

**Formation of national understanding of rare diseases:
from concept to national program**

Rare diseases in the EU and Ukraine

For the EU countries this concept is well known - "Rare diseases are a threat to the health of EU citizens, in so far as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity".

The specific problems and needs of patients with rare diseases have been reported and explained in several basic European documents, such as:

- Decision No.1295/1999/EC of the European Parliament and of the Council of 29 April 1999 adopting a programme of Community action on rare diseases within the framework for action in the field of public health (1999 to 2003);
- Regulation (EC) No 141/2000 of The European Parliament and of the Council of 16 December 1999 on orphan medicinal products;
- Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02).

It is necessary to highlight that there are no the same laws in Ukraine till now, as well as definition of "rare diseases". There are no statistics of rare diseases. In the neurology we have just one mention in the Reference to Directive of MINISTRY OF HEALTH from 28.12.02 N502 [2] - diseases of CNS are subject to the state statistical reporting:

- Inflammatory diseases CNS - G00, G03, G04, G06, G08, G09;
- Epilepsy G40 - G41;
- Diseases of peripheral nervous system G50 - G52, G54;
- Cerebral palsy: G80.

THAT'S ALL.

Prevalence in Ukraine

While occurrence of rare disease is 1 case per 2000 healthy citizens it seems insignificant number, but for Ukraine with the population of 46 million citizens it could means 235 people for each rare disease (based on [3]). This statistics proves the facts in Ukraine.

According to the Ukrainian patients' organizations were registered or estimated:

- Spinal muscular atrophy from 100 to 300 patients (by data for 7 years);
- Cystic fibrosis up to 500 patients;
- MPS about 50 persons;
- Muscular dystrophy - less than 50 (the data has begun since 2010)

It is in accordance with the forecast [3].

According to Institute of molecular biology and genetics of Ukraine (Livshits L.A.):

- Number of the analysed families with clinical suspicion on SMA (2006 - 2011) - 285;
- Number of the analysed families with clinical suspicion on CF for 5 years (since 2006) - 235;
- Number of the analysed families with clinical suspicion on PKU for 5 years (since 2006) – 105.

It is in accordance with the forecast [3].

BUT, Distinctions and inconsistency of data are on the graph.

What is first - the concept OR the national program (strategy)?

To approach to the problem decision is possible in two ways:

1. Will of policy makers to recognize the existence of a problem and to create legislative base.
2. Creation of the national program (strategy) with financing support (as a rule, from the budget).

It is obvious that countries which do not undertake steps to solve a problem of rare diseases, a funding from the budget is impossible. The situation becomes complicated by world financial crisis. It is necessary to move on the way of legislative lobbying, then to execute the approved laws will be obligatory natural step.

The EU countries passed one of stages. A problem of the RDs was legislatively recognized (e.g. the point 1 above). While the Council Recommendations witness the acknowledgment of the need for improving the conditions of rare disease patients by the Member States, and indicate the directions for the development of health policies, the European Project for Rare Diseases National Plans Development (EUROPLAN), has elaborated its “guidelines and recommendations” to facilitate the definition, implementation and monitoring of National Plans or Strategies.

The process for designing a National Plan or Strategy for rare diseases may be significantly different among countries. Some of them already developed their second plan (e.g. France), other countries have their first plan (Spain, Portugal, Greece and Bulgaria) and other countries have just limited knowledge about the situation of people living with a rare disease on their territories. Still, following the Council Recommendations, each member state should establish and implement plans or strategies for rare diseases at the appropriate level by the end of 2013. This fact will obviously be coordinated with the precondition (so, see the point 2 above).

EXAMPLES of existing national plans and strategies in different European countries.

