# FINAL REPORT OF THE HUNGARIAN EUROPLAN CONFERENCE

# I. Content

## II. General information

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| Country | Hungary | |
| Place and date of the National Conference | Hunguest Hotel Griff*** (H-1113 Budapest, Bartók Béla u. 152.);<br>15-16. October, 2010 | |
| Website | http://sites.rirosz.hu/europlan | |
| Organizers | • RIROSZ (HUFERDIS)<br>• Rare Disease Centre, National Institute of Healthcare Audit and Inspection<br>• Rare Disease National Coordination Research Team of University of Pécs Rare Disease Expert Committee | |
| Members of Steering Committee | Dorica Dan (EU Advisor)<br>Prof. Dr. Béla Melegh (scientific representation) –University of Pécs<br>Gábor Pogány Ph.D. president of Rare Disease Association of Hungary(civil representation)<br>János Sándor Ph.D. (EUCERD expert) – University of Debrecen<br>Ildikó Szy expert, NEFMI (governmental representation)<br>Dr. István Vályi-Nagy, AIPM (Association of Innovative Pharmaceuatical Industry) ( industrial representation) | |
| Names and list of Workshops | 1Day: Plenary lectures | Why Hungarian National Plan is necessary for Hungary?<br>Recommendations of European Committee and European Council<br>Presentation of EUROPLAN Program and recommendations<br>Present situation of Hungary based on EUROPLAN indicators |
| | 1, Section | Methodology and Governance of a National Plan |
| | 2, Section | Definition, codification and inventorying of Rare Diseases |
| | 3, Section | Information and training |
| | 4, Section | Laboratory diagnostics, screening and early intervention |
| | 5, Section | Research on rare diseases |
| | 6, Section | Standards of care for RDs - Centres of Expertise / European Reference Networks |
| | 7, Section | Orphan Drugs and Provision of Treatment |
| | 8, Section | Patient Empowerment and Specialised Services |
| | 2. Day: Closing Plenary lectures | Summarizing of all discussions, 10 minutes summary of all results of each section. Discussions and acceptance of the final European Union report |
| | First evening: POLKA Forum | |
| Chairs and Rapporteurs of Workshops | Yann Le Cam, Gábor Pogány, József Mandl, György Kosztolányi, Zsuzsanna Molnár, János Sándor, Béla Melegh, Vargha Helga Süliné, László Gulácsi, László Szőnyi, István Bitter, György Pfliegler, György Fekete, Veronika Karcagi, Katalin Komlósi, Ferenc Oberfrank, Mária Judit Molnár, Sándor Túri, Imre Boncz, Magdolna Dank, Pál Vittay, György Harmat, Csapó Judit Váradiné, István Nagy, András Falus, Ildiko Szy, András Rádics | |

## III. Summary

The Hungarian EUROPLAN National Conference for Rare Diseases (RD) has been organized in the framework of the EUROPLAN project. 148 participants – patients, scientific experts, representatives of governmental and industrial bodies – took part in the conference, held in Budapest. There were two plenary sessions, eight working group sessions and two POLKA meetings to discuss the EUROPLAN Recommendations. This publication was prepared for two years be the experts of European Council (2009/C 151/02), other EU institutions, including the Hungarian experts of Rare Disease Centre and Rare Diaseases Hungary (HUFERDIS). The conference offered the possibility to become better acquainted with the recommendations of both the Committee and the Europlan project as regards mapping the Hungarian situation, determining current strengths and shortcomings, and appointing common priorities. The Europlan documents and relevant materials served as a good basis for achieving the above. Due to stringent guidelines, important questions could be answered, while familiarizing ourselves with the opinions and needs of each sector's representatives.

The jointly worked out proposals served as practical, useful and forward-looking tools for the establishment and implementation of the National Plan for Rare Diseases in local political circumstances according to EU recommendations.

Participants of the conference determined the following as main priorities:

1. That the National Centre for Healthcare Audit and Inspection (OSZMK) assemble a National Plan Organising Committee by supplementing the current expert committee with representatives of sectors such as government and industry. The Ministry should designate a competent, responsible Head of the expert committee, - authorised to make decisions - to lead the development of the National Plan.
2. That OSZMK initiate transparent accreditation and the listing of centres of expertise, hospitals, and laboratories working in the field of rare diseases. Rare diseases should be included in the health care and social care systems, currently under reorganisation, and take into account existing resources and their concentration, as well as eliminating parallelism and formalizing existing informal relations and determining patients' pathways.
3. That external quality control of the accredited institutions by an independent supervising body must be insured, using existing EU compatible regulations (accurate patient registration, coding, multidisciplinary care, patient satisfaction etc.)
4. That the OSZMK rare disease information webpage be updated with information on the development of the National Plan, therapeutic options, organisation of healthcare pathways for patients, EU harmonisation (indicators and bio banks etc.), training, appointment of reference centres, research, social services etc.
5. That, the Hungarian participation in relevant EU programmes (EUROPLAN, ORPHANET, e-RARE-2, EUROCAT, BORQOL, and EUNENBS) must be motivated according to the strategic interest of the country.
6. Those European tools for measuring invisible disabilities, such as fatigue and pain are adapted so as to allow patients living with rare and chronic diseases to make use of care, support and benefits available to children with special training and care needs. When modifying the tax system, it should be considered that, in addition to tax allowances provided for big families, families raising chronic and disabled children should also enjoy tax allowances.
7. That awareness campaigns be launched for both expert and mainstream audiences on the area of education, development and empowerment of patient organizations. The mapping of applied procedures is necessary in the authorities dealing with rare diseases.
8. That an organisational body be developed to maintain an information helpline (or use, train and expand the scope of the existing one), taking into account the quality control of information (trained

personnel, adequate information). Join the European free helpline number network (116), in order to draw on the resources of this network. The implementation of monitoring satisfaction is also required.

9. That a detailed, appropriately scheduled plan be drawn up, together with the names of responsible persons. A continuous review would also be necessary. A forum by sectors should be organised twice annually, in order to share experiences and outcomes.

## IV. Detailed report

At the beginning of each theme, the relevant recommendations of the European Council are quoted, followed by the indicators calculated to monitor the current situation as well as to follow future developments.

### 1. theme – Methodology and Governance of a National Plan

**1. Section:** Methodology and Governance of a NP
**Chairs:** Melegh Béla, Helga Süli-Vargha
**Date, place: 15 October** 2010. 13:00-14:30, Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*1. Establish and implement plans or strategies for rare diseases at the appropriate level or explore appropriate measures for rare diseases in other public health strategies, in order to aim to ensure that patients with rare diseases have access to high-quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan drugs, and in particular:*

*a) elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems;*

*b) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies for a comprehensive approach;*

*c) define a limited number of priority actions within their plans or strategies, with objectives and follow-up mechanisms;*

*d) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level in the framework of the on-going European project for rare diseases national plans development (EUROPLAN) selected for funding over the period 2008-2011 in the first programme of Community action in the field of public health (1).*

**EUROPLAN indicators:**

| Actions | Indicators | | Type of indicators | Answers |
|---|---|---|---|---|
| Development of regulations/laws | 1.1. | Existence of regulations/ laws that support the creation and development of a RD plan | Process | Not existing, not clearly stated |
| | 1.2. | National/regional (percentage of regions) | Process | Not relevant |
| Establishment of coordination mechanisms | 1.3. | Existence of coordination mechanisms | Process | An expert committee exists but without real tether for coordination and preparing National Plan |

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|---|---|---|---|---|
| | 1.4. | Existence of an expert advisory committee | Process | Exists but partly functioning |
| Establishment of an external evaluation of Plan/Strategy procedure | 1.5. | Existence of an external evaluation body/procedure | Process | No |
| Degree of comprehensiveness | 1.6. | The number of priority areas included | Process | Not relevant |
| Establishing of a budget for developing the plan/ Strategy | 1.7. | Budget of plan/ strategy | Process | No |

### 1.1. Current situation

**1.1.1 In Hungary, social a**wareness of rare diseases and the life conditions of people living with a rare disease is at a medium level. We note that the Day of Rare Disease, organised three times already with continual media coverage following the event, has made important steps towards increasing awareness of rare diseases, which was very low before this event.

**1.1.2.** Within OSZMK, documentation of official resources, activity programs and research currently operating within the national health care and social system are being developed; this work began in 2009. However, goals can only be achieved if they are compatible with the Orpha.net system, which is user friendly, with a registry that is refreshed on a continuous basis, and that is accessible to everyone. An informal network of researchers and clinicians exists and informs participants about actual programs and research (such as the Day of Rare Diseases or the national EUROPLAN Conference, which both give room for participants to monitor and become acquainted with existing activities performed in the field of diagnostics, care and research relevant to rare diseases. Examination of the needs of patients living with a rare disease is the aim of our participation in the EurordisCare 3 research and the refill of EurordisCare2 and the BURQUOL-RD program are also in process.

