initiator and principal partner of the EUROPLAN project; Member of the EU Interdisciplinary Committee «IRDIRC»	International experience on creation of the national plans for rare diseases. EUROPLAN 1 - EUROPLAN 2
13.00 – 13.20	<b>Jury Zhulev</b> , Russian Patients Union, Co-chairperson; Russian Society of Haemophilia, President	Development of cooperation in the field of rare diseases in anticipation of the National Plan

13.20 – 13.40	<b>Svetlana Karimova</b> - National Association of Organizations of Patients with Rare Diseases “Genetics”, President; National Charity Foundation of the Assistance to People with Rare Diseases "Rares", President; Eurasian Alliance for Rare Diseases, Secretary General	Regional programs in the field of rare diseases: features of development in a context of uniform strategy
13.40 – 14.00	<b>Igor Lisukov</b> , MD, PhD, Professor, Deputy director of the Institute of Children’s Haematology and Transplantology named after R.Gorbacheva	RD Definition in Russia; Registries and Registers
14.00 – 14.20	<b>Valentina Larionova</b> , Professor, Saint Petersburg State Medical University named after I.Pavlov, North-Western State University named after I.Mechnikov, “International Society of the Personalized Medicine”, Representative in Russia	Problems of education in the field of rare diseases. Implementation of training programs. Informing.
14.20 – 15.00	<b>Ekaterina Zakharova</b> , MD, PhD, Head of Laboratory of Hereditary Metabolic Diseases of FSBI “Medical Genetics Research Center” of RAMS, Russian Society of Rare (orphan) Diseases, Chairperson	Researches in the field of rare diseases – from the exact diagnosis to the discovery of new methods for treatment
15.00 – 16.00	<b>LUNCH</b>	
16.00 – 16.20	<b>Vera Izhevskaya</b> , MD, PHD, Medical Genetics Research Center of RAMS, Deputy Director on Science	Centers of rare diseases diagnostics. World and Russian experience
16.20 – 16.40	<b>Ali Asanov</b> , MD, PHD, Professor, Chair of the Department of medical genetics, Moscow State University named after Setchenov	Formation of expert community in the field of rare diseases in Russia
16.40 – 17.00	<b>Veniamin Chernov</b> , MD, PHD, Deputy Director of FSBI «FSCC of Children’s Haematology, Oncology and Immunology named after D.Rogachyov»	Orphan drugs: clinical trials
17.00 – 17.20	<b>Elena Chadova</b> , Deputy Minister of Health of Sverdlovsk region	Experience of Sverdlovsk region in the organization of medical care for the patient with rare (orphan) diseases
17.20 – 17.40	<b>Ludmila Nazarenko</b> , Deputy Director on Scientific and Clinical fields of Scientific Research Institute of Medical genetics of the Siberian Department of RAMS	Experience on orphan diseases treatment in the Siberian federal district

17.40 – 18.00	<p><b>Irina Myasnikova</b>, Russian Patients Union, Rare Diseases Working Group, Chairperson</p> <p><b>Marina Terekhova</b>, Interregional Public Organization for support to people disabled since the childhood, suffering from Gaucher's disease, and their families, Chairperson</p>	Role of public organizations in the assistance for patients with rare diseases
18.00 – 18.15	<p><b>Anna Sonkina</b>, Consultant physician in palliative care of the Orthodox Charity service</p>	Problems of rendering the palliative care to the patients with rare diseases
18.15 – 19.00	<p><b>Wrap-up session; Adopting the Resolution</b></p>	