- THE BULGARIAN NATIONAL PLAN ON RARE DISEASES 2009-2013;
- THE FRENCH NATIONAL PLAN 2004-2008 and 2011-2014;
- THE GREEK NATIONAL PLAN FOR RARE DISEASES (2008-2012);
- THE PORTUGUESE NATIONAL PLAN FOR RARE DISEASES (from 2008);
- THE SPANISH STRATEGY FOR RARE DISEASES;
- IRELAND: The preparation for National Plan is planned to be launched in 2010, with the main elements of the Plan to be advanced in 2011;
- In POLAND, the Ministry of Health created in June 2008 the Rare Diseases Task Force;
- CZECH national strategy for rare diseases 2010-2020

The unique program - EUROPLAN

The European Project for Rare Diseases National Plans Development (EUROPLAN) is a three-year project of the Programme of Community action in the field of Public Health (2003 - 2008), which began in April 2008. The Council Recommendation of the 8th of June 2009 on action in the field of rare diseases states that EUROPLAN has the task of developing “guidelines and recommendations”. It has been agreed that Recommendations are to be meant as “guidance” for the development of National Plans or Strategies, implementing the contents of the main European documents on rare diseases and in particular the aforementioned Council Recommendation. Great importance has also been given to their use as a “toolbox”, in relation to the fact that the EUROPLAN recommendations provide a set of “tools and examples” of how activities for rare diseases can be organized at national (and European) level. In fact, the actions recommended in this document will be implemented and developed differently in different member states, based on the organization of the national health

and social system, on the population size of the country, on the availability of expertise in the field of rare diseases, on the integration with already existing initiatives and on budget issues.

An interaction of Patients' organizations with the EUROPLAN creates the perfect tool for influence on rare diseases as in social, ethical, and the scientifically-practical plan. In fact, our vision, that the EUROPLAN role can be wider than simple "recommendations", and should be directed to informing the First Persons of the countries which aspire to enter into the international cooperation about the RDs problem. Frequent meetings on high level in the EU is an occasion to pay attention of participants that close interaction from the EU cannot do without performance of one of humanity principles - recognitions of the right of the patient with rare disease on the adequate level.

Empowerment of Patients' Organisations

As a result of empowerment, patients with rare diseases have in many cases played an active and instrumental role in determining research projects and forming health care policy. In addition, empowerment may result in better management of the daily needs of patients and better compliance with care protocols, in coping with the associated psychological conditions and in improving social inclusion. Due to the large number of different rare diseases there are over 1700 different patients' organizations in Europe. They play an important role in offering information and to patients, raising funds for research and lobbying for better quality of care and treatment. Many of these people (patients and their relatives) are organized into national alliances, sometimes affiliated to European umbrella organizations, the most important of which is, by far, EURORDIS. Disease-specific websites run by patients' associations, are often very important sources of information frequently used by patients.

Spinal Muscular Atrophy – is a rare genetic disease affecting each of 6000 newborn. In 2004 parents of children with SMA created Kharkiv Foundation «Children with a spinal muscular atrophy» which unites parents from all Ukraine, providing with their necessary information and encouragement. The Foundation acts as the initiator of maintenance of priority in public health services and social life for patients with SMA. Carrying out its mission, the Foundation is the full partner of the parental both professional organizations of Europe and the USA [5].

In Ukraine the Foundation aspires to draw public to a problem of the RDs through a prism of spinal muscular atrophy, patrons and government officials to a problem of rare diseases, stimulate national debate among relevant stakeholders; aspires to raise knowledge of practical public health services and to push processes on problem RDs at the state level and through public health services bodies. As an example, in September, 2010 the information letter «About innovations in public health services system» with support of MINISTRY OF HEALTH of Ukraine in which questions on RDs (neuromuscular type) were published, doctors were informed on SMA, DMD united registration system.

Many countries have to face specific problems for developing health care policies for rare diseases. Indeed, many diseases are not present in the population or occur only occasionally. Moreover, the proportionately limited funds and staffing of the public health system might not allow a sufficient diversification of health services. Combined, these features result in lack of awareness in the general population; lack of advocacy and pressure on the health policy agenda; lack of attention towards the problems of rare disease patients; lack of on-site specialized health care professionals and centers of expertise for rare diseases; and insufficient research on rare diseases.