### 1.2. Strategy and structure of a National Plan

There is no legal frame program dealing with the health care and social needs of rare diseases patients currently in Hungary. Neither does there exist a special activity program with the aim of developing a national plan or strategy in this area. In 2008, within the frame of OSZMK, the Rare Disease Centre was set up (RBK). Its task is to coordinate the informally appointed regional centres of expertise. This body does not have the authority to develop a National Plan. The first productive step toward this goal was the active participation of each sector in the EUROPLAN conference.

### 1.3. Management of a National Plan

Currently there is no appointed Organizing Committee, state or governing body to coordinate or implement the Plan. The professional body of the above mentioned RBK would be competent to undertake this task; however, this would require extending its authorisation as well as involving the representatives of the pharmacy and the governmental sector into the process.

**1.4.National Plan monitoring:** This part is not relevant because of the absence of a National Plan

### 1.5. National Plan Sustainability:

Regarding lack of a National Plan, the question is not relevant. However, we emphasise that the issue of rare diseases should be adapted into the present, on-going reorganisation of the health care and social care system. By rationalisation and reorganisation of the current system, a great number of issues could be solved without using extra resources (i.e. concentration of national resources, elimination of parallelisms, making informal relations into formal). In the long term, however, the assignment of separate financial resources within the state budget is needed to ensure the sustainability of the Plan.

### 1.6. Recommendations

1.6.1 It is necessary to increase the awareness on rare diseases in the national health care and social care systems by setting up registries on the available resources, activity programs, research (i.e. by turning initials of the OSZMK webpage to be useful and functioning. The first steps of achieving this goal could be realized, none the less, by setting up an Orphanet compatible Hungarian registry, through the integration of the Orphanet registry, by establishing a compatibility with the existing Hungarian registries, as well as by insuring the availability of the whole Orphanet database in Hungarian.

1.6.2. Support of the current international research programmes (**EurordisCare2, Burquol-RD)** on the needs and life quality of RD patients through the integration of the patients as well as their representatives is crucial.

1.6.3. **Legal frames** must be developed to solve the heath care and social needs of RD patients, as well as a special program in order to set up a National Plan and strategy.

1.6.4. The Ministry should appoint and - if needed- enlarge the **organizing committee** responsible for the development and implementation of the plan. Participation of each stakeholder must be ensured, such as health care authorities, patients, medical experts, researchers, representatives of the industry etc. These participants could cover all the affected territories, i.e. pharmaceutical industry, state, clinics, health care and social care services, epidemiology, administration etc. It is necessary to nominate a responsible coordinator as well as an independent external supervisory committee.

1.6.5 The organizing committee should meet regularly (at least 3-4 times a year). At least once a year, a public report should be produced on the committee's activities, on the outcome of its goals and on the activities of persons with key responsibilities. This report could be released on the International Rare Disease Day and could be available later on the webpage of RBK.

1.6.6. When working out and practicing the National Plan, the Europlan indicators and the research on evaluating patients' satisfaction could be used as a basis for evaluating and controlling the implementation of the NT itself. This review must be done by a **separate, independent supervisory body**.

1.6.7. The currently actual health care reform must integrate the organisation **of RD patients' care based on the European examples.** When implementing the prioritised activity programmes, a special budget should be assigned to each of them. (i.e. orphan drugs, centres of excellences, availability of diagnosis, research etc.) A separate and transparent budget must be made available for these purposes.. In order to ensure the long term sustainability, this budget should not be an addition, but may come from the reorganisation and rationalisation of existing resources and from the elimination of redundancies.

1.6.8. **Introduction of a Rare Disease plastic card for RD patients.** At a later stage, services could be assigned to a bare-coded, plastic chip card. At the time of its introduction, it would be practical to ensure, that patients and their relatives could recover the travelling costs to examinations, controls from National Health Insurance Fund (OEP). An 80% salary allowance could also be introduced for the frequent one day examinations. In order to achieve cost and time effectiveness, RD centres should centralise the necessary examinations. In the case of those diseases where it is known that the active care protocol will exceed three months, the RD patients or their parents should be eligible for temporary care support, equal to the normal actual minimal salary. Parents and relatives raising RD kids, when on care support, should be eligible to all the allowances that are provided to parents living on maternity benefit (GYES) and child care fee (GYED), such as travel allowances and (small) children's season tickets.
The activity of parents and relatives on care support should be acknowledged as working time, as this would relieve the state from raising these children in institutions; this work undertaken by parents should be estimated according to the normal minimal salary. It goes without saying that this system must be closely controlled to insure that the parent/or relative does not abuse these supports for anything other than the interests of the RD patient. Methods of monitoring and control should be detailed taking into account the personal rights and the interests of the financing body in a way consistent with controls used in other cases where financing comes from public sources.

## 2. theme – Definition, codification and inventorying of Rare Diseases

**Chairs:** László Gulácsi, László Szőnyi

**Date, venue: October 15,** 2010, 13:00-14:30, Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*2. Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.*
*3. Aim to ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD while respecting national procedures.*
*4. Contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks as referred to in the Commission Communication on rare diseases.*

**EUROPLAN Indicators:**

| **Actions** | **Indicators** | | **Type of indicators** | **Answers** |
|---|---|---|---|---|
| To officially adopt the EC RD definition (no more than 5 cases/10,000 inhabitants) | 2.1 | Adoption of the EC RD definition | Process | Yes |
| To include the best RDs classification currently existing into the public health care related services | 2.2. | Type of classification used by the health care system | Process | ICD-10 |
| | 2.3. | Developing policies for recognising RD by the care information systems | Process | Not existing, not clearly stated |
| Defining a surveillance system based on a patient outcomes registry | 2.4. | Registering activity | Process | Multiple RD registries, not standardised |
| | 2.5. | Number of diseases included | Outcomes | 26 |

### 2.1. Definition of the rare disease (RD)

Presently the EU's official definition functions as the accepted definition, with a rare disease defined as one in which there are no more than 5 cases/10,000 inhabitants. Unfortunately awareness of and acceptance of rare diseases is not as consistently shared in different national contexts.

### 2.2. The codification and follow-up of the RDs within the national healthcare system

**2.2.1.** In Hungary the ICD10 codification system is applied.
**2.2.2.** **The codification system** is used for the registries and the normative **cost refund.**
**2.2.3.** Currently there are no **aimed measures** for introducing the new (ICD11) system which will be ready for 2014 by the WHO and which is recommended by the Council for the rare diseases. At the same time the OSZMK's Congenital Anomalies National Surveillance Department, as a member of the European Surveillance of Congenital Anomalies (EUROCAT) fulfilling the organization's request, has already adopted the recommendation for the modifications of the ICD 11 version. The professional body concerns the future appliance of the ICD 11, as an evident.
**2.2.4.** **The level of the health experts' consciousness and knowledge in the territory of the RDs' classification and codification** can be measured adequately given that,, less than 300 RDs have their own ICD code. At the same time the codification is the basis of the claim to a normative subsidy, and often the physicians have opposing interests and prefer not to use the codes belonging to the exact diagnosis for reaching better financing through symptomatic treatments.

### 2.3. Recordings, registers and lists

**2.3.1.** There is a register, the National Register of the Congenital Anomalies (VRONY), which is already overlapping with the future RD recordings. The physicians are obliged to report on each case concerning children younger than 1 year according to the Act no. XLVII. from the year 1997. (Public Wealth Bulletin, 1997. 12. 1722-24'). These data can be requested officially, taking into consideration the protection of personal data. The reporting system had been paper-based for a long time, so the data processing was a difficult task. As of October 2009, the reporting of congenital anomalies has been conducted electronically. The European Surveillance of Congenital Anomalies (EUROCAT) recommends on-line reporting for all of the associations (until now, other than Norway, Poland and Wales, Hungary uses an electronic reporting system). The VRONY can be judged as a favourable modification not only for financial reasons but for its up-to-date status too. Additionally, several years can pass until a correct diagnosis of a RD is found or, in some cases, symptoms can emerge only later; consequently, reporting liabilities are not as relevant for such cases. At the same time Paragraph 10 of the Act mentioned above in connection with VRONY, makes possible the forwarding of data: "In case of data management within the healthcare service network the health and personal data can be forwarded and associating (till that time and level which is absolutely necessary for serving the required measures)". Independently from this, there is a need, so the OSZMK initiated the establishment of an overall RD registration, but this program is only under way.

**2.3.2.** Currently these registers and programs are not separately supported by the government.

### 2.4. Recommendations

2.4.1. The use of the official EU's RD definition (≤5/10000) should be compulsory for alleviating the international co-operation and the community-level acts (e.g. co-operation at diagnosis' establishment, treatment, caring and registration).

2.4.2. The use of the RDs' common EU registration (Orphanet) should be supported within the national health care service systems, and it should be co-operatively and continuously updated.

2.4.3. The suitable codification of RD should be supported by careful consideration of the financing mechanism, by introduction of the use of the ICD11 and Orpha codes to be expected by 2014, prepared by experts for ensuring follow-up possibilities within the national healthcare service systems.

2.4.4. The cross-references between the different classifications systems used in the country should be ensured for harmonization and synchronization with such European initiations as the Orpha-code system.

2.4.5. The co-operation within the ICD 10 supervision process should be ensured for making possible the immediate application of the ICD 11, when it is ready.

2.4.6. The healthcare system should have perfectly trained experts for the recognition and codification of the RDs.

2.4.7. The integrated use of the administrative, demographic and healthcare service systems' data should be supported on the national level by adequate measures for the right management of the RDs. The knowledge concerning the EU-compatible establishment and management of the healthcare registrations and registers, the appropriate codification should be involved into the healthcare-informatics experts' education.

2.4.8. The healthcare authorities have to encourage data collection and circulation and healthcare purpose accesses from all authentic sources, included the experts' centres to be established, according to national regulations.
The patients' organizations and the individual patients should also be involved in a healthcare data sharing system; that is, the data access should be ensured for them, likely as the National Health-Insurance Fund's (OEP) patient-way's supervision system. The patient, who provided any data for the system must have the right

to access the system, hereby to check the correctness of the data, and to compare his own data with the summarized data and statistics to be obtained from the system.

2.4.9. In each reference centre an adequately trained person should be employed who must be able to manage the registrations and the registers' data-basis based on the codifications.

2.4.10. Promote the presence of the national registrations in the existing European/ international registrations.

2.4.11. Identify the means for combining the registers' EU and national financing possibilities.

2.4.12. For researching, curing and preventing purposes, encourage and support the special RDs' and RD-groups' international, national and regional systems, associations, including the entities maintained by the researchers too.

## 3. theme – Information and training

**3. Section:** Information and education in Hungary

**Chairs:** István Bitter, György Pfliegler

**Date and place:** 15 October 2010. 15:00-16:30, Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*5. Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance.*

**EUROPLAN indicators**:

| Actions | Indicators | | Type of indicator | Answers |
|---|---|---|---|---|
| Existence of a information sites for both professionals and patients provided by the plan/strategy | 10.1. | Existence of a comprehensive national and/or regional RD information system supported by the government | Process | Yes, covers only some RD |
| | 10.2 | Help lines for professionals | Process | Not formal decisions have been taken |
| | 10.3 | Help lines for patients | Process | No |
| | 10.4 | Clinical guidelines | Outcomes | 17 |
| Promoting training activities and awareness educational campaigns among professionals and patients | 10.5 | Number of such as activities promoted by the plan/strategy | Process | Not relevant |

### 3.1. Facilitating the access to information for all interested parties on the services available to RD patients

**3.1.1. The existing sources of information and their quality**

**3.1.1.1. There is one official website maintained by the government**, under the link www.oszmk.hu, however it is only a few pages and its information content is very limited. It would be very important to make it more user-friendly and to extend its content with the following: links to patient organisations, list of institutions dealing with rare diseases, scientific associations, and research in progress, databases, recent events and news. The website that has the widest coverage is the site www.rirosz.hu together with its subsidiary sites, all maintained by non-governmental organisations.

The use of the major international websites (e.g. orpha.net; NORD etc.) is fairly occasional.

There are articles published on certain medical and pharmaceutical websites, which deal also with rare diseases, but these are quite rare, depending on the ability of civil organisations to promote their interests: e.g. www.webdoki.hu, www.patikamagazin.hu, www.medicalonline.hu. There is also a non-professional site with collection of links: http://ritkabetegsegek.lap.hu/

**3.1.1.2. There are no help lines specialized in rare diseases**. Most of the patients find proper information meeting their special needs (e.g. healthcare pathways, social care, advice on daily life issues, mental aid, etc.) through searching for patient organisations' websites or calling the patient organisations themselves. However, these lines are not toll-free, it is serendipity whether the relevant patients are able to find them, and they are fully sustained by non-governmental organisations, and are disease specific. There is no government call centre trained on rare diseases that would facilitate the access to the European number 116.

**3.1.1.3.** Currently there are no inspiring initiatives or programs for the development of informative and educational materials targeted to patients or to other groups of special interest (teachers, social workers, etc.).

**3.1.1.4. We make use of the information sources already existing at the European level (e.g. ORPHANET, EURORDIS)**, e.g. in case of clinical studies, research projects, etc. The registration of institutions and experts has not been done yet, except for a few cases (e.g. the registration of the biobank in Pécs for the E-RARE2). The involvement in the on-going research projects and clinical tests is incidental; often it happens through the mediation of the patient organisations with activities on the international level. The representative of EURORDIS in Hungary is HUFERDIS.

**3.1.1.5. There are initiatives to raise the public awareness related to rare diseases**, but mostly from non-governmental sources.

a) The Rare Disease Day has been organized by HUFERDIS since 2008 (in cooperation with member organisations and professional organisations). On top of this, since its formation and since its joining to EURORDIS, HUFERDIS have been participating also in the other projects coordinated by EURORDIS (e.g. POLKA, EUROPLAN, BURQOL, EurordisCare 3). In addition, HUFERDIS and its member organisations, of which there are 35 by now, are regularly present at conferences, family days and as part of other programs where they raise the public awareness through information materials and through access to personal consultation.

b) Media interest for rare diseases has been continuously growing, particularly in relation to the Rare Disease Day.

c) There is more and more mentioning of rare diseases in the basic education, postgraduate courses, conferences and publications for medical experts, but it is necessary to further increase the effectiveness and intensity of these. The same is yet completely missing from the education of teachers and of social workers.

**3.1.1.6.** These programs are promoted and financed mainly by the patient organisations. Governmental support is incidental and is only through tenders.

### 3.2. Development of availability of quality information on rare diseases

**3.2.1.** These information sources and initiatives are publicized only through the lobbying activities, media presence, newsletters, information events, websites and conferences delivered by non-governmental organisations yet.

**3.2.2.** On top of the lobbying activities of non-governmental organisations it is necessary to provide official and controlled information to the target group and to the wider public.

### 3.3. Provision of appropriate education and training to health professionals in relation to rare diseases

**3.3.1. The education to health professionals in relation to rare diseases currently includes the following:**

- Information about the existence of rare diseases and about the resources available for their care: partially;
- Medical training in fields relevant to the diagnosis of rare diseases (genetics, oncology, immunology, neurology, paediatrics): partially;
- Further education for young doctors and scientists working in the field of rare diseases: partially;
- Exchange and sharing of expertise between the centres of expertise in the country: partially.

Examples:

a. Courses on Rare Diseases (graduate, postgraduate) Debrecen, Department of Rare Diseases, since 2003, annually, 40-100 participants;

b. Days of Internal Medicine of Debrecen (so far seven times), in the frame of levelling courses for specialist doctors and candidates, annually, 2-3 hours within the seven days, 100-150 participants;

c. Family doctor education: one hour lecture per year (Debrecen)

d. Other conferences (e.g. Pécs, 2007, 2008), Conference of the Hungarian Society of Human Genetics (Debrecen, September 2010), etc.

**3.3.2. The EU directives are implemented and applied** by the respective deadline (e.g. cross-border care, genetic tests, etc.). The introduction of treatment protocols is accidental; often it is the patient organization that informs the experts about these.

**3.3.3. The sharing of the expertise existing on the EU level** and the access to adequate trainings is supported via OSZMK *(National Centre for Healthcare Audit and Inspection)* and ÁNTSZ *(National Public Health and Medical Officer Service)*, so that it reaches all medical experts.

**3.3.4.** Hungary is supporting the participation of local experts in the development of international guidelines that should help the diagnosis and care of rare diseases on the national level. For example, there are two official delegates to EUCERD, one to the development of ICD11, and one to the Orphanet program.

### 3.4. Recommendations

3.4.1. The OSZMK website on rare diseases should be made more up-to-date, extended with relevant pieces of information, made more user-friendly, and be further promoted. As part of this development, an information database should be created, in harmony with the structure and objectives of the Orphanet database, in Hungarian language and – if possible – on the original Orphanet website.

3.4.2. A new information help line should be established (or an existing one extended and trained), considering also the quality assurance on the information provided (properly trained experts, adequate information). Join to the European toll-free help-line (116) network and reapply its experiences.

3.4.3. The correct identification of healthcare pathways in the hospitals should be secured through the training and involvement of paramedical experts and social workers working in the centres of expertise (that are to be appointed) and through the provision of the information materials of the patient organisations to those concerned.

3.4.4. The preparation and circulation of information materials on rare diseases should be financially supported – this could even include the setting up of a 'dedicated fund' by the relevant authorities, the provision of which should be seriously considered by government offices. Nevertheless the needs of the patients would be better addressed through a support system that is transparent and is independent from the government (like the NCA for example *(National Civil Fund)*), as opposed to either a dedicated fund with unclear principles of fund allocation or the direct support through commercial entities that might not guarantee equal treatment on ethnic, religious, political or social grounds. Therefore it is recommended that the tenders of the NCA are extended in this direction, because it could support patient organisations (including

even the smaller ones) in their provision of information materials, social studies, contact building and information exchange with foreign country and international umbrella organisations, and even their professional conferences.

3.4.5. There should be presentations/lectures in high schools and in the universities calling attention to rare diseases. Raising awareness about the life, opportunities, difficulties and the socially useful activities of the people suffering from rare diseases should include the promotion of solidarity and respect for differences between people.