Despite the fact that threshold referring to rare diseases differs in different countries and ranges from 1:1500 to 1:2500 of the population, in Ukraine still there is a barrier of recognition of adequate number of patients which has RDs:

- in the U.S. - less than 1 per 200 000 affected Americans (based on Rare Disease Act of 2002) or 1 per 1 500;
- in Canada - at least 1 per 2 000 population;

- in Japan - less than 50 000 affected Japanese or about 1 in 2,500 people;
- Australia - less than 1 per 2 000;
- and the European Union - if it occurs no more frequently than 1:2 000.

What is the situation in Ukraine?

This summer, on public consultation there was Conception of State Program: "Health 2020: Ukrainian perspective" on 2012-2020". The main feature was total absence of a mention of rare diseases in the document context. The Foundation had been initiated a meeting of POs' and joint recommendations were developed for this document. Reaction from the Ministry is expected.

Construction of reliable communications and cooperation with the state structures demands the initiative from POs. Despite the fact that Ukraine isn't integrated in the EU, the Foundation aspires to represent Ukraine at international level by putting a keystone of prospect for the future. For the international community there are no political borders, each element which helps to solve a global problem of rare diseases is important. An example of how each voice is accepted, see «Open consultation of European Commission" in 2007 where the Foundation took direct participation [7].

Tools for work

- The international action The Rare Disease Day, which lobbying action at national and global level, provides the momentum in pushing for National Plans or Strategies;
- The EUROPLAN 2;
- National Registers for rare diseases. (Close cooperation of Foundation with the European network of neuromuscular diseases TREAT-NMD allowed creating the Ukrainian national Register of patients with SMA and DMD, becoming a part of the International registry.
- Social councils (since 2011 in Ukraine the public monitoring system of actions of state structures is reorganized - Social councils are created at regional administrations, the Ministries. The Foundation became a member of such structure at regional and national level that gives prospect to initiate questions of rare diseases).

CONCLUSIONS

Today, even that known rare diseases is still very limited, it is possible to ascertain that the public health services system undertakes certain practical actions. However these actions remain not coordinated and unsystematic in the conditions of absence of understanding of a way from the Concept to the Program.

However, we should despite of everything continue to struggle for patients with rare diseases ... The aim is to ensure that all patients with a rare disease in Europe have equal access to high-quality care, including diagnostics, treatments and rehabilitation.

The countries where there is no concept or the national program should recognize at legislative level principles and concepts of "RARE DISEASES».

President of Foundation Vitaliy Matyushenko

[1] Communication from the commission to the European parliament, the council, the European economic and social committee and the committee of the regions on Rare Diseases: Europe's challenges. Brussels, 11/11/2008 COM (2008 679 final).

[2] «Time specifications of granting of medical aid to the children's population in the conditions of out-patient-polyclinic institutions» (children's neurology and children's surgery). It is confirmed by order MINISTRY OF HEALTH of Ukraine of 28.12.02 N502, p. 120.

[3] Rare Diseases: understanding this Public Health Priority. EURORDIS, November 2005

- [4] Prevalence of rare diseases: Bibliographic data <http://www.orpha.net/>
- [5] KCF «Children with SMA». Gogolia street, 7, Kharkiv, Ukraine 61057. Registration Code 33288562. The certificate №14801200000004348, the decision of Executive committee of the Kharkiv City Council 25.11.04. Ph. +38-050-364-06-73 WEB www.csma.org.ua E-mail info@csma.org.ua
- [6] Information letter «About innovations in health protection system». MINISTRY OF HEALTH of Ukraine, the Ukrainian Center of the scientific medical information and patent work, based on the decision of problem commission MOZ of Ukraine "Neurology" № 9 14.09.10. Martinyuk V., Matyushenko V, Suhonosova O.
- [7] http://ec.europa.eu/health/ph_threats/non_com/consultation_en.htm#3 (R-004 Vitaliy Matyushenko President of Foundation Children with SMA UA 11/29/2007)
- [8] Recommendations for the development of national plans for Rare Diseases. Guidance document/Guidance_doc_europlan_20100601. Revision Date 8 Jul 2010.