3.4.6. There should be more regular and more predictable presence in both the public and the expert media (e.g. mandatory column, social solidarity programs)

3.4.7. **There should be more opportunities secured for the patient organisations to appear on the mandatory vocational trainings and on the relevant conferences, etc. (e.g. poster, presentation, brochure)**

3.4.8. The support coming from commercial entities to the patient organisations should be encouraged, e.g. through the provision of tax credits – this way there would be additional funds generated for the purposes of education and information provision. We request that donations should again be made tax free and that the donors receive a tax benefit in same way as they did previously.

3.4.9. The rare diseases that are relevant to certain medical specializations should be addressed in the mandatory levelling courses of those specializations - i.e. not only in the courses of internal medicine.

3.4.10. The database for diagnostic options in Hungary should be created (indication, type of sample, price, location, time needed for the diagnosis), and made available on the OSZMK website.

3.4.11. It should be made clear that 20% of rare diseases are not genetic diseases, but are from different cause (e.g. infections).

## 4. Theme – Laboratory diagnostics, screening, and early intervention

**4. Section:** Laboratory diagnostics, screening, early intervention

**Chairs:** György Fekete, Veronika Karcagi

**Date, venue:** October, 15, 2010, 15:00-16:30, Hunguest Hotel Griff***

### Council Recommendation (2009/C 151/02)

17. Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support:
(a) the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases;
(b) adequate education and training for all health professionals to make them aware of the existence of these diseases and of resources available for their care;
(c) the development of medical training in fields relevant to the diagnosis and management of rare diseases, such as genetics, immunology, neurology, oncology or paediatrics;
(d) the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;

**EUROPLAN indicators:**

| Actions | Indicators | | Type of indicator | Answers |
|---|---|---|---|---|
| Develop screening policies | 8.1 | Number of diseases included in the neonatal screening programme | Outcomes | 26 |
| | 8.2 | Number of diseases included in the neonatal screening programme properly assessed | Outcomes | 26 |
| Ensure quality of RD diagnosis laboratory | 8.3 | Existence of a public directory/ies of both genetic tests on Rare Diseases | Process | Under discussion |
| | 8.4 | Proportion laboratories having at least one diagnostic test validated by an external quality control | Outcomes | 20 % |

### 4.1. Access to the adequate diagnosis

Presently, the RD's diagnostic can be implemented in the indicated diagnostic centres and the genetic consultative centres. The time elapsing for the first visit is averagely 4.9 months, which is can be characterized as too long.

The EuroCare2 survey, which is can be served further data to the issue, is already on way. The new-born babies' screening program contains 26 compulsory examinations, mainly in case of metabolism diseases, for which examination two centres, have been assigned in the country.

### 4.2. Genetic diagnostics and communication on genetic results

For this issue the professional protocol of the Ministry of Healthcare, the document titled as Genetic Consultation, and prepared by the Genetic Professional College in 2004, is the basis. This defines the conditions necessary for supplying the laboratory background, the infrastructure, the personal conditions, the personal/operational costs. It provides a transitional period for supplying all of these conditions. It is problematic that these centres and laboratories have been established up and down in the country without any method, randomly. Without a national registry, these centres and laboratories are not known and are not as visible as they need to be.

### 4.3. Quality-control

The supervision of the accredited laboratories is performed regularly by foreign accreditation bodies.

### 4.4. Financial support and sustainability of the centres

Presently these institutions do not get separate financial aid from the state budget. So it definitely requires regulation, development and operation of the institutional system and development programmes. Furthermore, it is important, that the operating centres become known (central homepage) and reachable, for ensuring the most effective marking of the patients' ways. The patients' interest is that the earliest possible diagnosis, the treatment and the development could be in one place, near to their residences.

### 4.5. Establishment of diagnostic registries

There are positive European examples: e.g. the EuroGentest, Orphanet, Gentests which contain easily accessible data: description of the diseases, lists of the diagnostic laboratories, quality insurance, registers of patients' organizations, databases, experts, but unfortunately in Hungary there is no adequate register (the existing one contains obsolete data), there is no Orphanet representation.

### 4.6. Recommendations

4.6.1. The earliest possible implementation of the **Orphanet registration** at OSZMK level. But before the foreign harmonization there is a need for the Hungarian harmonization.

4.6.2. The earliest preparation of the **national genetic examinations' register**.

4.6.3. Further education of the experts, improvement of the information's accessibility for exact assigning of the **patients' way** following the diagnosis (social assistance's possibilities, civil organizations, mental hygienic assistance – not the patient is who had to search for the diagnostic possibilities).

4.6.4. **Taking over**, adaptation and following of the already existing **diagnostic protocols**, spreading of the national experiences.

4.6.5. **Improvement of the genetic examinations' accessibility** with rationalization of the existing capacities, with provision of separate state budget's aid.

4.6.6. Providing a **cross-border healthcare service**, providing the OEP's financing for those necessary cross-border examinations which are not accessible in Hungary. The simplification and speeding of the assistance mechanisms have to alleviate all of these tasks. The competence of the professional college has to spread on the decision that laboratories can receive the samples with OEP's financing, and the determination of the yearly contingent.

4.6.7. The laboratories and the advisory centres have to be **accredited based on the international standards**, by promoting the participation on the international round-examinations.

4.6.8. The quality controlled molecular and cytogenetic procedures have to be available, furthermore the professional supervision system for controlling the laboratories, with definition of the competence levels and with the involvement of the Clinical Genetic Professional College & the Hungarian Scientific Academic's Physician's Department have to be attainable too.

## Theme 5 - Research on RD

Workshop 4: Research on RD
Chairs: Katalin Komlósi and Ferenc Oberfrank
Date, time and place: 9:00-10:30, 16 October 2010. 2010.10.19. Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*III. Research on rare diseases*
*(6) Identify on-going research and research resources in the national and Community frameworks in order to establish the state of the art, assess the research landscape in the area of rare diseases, and improve the coordination of Community, national and regional programmes for rare diseases research.*
*(7) Identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them, and promote interdisciplinary cooperative approaches to be complementarily addressed through national and Community programmes.*
*(8) Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.*
*(9) Include in their plans or strategies provisions aimed at fostering research in the field of rare diseases.*
*(10) Facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases and more generally with regard to the exchange of information and the sharing of expertise.*

**EUROPLAN Indicators:**

| Activity | Indicators | | Type of indicator | Answer |
|---|---|---|---|---|
| Building a research programmes for RD | 3.1 | Existing a RD National/Regional research programmes | Process | *RD research programme included in the general research programme as a priority* |
| | 3.2 | RD research programme monitoring | Process | *Existing, clearly stated, partly implemented* |
| | 3.3 | Number of RD research projects approved by year (if possible yearly starting the year before plan commencement) | Outcomes | *There isn't any independent database* |
| | 3.4 | Clinical trials funded by public bodies | Outcomes | *No actions have been taken* |
| | 3.5 | E-RARE joining | Process | *In process* |
| | 3.6 | Including public health and social research, in the field of rare diseases | Process | *Yes, partly implemented* |
| | 3.7 | Research platforms and other infrastructures are also funded by the research programme | Process | *Yes* |
| Existence of national policy in support of the recruitment of young researchers/ scientists specifically for rare diseases | 3.8 | Number of young scientists recruited every year to work specifically on rare diseases | Process | *There is a registry but we have no exact data.* |
| Allocate funds for the RD research programme | 3.9 | There are specific public funds allocated for RD research | Process | *No* |
| | 3.10 | Funds specifically allocated for RD research actions/projects per year since the plan started | Outcomes | *Not relevant* |

### 5.1. Mapping of existing research resources, infrastructures and programmes for RDs

**5.1.1.** Mapping of existing research sources and infrastructures of frontier fields has not been carried out yet. Therefore, evaluation of public and private founding prospects is not performed either.
**5.1.2.** At present, there is no special national RD research program based on separate source; however there is no obstacle to start such a program in principle.
**5.1.3.** At present, most of research projects investigating quality of life and needs of patients in Hungary are initiated by Eurordis through HUFERDIS. Regarding basic, translational and clinical public health research, interest and capacity of university clinics and pharmaceutical industry is determinant. In these cases, civil organizations take part in the recruitment of patients, in the development of research protocol and in sharing international results.

### 5.2. Measuring requirements and priorities concerning the basic, clinical, translational and social researches

In the field of social research, mapping of priorities is attained mainly through Eurordis projects (EurordisCare2,3, Burqol-RD)., A workshop of E-RARE2 programme deals with the mapping of priorities by international questionnaires in 2011, concerning basic, translational and clinical research.

### 5.3. Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects

5.3.1. Linking of basic and translational research to CoEs is not yet regulated..
5.3.2. Realization of an overall interdisciplinary approach in research is incidental and depends on the actual research project.
5.3.4. Regarding research in the field of social sciences, joint organizations o HUFERDIS show active cooperation and HUFERDIS also has a strong mobilizing force to the joint organizations.
5.3.5. There are no special programs for supporting and recruiting young researchers but general opportunities are available in the field of RDs (ex. scholarships, study-tours).

### 5.4. EU collaboration on research on RD

There are collaborations, however, encouragement and broadening is needed in the fields of basic, translational and clinical public health research, especially:

- BioBank Pécs joined E-RARE
- Corvinus University: BURQOL-RD program
- DEOE: EurordisCare2
- EU supported „Joint Action on EUROCAT" project of National Centre for Healthcare Audit and Inspection contains epidemiology research programs regarding rare diseases (mainly syndromes).

### 5.5. Recommendations

5.5.1. Special national research programs concerning RDs are necessary (basic, translational, clinical, public health and social), and these should be supported from founds dedicated to these fields, possibly on the long run. However, not a transformation but an improvement and sustainability of present research founding system could move this filed forward (more RD tender from the Medical Research Council - ETT).
Research tenders related to RDs should be made identifiable and traceable within the wider national research programs.

5.5.2. In defining research priorities and in the transparency of on-going research, a constant interaction between researchers and patient organization is needed.

5.5.3. Patient organizations should be regularly updated about recent research and their results, ex. research centres invite patient organizations to their scientific councils.

5.5.4. Following EU examples, RD related tenders should also originate from patient organizations (national, or international tenders), presuming that a financial basis is available.

5.5.5. National Plan or Strategy should include contribution for the cooperation of CoEs and /or other public health structures, health and research authorities in order to broaden knowledge about different aspects of RDs ex. founding of CoEs through tenders.

5.5.6. National networks should be motivated to investigate RDs. Special attention is needed in the fields of translational and clinical research in order to facilitate application of new knowledge in RD therapy. In the same time, registry of research teams working on RDs should be developed.

5.5.7. Technical platforms and infrastructure including clinical, social and health industry research related to RDs should be developed and supported in order to improve everyday care of patients with RD. A possibility of cooperation between public and private services should be investigated.

5.5.8. Multidisciplinary national and international research should be promoted to reach a critical number of patients for clinical tests and to use international professional knowledge.

5.5.9. Specific programs should be initiated to support and recruit young researchers working on RDs. To motivate professional after growth (of medical and paramedical professionals) and young researchers for studying RDs, a special scholarship found should be established (following EU example, based on either governmental or public funds).

5.5.10. Survey of new purpose use and new combinations of existing drugs should be promoted by specific clinical research tenders because it could be a cost effective way of the development of RD treatment.

5.5.11. Appropriate initiatives should be formed to foster participation in international research enterprises related to RDs including EU framework and E-RARE. National support of these initiatives should be consistently raised. Hungary has become a member in E-Rare2 project (through Centres of Expertise Pécs).

5.5.12. Specification of separate source of support is needed for patient organizations to join EU research.

5.5.13. Access to EU projects and information should be guaranteed for national research centres.

## Theme 6 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)

Workshop 6: Centres of Expertise and European Reference Networks for rare diseases
Chairs: Mária Judit Molnár and Sándor Túri
Date, time and place – 11:00-12:30, 16 October 2010. Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*(11) Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.*
*(12) Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.*
*(13) Organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts and exchange of professionals and expertise within the country or from abroad when necessary.*
*(14) Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.*
*(15) Include, in their plans or strategies, the necessary conditions for the diffusion and mobility of expertise and knowledge in order to facilitate the treatment of patients in their proximity.*
*(16) Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.*

**EUROPLAN Indicators**

| Activity | Indicators | | Type of indicator | Answer |
|---|---|---|---|---|
| Improve the quality of healthcare by defining appropriate centres with experience on RD as well as pathways that reduce the diagnosis delay and facilitate the best both cares and treatments to patients | 4.1. | Existence of a policy for establishing centres of expertise at the national/regional level | Process | Not existing, not clearly stated |
| | 4.2. | Number of centres of expertise adhering to the policy defined in the country | Outcomes | 0-Officially, approx. 8-informally |
| | 4.3. | Groups of rare diseases followed up in centres of expertise | Outcomes | Covering all or most of rare diseases |
| | 4.4. | Centres of expertise adhering to the standards defined by the Council Recommendations paragraph d) of preamble | Outcomes | 0 % |
| | 4.5. | Participation of national or regional centres of expertise into European reference networks | Outcomes | Approx. 10 % |

### 6.1. Identification of National or Regional centres of expertise throughout Hungary by the end of 2013

**6.1.1.** At present, owing to a general lack of information, access of the centres for patients is occasional and pathways are often informal through civil organizations or their homepages. In the past years, there was an improvement due to the raising awareness, the activities of national Rare Disease Centre (RBK) as a part of the National Centre for Healthcare Audit and Inspection, and also the trainings and conferences of the centres and the growing media interest. Health care in adulthood is exceedingly difficult and a main issue even in those successful patient groups where a child centre is organized because rare disease centres for adults are very scarce. Most adults visit paediatricians in an informal way leading to an overload and a transfer of costs to that side. Therefore, the organization of management, treatment and care of adults with rare diseases are especially needed. Until then, health care of adults should be legalized in paediatric practice and consultation, special knowledge and equipment tailored to adult needs should be available with appropriate accounting of National Health Insurance Found points. This could be a temporary solution until the organization of an adult centre. In the same time, specialists of paediatric practice should be allowed to involve adult specialists if needed. We suggest that the centre of National Centre for Healthcare Audit and Inspection could help in working out its professional details.

**6.1.2.** **The RDTF (RD Task Force) criteria** that are likely to be included in the 15 paragraph of the EC regulation of cross border health care regarding the function of Centres of Expertise, can be realized by the official identification of centres of expertise and own budget allocation for the extra tasks.
A HURO-euro programme plan is in process for a number of Hungarian centres in which Universities of Szeged and Debrecen jointly with Romanian counties wrote a tender for the establishment of centres of expertise in Romania and the development of them on both sides.

**6.1.3.** Since these are the largest University centres they are involved education, research as well as in medical attendance and function in the above fields as centres of expertise after task distribution. At the development of the centres we highly regard multidisciplinarity, research and educational activity, beside the clinical work and also the opportunities of social care. This way it can be ensured that the centres become experts of a given rare disease both in the clinical and research sides.

**6.1.4.** The four university centres provide regional service which should be coordinated. It postulates continuous communication and the coordination of all the centres which could be managed by RBK.
For this, the exact mapping of the activities and capacities, a public report and regular meetings of the RBK is needed.

### 6.2. Sustainability of CoEs

**6.2.1.** The founding of CoEs of rare diseases should be accentuated covering diagnostics, therapy and care.
In the field of diagnostics, beside the costs of examinations and tests costs of amortization of the equipment should be taken into account when calculating the budget. In every case, CoE teams should have a member who is responsible for the operation of registers and biobanks apart from medical doctors, psychologists and physical therapists.

Point of human genetics has been significantly reduced in favour of microbiological-genetic examinations; therefore, examinations in the field of human genetics became incapacitated. Cover for metabolism tests defray only the actual costs of the tests but do not defray the maintenance of equipment and the costs of chemical agents. Therefore, it is feared that the present equipment park is going to be amortized in the next 1 to 2 years. Purchase of mass spectrometer, gas chromatograph, light-cycler and sequencing system is impossible from the university centres' own resources. Beside the 26 screening tests, metabolism centres also wish deal with the investigation of genetic background of MPS or other metabolic diseases. Without a proper financial background it is insoluble. Amortization of the present equipment park endanger the current examinations, its sustenance needs an own budget. In some neighbouring countries this problem was resolved by privatization (ex. Czech Republic). Until then, human genetics and microbiology should be fully separated and appropriate conditions should be applied for them respectively.

### 6.3. Participation in European reference networks

**6.3.1. Fostering the participation of centres of expertise in European reference networks**
The identification and the consequent extra budget of National and Regional CoEs should assume the obligation of registration in E-RARE, orpha.net and reference centres of the attended diseases. The optimal conditions of national CoEs and the conditions of admission to Orpha.net network should be ensured by accreditation processes and participation in international research. Namely, the condition of participation is the continuous accreditation of laboratories and workstations and participation in international conferences-

**6.3.2. Fostering transnational mobility of patients and experts**
At present, the mobility of patients and experts applies only when a therapy or a diagnostic method is not available in Hungary. It works through a far too lengthy application process to the National Health Insurance Found.

### 6.4. Shortening healthcare pathways to the diagnosis

**6.4.1. Mobility of experts and expertise** (including information and information technology) can be facilitated by development of a network of experts where anyone can be informed about where a given disease is taken care of.
**6.4.2. In order to make a national network of the present laboratories** and to support them, a survey of the present conditions, capacities and activities is necessary at a national level. Results should be regularly updated and made available for anyone on the homepage of National Centre for Healthcare Audit and Inspection. Accreditation and regular supervision of the laboratories is obligatory for quality assurance.

**6.4.3. Interchange of DNS and other samples** can be organized and is already working in an international level in the daily practice. Genetic diagnostic testing abroad is available through an application process to National Health Insurance Found and in many instances National Health Insurance Found refunds its costs.

**6.4.4. Guide to diagnostic testing and testing regarding the whole population in Europe can be promoted by facilitation of joining European Reference Networks.**

**6.4.5.** Medical knowledge of special centres reaches general practitioners through local presentations where current protocols are introduced. Members of the National Centre for Healthcare Audit and Inspection can work out diagnostic and therapeutic protocols by the adaptation of international literature. These protocols will be accepted by a board of professionals of a given field and popularized in professional forums.

### 6.5. Organise healthcare pathways for patients suffering from rare diseases to access national CoEs or CoEs abroad. Facilitation of development of CoEs.

There are four university centres of expertize with diagnostic and therapeutic facilities: Budapest, Szeged, Pécs, and Debrecen. In addition to that, three more Care Centres are needed: Miskolc, Szombathely and Győr. Official identification of these centres should be fulfilled in an objective way through an accreditation method judged by RBK. National Centre for Healthcare Audit and Inspection initiated an open registry about the activities of centres of care and expertise, and activities of consultants and laboratories that require accreditation. Continuation of this work is absolutely necessary. The registry would contain diagnostic tests and the treated diseases. Apart from that, display of the links of European Reference Networks would be important to facilitate development of healthcare pathways.

### 6.6. Assurance of a multidisciplinary approach and integration of medical and social care in centres of expertise

Certain paramedical activities are already available in or in cooperation with university hospitals (ex. hospital teachers, physical therapists, social workers, psychologists). However, the above problems appear more frequently in these institutions, therefore, the need for such services is also increased. It would be important to improve personal conditions and capacity of these paramedical fields. Moreover, the homepage of National Centre for Healthcare Audit and Inspection that is under preparation should display supplementary social and educational institutions and the competent civil organizations. The propagation and distribution of flyers for patients in the centres would be also important.

**6.6.1.** System adapted to clinical treatment protocols for RDs works through an individual equity application in those cases when distant access to the specific healthcare needed. It covers multidisciplinary consultation and travel costs. In case of inland travel, the Health Insurance Found support travel cots of accessing healthcare services. Regarding inland travel support there is detailed information in the public utility menu on the public side of the homepage of the National Health Insurance Found. For relatives (in case of children), there is accommodation/bed in the hospital or McDonalds houses are available. There is no regulation on account of that not only children's parents but also adults themselves could go on sick-leave. We suggest a modification of labour law in this case.

**6.6.2.** National Centre for Healthcare Audit and Inspection and Hungarian National Public Health and Medical Officer Service could be the mediators of medical expertise between special centres and healthcare and social sphere.

**6.6.3.** Good cooperation with national and foreign professionals could be realized through connection to European reference networks an order that the most effective evidence based protocols became accepted. It has been realized only in part by now.

### 6.7. Feasibility of the evaluation of CoEs

6.7.1. System applicable to the evaluation of CoEs would be based on clinical results and patient satisfaction according to the below points:

- Type and number of diseases attended
- Number of registries of patients attended

- Quality of medical care
  - Multidisciplinary care
  - Tracking of Orphan drugs
  - Technological platform
  - Networking
  - Information flow

- Efficiency of medical care
  - Research
  - Publication
  - Clinical and research projects obtained

- Formation of clinical policy

- Gathering data for epidemiological database and development of health indicators

- Number of trainings organized
  - for medical doctors and professionals
  - Patients with RD/ Public health education

### 6.8. Recommendations

6.8.1. Mapping of the presently informally functioning CoEs, centres of care, genetic counselling and laboratories in Hungary and the development of an official accreditation process (through tender).

6.8.2. Assignment of the institutions and publication on the homepage of National Centre for Healthcare Audit and Inspection.

6.8.3. Support of the development of CoEs having extra tasks by separate financing mechanisms.

6.8.4. Control of accredited institutions based on EU compatible standpoints (exact patient registry, coding, multidisciplinary care, patient satisfaction etc.) by an independent supervisory organization. Data of patient registry (satisfactory) would be utilized in the evaluation of efficiency and in the quality assurance in a multilevel control approach where publicity would be realized in a number of ways including patient access.

6.8.5. Assurance of joining medical and paramedical services when determining healthcare pathways. Assurance of the missing human resources.

6.8.6. Accomplishing the connection to European Reference Networks and other EU programs even with application of sanctions.

6.8.7. Ensuring cross border access to the specific healthcare by shortening time of the application process when needed.

6.8.8. Introduction and spreading the information about the homepage constructed and the structure and function of the forming care system for RD's through the National Centre for Healthcare Audit and Inspection

and the Hungarian National Public Health and Medical Officer Service and also by programs, flyers and media appearance.

6.8.9. Involving civil organizations in every step of the development of the National Plan.

6.8.10. There is a great need for the establishment and operation of the National Habilitation, Development and Service Centre for Rare Diseases in those RDs that require intervention in order to facilitate social integration. It would be a coordinated, team-based, multidisciplinary habilitation and prevention centre which also fulfils social and re-educational goals and could be a treasury of intervention methods and possibilities, similar as in Scandinavia. The patient's family, after having received the diagnosis, could move in to the centre for a few weeks, where it would receive an all-inclusive lifestyle counselling and consultation. This centre would be closely connected to healthcare centres where diagnosis and therapy would be available. Their cooperation would be especially important in the field of prevention, orientation, indication, and recognition and guiding.

According to its previous plan, HUFERDIS would be willing to coordinate this project by holding together civil organizations and professionals with the help of EU support.

Until the development of an independent centre, it should be mapped that how present rehabilitation facilities could be transformed suitable for RD patients at least in part. EU surveys show that in comparison to the medical services, these services are even shorter and there is no such Centre in Central Europe at all.

## 7. topic – Orphan drugs and treatments

**7. Section:** Orphan drugs and treatments

**Chairs:** Imre Boncz, Pál Vittay, Magdolna Dank

**Date, place:** 2010.10.16. 11:00-12:30, Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*19. It is of utmost importance to ensure an active contribution of the Member States to the elaboration of some of the common instruments foreseen in the Commission communication on rare diseases: Europe's challenges of 11 November 2008, especially on diagnostics and medical care and European guidelines on population screening. This could be also the case for the assessment reports on the therapeutic added value of orphan medicinal products, which could contribute to accelerating the price negotiation at national level, thereby reducing delays for access to orphan drugs for rare diseases patients.*

**EUROPLAN Indicators:**

| Activity | Indicators | | Type of indicator | Answer |
|---|---|---|---|---|
| Ensure the mechanism that facilitates ODD access and the reimbursement of their cost to patients after they got the market authorization by EMEA. | 7.1 | Number of ODD market authorizations by EMA and placed in the market in the country | Outcomes | 70 % |
| | 7.2 | Time between the date of an ODD market authorization by EMA and its actual date of placement in the market for the country | Outcomes | 0-10 nap |
| | 7.3 | Time from the placement in the | Outcomes | 90 day in normal procedure, however, |

| | | | | |
|---|---|---|---|---|
| | | market in the country to the positive decision for reimbursement by public funds | | this could be elongated in case of complicated cases. |
| | 7.4 | Number of ODD reimbursed 100% | Outcomes | 33 |
| To develop mechanisms to accelerate ODD availability | 7.5 | Existence of a governmental program for compassionate use for Rare Diseases | Outcomes | • No |

### 7.1. Future of orphan drugs

#### 7.1.1. Available orphan drugs and number of patients treated with these drugs

30 claims for orphan drugs have been accepted by social insurance between 2004 and 2010.

Currently in Hungary 14 orphan drugs in different stripping and component content receive support; altogether 23 orphan products are supported. Most product 0% (hospital financing) and accentuated, indication linked 100% (distributed in pharmacies) accepted in support category. One single orphan drug support: 0% and accentuated, indication linked, 90%, two products: 0%

Within the framework of outpatient service, in 2008, 1431 patients, in 2009, 1732 patients have been treated with orphan drugs. In hospitals two orphan drugs have been reported. In 2008, 18 patients, in 2009, 20 patients received these drugs. In 2009, 289 patients' discretional fair claim for orphan drugs has been accepted. Around 13 rare diseases receive support within the framework of discretional fair claims.

#### 7.1.2. Rate of obstruction to access orphan drugs within the time frame set by EU law (180 days)

Access to orphan drugs that are already TB supported is legally assured by the service providers contracted with the National Health Insurance Fund. These service providers are the pharmacies in the case of the civil population, and the hospital pharmacies in the case of hospitalisation.

In Hungary, the drug acceptance process (transparency) to the social insurance is based on the EU transparency Directive (89/105/EK). The relevant Hungarian legislation is implanting the EU Directive, according to which a difference is made among original/innovative and generic drug during the acceptance process. Producers of innovative orphan drugs which have already received approval for market release can ask for social insurance support via the regular process. In this case, within the time limit set by the EU, 90 days are open for price discussion, plus another 90 days to determine the rate of support, altogether 180 days are open. In Hungary, during the transparency process, governmental organs will only decide on the rate of the support. Since the local legislation is based on the EU one, therefore in normal process, 90 days are the time limit of acceptance. If the change of the legislation is needed for the acceptance of the orphan drug, then the process is suspended until the decision of the competent minister for medication (until the change on decree for acceptance and supports of drugs comes into force or until a negative decision), but up to a maximum of 90 days from submission of the claim. First of all those new, not yet supported components 'claims are in these circle, for which new definition is needed for ATC group, support category or rate within ATC group, or new disease category not yet included in the legislation for acceptance and support of drugs, or definition of new indication category. In the case of generic products, in the course of a simplified procedure the decision on the acceptance is made within 60 days.

In the case of discretional claims the law sets 22 working days as time limit for procedure. Drugs not supported by social insurance can only ask for a support within this procedure.

We can state that when accepting the orphan drugs, equal judging exists on legal level as well, while judging all the criteria of support (relative efficacy, cost efficacy ....) is difficult or is subject to modification due to their special status assured by the EU during the centralised market release procedure.

The Hungarian time limit is said to be very long in the case of oncological drugs (not always orphan drugs) within the EU. Due to the limited resources, drugs with high price are constantly handicapped and this is

especially true in the case of orphan drugs. The current practice means that they are only accepted if there is a significant spare in the drug budget and this harms the acceptance of orphan drugs. Therefore, the time for availability is significantly longer than 180 days it can even be multiplied

### 7.2. Availability of orphan drugs. Pricing and support.

**7.2.1. Possibilities of fastening and developing the national procedures for price/support determination of orphan drugs:**

There is no specific procedure for accepting orphan drugs, the increased acceptance threshold is especially lacking. (QALY)!

Many times support is only available via discretional claims. Consideration of claims is difficult and time consuming, simplifying and speeding up the procedure is needed. Financing capacity of the state significantly limits the yearly budget (macroeconomic situation). Currently the number of discretional claims is limited to 50 patients which challenge their constitutional rights.

National Health Insurance Fund (OEP) manages within the budget separated for discretional claims in E Fund. Since these products do not receive support, price determination is allocated to the producer; OEP does not have a direct impact on pricing. When considering discretional claims, support is determined in a discretional procedure, taking into account the financial status of the claimant.

In view of pricing, decisions should be based on EU level joint valuation and on reference prices, working out special acceptance policy procedures for entering into the supported circle, developing risk sharing models, shaping well defined, narrowed patients' groups for being supported are necessary. Support claims should never be rejected due to lack of information or due to financial reasons.

**7.2.2. Introducing mechanisms, necessary to minimise the time limit of availability of drugs, based on the EU level report of EMA European Medicine Agency on the clinical added value of orphan drugs, determining the national decisions on pricing and support.**

When evaluate the outcome of clinical medication, the procedure could be fastened by facilitating the access to data, rationalising the legislation on data protection, specifying and simplifying the regulation of decision making. In the case of orphan and ultra-orphan drugs, it should be considered to use specific procedural regulations.

Orphan drugs, in comparison to other innovative drugs, are approved and accepted for support using different criteria, i.e. the number of patients in clinical examinations is limited , often certification of evidences are not so well based. Considering the relative efficacy, it can be stated that EMA, when it approves the drug, does not require the prior condition of randomised, controlled, two-blind examination, resulting in two concordant opinions. Costs of therapy pear head (actual time of the therapy, or if the therapy exceeds a year, then therapy per year), similarly to the potential number of patients, vary in a large interval, between 2-135 million HUF per patients, 3-442 patients per indications.

In the case of a positive acceptance procedure, greater flexibility of producers, traders, and their willingness for risk sharing should be reasonable. Specific price of orphan drugs is higher in comparison to other products; therefore greater value/performance based approach of producers/traders should be expected. In our view, it would be possible to use such an aid mechanism, which would allocate resources on discretional bases from a special fund, worked out for those diseases where the patients are innocent in the cause of the disease. Rare diseases are such.

**7.2.3. Measures promoting availability of orphan drugs in the national policy on discretional pricing and support, as recommended by the EU Pharmaceutical Recommendations:**

Availability and support is assured via discretional claims. It is the right of the producer/trader to determine whether it initiates the acceptance of the drug for regular support or not. At the same time, it is worthwhile to fasten the time frame of accepting orphan drugs to the supported circle.

Development of national network of care centres and centres of expertise will simplify the cooperation between the financing organ (OEP) and clinical professionals and practitioners. It would be worthwhile to consider working out a special method for the financial support of orphan drugs.

**7.2.4. Availability of orphan drugs via the centres of expertise**

In the case of oncological care, orphan drugs are available via assigned oncological centres. Using this model it would be worthwhile to set up orphan drug care centres. In the availability of treatment, there are regional differences within the country. Equal availability of orphan drugs both in quality and in quantity should be assured. Treatment cannot depend on the financial status of the hospital as no one should have to jeopardise his/her quality of life due to financial problems of an institute. Such approach would be cost effective as well, since a patient with a good quality of life will not fall out of the working society.

**7.2.5. Hungarian participation is supported in the survey of orphan drugs' clinical added value within the EU level cooperation of EMA (**European Medicine Agency).

### 7.3. Temporary authorisation of orphan drugs and discretional use. Off label use.

**7.3.1. Promoting availability of orphan drugs via discretional claims:**

We should consider support of experimental drugs/treatments by state resources (social security or clinical) together with protocol and data collection similar to the circumstances used in drug research. Pharmaceutical companies may support the diagnostic background tests.

**7.3.2. Subscription of drugs outside indication and preferential support are possible, provided that their benefits for patients are certified. Current Hungarian protocol makes it possible; however, procedure is very bureaucratic. In the future the current procedure should be simplified and rationalised.**

Frames of drug orders outside indication exist.

It must be noted that the off label use is not equal to the use of orphan drugs. Not all rare diseases are treated with orphan drugs and off label permit to use the necessary drug is not always needed. Information on orphan drugs receiving social security support and on off label use indication, number of patients diagnosis, treatments etc., are managed by OEP. Due to the limited number of patients, however, sharing these data with outside partners would increasingly raise the question of data security.

**7.3.3. Treatment outside pharmaceutical products. Further studies and adaptation are needed on the area of treatments other than pharmaceutical if their benefits for patients are certified.**

Early development, physical therapy, conductive training, TSTM, HRG, Dévény method, therapeutic pedagogy, family care, psychological treatment etc. Set up of the National Habilitation Development and Care Centre of Rare Diseases would promote feasibility to concentrate all these possibilities

### 7.4. Recommendations

7.4.1. Taking into consideration the positive examples of the international practice is recommended in the local practice.

7.4.2. Promotion of availability to orphan drugs by simplification and fastening the procedure of discretional claims. Set up of Orphan Drug centres using the sample of oncological centres

7.4.3. Promotion of availability to drugs outside indication by simplification and fastening the procedure.

Professional's information supporting the decision making is needed for ordering drugs outside indication.

7.4.4. We recommend development of a unified national concept on introducing central venous catheter in the case of infant cancer patients and of all those rare disease patients where several operations are needed. Introduction of a central venous catheter adapted to the age of the child and to the protocol of the disease is recommended before starting the chemotherapy or other treatments where such catheter would be needed. Adequate information for parents, consideration of their points of view when choosing the CVC is also needed. Such methods would eliminate much unnecessary suffer for children.

7.4.5. In the case of a change in therapy recommended by the specialist, a flexible update possibility of the drugs' list where full support is available on certain family conditions would also mean an instant help for the families. This way it should be avoided that while there are unused financial support frames, the family must pay significant amount for other drugs even if it would be entitled to full financial drug support.

## 8. theme – Patient Empowerment and Specialised Services

**8. Section:** – Empowerment of patients' organisations

**Chairs:** György Harmath, Judit Váradiné Csapó, István Nagy

**Date, place:** 2010.10.16. 11:00-12:30, Hunguest Hotel Griff***

**Council Recommendation (2009/C 151/02)**

*18. Consult patients and patients' representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases.*

*19. Promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking and outreach to very isolated patients.*

**EUROPLAN Indicators:**

| Activity | Indicators | | Type of indicator | Answer |
|---|---|---|---|---|
| Promoting the existence of a RD patients' organizations that represent all RD patient associations | 5.1. | Number of umbrella organisations specific on rare diseases | Process | Existing only one organization |
| | 5.2 | Having a directory of RD patients organizations | Process | Yes |
| | 5.3 | Number of Patients' associations | Outcomes | 35 |
| | 5.4 | Number of diseases covered by patients' associations | Outcomes | approx. 100 |
| Patients' organizations involvement in decisions affecting RD | 5.5 | Permanent and official patients' representatives in plan development, monitoring and assessment | Process | Yes, clearly established, substantially implemented and participation reimbursement considered |
| | 5.6 | Participation of patients organizations in the development of RD research strategies | Process | Sometimes are consulted before the final document is approved |

| | | | | |
|---|---|---|---|---|
| | 5.7 | Participation of patients organizations in the RD centres of expertise designation and evaluation | Process | Yes |
| Support the activities performed by including patient organizations, such as awareness raising, capacity building and training, exchange of information and best practices, networking, outreach to very isolated patients | 5.8 | Resource (funding) provided for supporting the activities performed by patient organisations | Outcomes | There is no separated funding for patient organizations |
| | 5.9 | Support to sustainable activities to empower patients, such as awareness raising, capacity-building and training, exchange of information and best practices, networking, outreach to very isolated patients | Outcomes | not relevant |
| Building - supporting the existence of comprehensive help line for patients | 5.10 | Availability of Help line for RD | Process | Not formal decisions have been taken |

### 8.1. Involving patients and their representatives to the decision making procedures on rare diseases.

**8.1.1. Possibilities of involving patients and strengthening their roles in the following territories:**

- **Development of National Plans**: assuring participation of HUFERDIS in the work of the organising committee, at each phase of the elaboration and the evaluation of the plan.

- Professional responsible person must be named for the territory of rare diseases within the government, actively taking part in the decision making must be named in order to insure his/her identification. He/she must report/inform RIROSZ on the relevant work.

- **Providing information:** after finding of the diagnosis, centres of expertise and genetic consultants should provide information on existing specific civil organisations (using leaflets, availability, etc.) Their presentation materials and web links should be available on the information homepages of National Centre for Healthcare Audit and Inspection, of the centres as well as on conferences

- **Development and management of Centres of Expertise and European reference networks**

  Centres of Expertise must keep continuous contact with HUFERDIS and with the representatives of the relevant civil organisations, involving them into the activity, research and program of the Centres when they are concerned. Centres should inform patients on the achievements of the European reference networks.
  Evaluation of supervision should contain a survey of the results of patients' complacency level.

- **Determination of guidelines on the research of rare diseases:**
  Representatives of patients' organisations should be involved in the setup of the protocol, in the information and recruit of patients. Results must be shared from time to time.

  **Involvement on territories, such as set up and management of registries, clinical experiments, evaluation of clinical added value of drugs, therapeutic education and care programs, medical and social care training etc.**
  Involvement of these organisations in the crediting procedure and in the development of its consequences is also recommended.

**8.1.2. Assuring referred tender support or state normative support should help patients' taking part in the decision making process on the territory of rare diseases.**

### 8.2. Supporting the activity of patients' organisations

**8.2.1. Currently neither HUFERDIS, nor any rare disease organisation receive normative state support, contrary to similar other umbrella organisations, like:** ÉFOÉSZ, SINOSZ, MEOSZ, MVGYOSZ, etc.), while the number of patients (600-800 thousands) represented by HUFERDIS is multiple of those represented by the other above mentioned organisations (even if removing the overlaps). At the same time HUFERDIS works on the same high level and its work is similar to that of the above mentioned organisations, it is present on both national and international levels, undertaking several state functions. All these significant amount of work can only be performed by the help of civil volunteers, the using infrastructure of existing old member organisations. Long term maintenance of its work cannot be ensured without infrastructure and without at least part time employees. Its programs aimed at national adaptation of good EU practice and at approaching EU standards is executed using ad hoc tenders and sponsor resources.

**8.2.2. Alongside to the assurance of the state normative support, charity activity of companies can be triggered by tax allowances.**

**8.2.3**. Rare diseases are chronic, life threatening, need daily supervision and care. While in general rare diseases are associated with disabilities, this is not always the case. In order to compensate the handicap both the solidarity of the society and the state support must be increased. In these families often only one member is working and due to this their status is cumulatively handicapped. Therefore those disabilities where no sign can be seen (such as chronic fatigue and pain) should also be measured by adopting existing European tools. It is also necessary to ensure the availability of special care, support and allowances for all those patients who live with a rare, chronic disease. The supporting structure must also include the whole family, not only the patient.

**8.2.4.** When modifying the tax system, it should be taken into consideration that families raising children suffering from a rare disease that cannot be linked to any other group should also receive tax allowances.

**8.2.5.** Campaigns raising social awareness should be launched, such as starting social solidarity programs, preparing and distributing leaflets, triggering training of specialists (social, educational and health care sphere) etc.

**8.2.6.** Centralised sourcing, knowledge base, supporting institutions (incubation houses), enlargement of training centres/capacities to professionalise civil associations must be initiated.

**8.2.7.** There are several good examples of other types of support and cooperation, i.e. several staff member of OSZMK also work/are honoured member in civil associations (David Short People Association (achondroplasia), Association of People Living with Limb Shortage, Csupaszív, Kézenfogva etc). In these associations professionals of OSZMK help the patients and their relatives by providing human genetic consultation, by educating and presenting. Via phone or e mail or by post, they help the rare disease patients on a regular basis.

### 8.3. Special social care: temporary care, therapeutic recreational programs, programs to help patients in the everyday life integration

**8.3.1.** On the territory of early development and temporary care giving, there are good and high quality programs, which support the patients and their families living with rare diseases. **Capacities of these programs are, however, not enough and they do not cover the entire country. The majority of them are held by civil associations.**

**8.3.2.** The national strategy „Our Common Treasure, the Child" gives overall concept of principles on how to integrate and develop children living with disabilities, but no action program is linked to the principles. Legislation exists on care, training, integration, work help of SNI Children (obligatory training hours, increased normative, increased possibility for home care for the first years, fee for home care etc.); however, these possibilities are not available for all rare disease patients.

**8.3.3. In order to make these possibilities more available for the patients, several measures should be taken, including state support:**

Applications, claims procedures should be simplified, rationalised, harmonised and parallelism should be cancelled.

- Training the social sphere and care givers for the special tasks associated with rare diseases.
- Existing support possibilities should be more publicised.
- Centres of expertise should also give room to consultation and information.
-

**8.3.4. Financing special cares show mix picture.** These tasks are undertaken either by the state institutions using state budget or by private companies or by civil associations.

### 8.4. Helplines

**8.4.1. There are no RB specific helplines, neither for the professionals nor for the patients.**
Civil associations partly provide such services on an informal basis.
**8.4.2.** Civil associations do not have enough capacity either in staff or in financials to maintain a helpline which is free for the caller. Being a small country, there is no need to maintain separate helplines for all rare diseases groups. It would be a good solution to set up an operation body with trained personnel, either financed by the state or using normative support. To extend existing helplines, such as Blue –line or Association of Mental Help Phone Lines) would also be a good possibility.
**8.4.3. To set up the 116 European dial up number**, an organising body should be appointed, trained and financed, together with establishing the information and knowledge base necessary for their work. Promotion of the helpline in hospitals, webpages, in mass media etc. should also be worked out.

### 8.5. Recommendations

8.5.1. See 8.1.1.

8.5.2. Representatives of patient associations should be involved in the process of working out the protocol, information and recruitment. From time to time, patient associations should be informed about the outcomes.

8.5.3. See 8.2.2.

8.5.4 See: 8.2.3.

8.5.5. See: 8.2.4.

8.5.6. See: 8.2.5.

8.5.7. See: 8.2.6.

8.5.8. See: 8.3.3.

8.5.9. It is necessary to enlarge the possibilities for helping social integration, care giving programs and institutions. Special emphasis should be put on the territory of social even chance.

8.5.10. To be linked to the international 116 number, a local organising body must be appointed, set up, trained and financed.

## V. Consequences of the final report

**1, The role of the Europlan recommendations and indicators in serving as support to the elaboration of our national strategy:**

The well-structured recommendations (together with the supporting documents) have served as a wonderful basis both for the professional organisation of the conference and for its high level execution, for the final report and for the steps starting the elaboration of the National Strategy. Indicators served as objective frame for evaluating the current situation as well as for monitoring the future development. In several cases it turned out that adequate measures need rearranging currently available data and elaboration of the possibility of a new statistical separation.

**2, The possibility of transplanting the EUROPLAN recommendations into our National Strategy.**

General principles and consequences of the recommendations served as valuable guide and proved to be simply followed. However, their support has not been enough for the implementation of the specific measures. For this purpose, specific observations of other countries served as valuable support too. These examples (framed in the text of the recommendations) naturally reflected rather the situation of the other country, therefore only a minority of them fitted to the Hungarian circumstances. However, these latter served as good practical ideas!

## VI. Document history

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## VII. Attachments

### 1. Attachment: Glossary

| Abbreviation | Meaning |
|---|---|
| CoE | Center of Expertise |
| ERN | European Reference Network |
| OD | Orphan Drug |
| RD | Rare Disease |
| OEP | National Health Insurance Fund |
| ÁNTSZ | National Public Health and Medical Officer Service), |
| OB | Organizing Committee |
| NP | National Plan |
| RBK | Rare Disease Centre |
| OSZMK | National Centre for Healthcare Audit and Inspection |
| BURQOL-RD | Social Economic Burden and Health-Related Quality of Life in Patients with Rare Diseases in Europe |
| QALY | Quality Adjusted Life Year |
| ETT | Medical Research Council |
| EMA | European Medicine Agency |

| | |
|---|---|
| TSMT | Planned sensomotor training |
| HRG | Hydrotherapeutic Rehabilitation Gymnastics |
| CVC | Central Venous Catheter |
| ICD | International Classification of Diseases |
| NCA | National Civil Fund |