Sharing best practices on integrative approach to rare diseases in different countries

Hungary

Janos Sandor
National Centre for Healthcare Audit and Inspection
Legal environment of rare disease health policy development

Ministry of Health

Hungarian Parliament

Health Commission of the Hungarian Parliament

National Public Health and Medical Officer Service

Public Health Program

National Centre for Healthcare Audit and Inspection

Rare Disease Centre (est. November 2008 by modification of Deed of foundation)
Structure of Rare Disease Centre

National Centre for Healthcare Audit and Inspection

Rare Disease Centre

National RD Advisory Board (university, governmental, patient representatives)

National RD Research Coordination Centre outsourced to Pecs University

IT center: hospital and outpatient discharge records analysis registry of RD providers, labs, research programs, patient groups

Universities’ internal RD coordination units

Nominated RD responsible persons in the Ministry of Health, in the National Institute of Pharmacy

Regular consultation with National Board for Genetics

Project based collaboration with sociological centers, National Foundation for Disabled Persons, National Centre for Statistics
Rules and Regulations of RDC

- defines public health indicators for rare diseases (RD);
- elaborates its own data collecting technology and co-operates with other agencies in order to get RD related data, in order to prepare these indicators;
- initiates the elaboration of RD guidelines and carries out the audit projects;
- maintains the national database of RD specialized health care providers;
- contributes to the assignment of national centres of expertise and their participation in European networks;
- facilitates the establishment and operation of the quality management programs for the Hungarian RD laboratories;
- facilitate the E-health application in RD related care;
- initiates the RD teaching programs launching in the universities;
- participate in the work of national agencies responsible for orphan drug and orphan medical device legislation;
- supports the improvement of the availability of special social services for RD patients;
- supports the effective primary preventive program;
- evaluate the efficacy of the RD screening programs;
- facilitates the RD research projects, the international co-operations;
- contributes to the development of collaboration between governmental bodies, providers and patient organizations;
- supports the Hungarian participation in the European RD projects;
- initiates programs, which contribute to the improvement of RD perception among lay people;
- co-ordinates the elaboration and monitoring of national policy on RD;
- reports on the Hungarian achievements regularly;
Primary preventive measures

1st Central and Eastern European Summit on Preconception Health and Prevention of Birth Defects (August 27-30, 2008)
To provide a platform for review, analysis and discussion of the promotion of women’s health before, during and beyond pregnancy, and the role of preconception health and health care in the prevention of birth defects in the Central and Eastern European region.
Socio-economical determinants of periconceptional folate supplementation (39% is supplemented)

Mothers’ education

Fathers’ education

(significant SES-gradient)

(Governmental supported project on the periconceptional folate status and on attitude towards different supplementation programs)
Psychological determinants of being supplemented by folic acid during early pregnancy (Trait Anxiety Inventory)

The anxiety during early pregnancy is connected with low prevalence of supplementation (ORs adjusted to age, mothers’ and fathers’ education)

„I worry too much over something that really doesn't matter”

OR = 0.427 (0.211 – 0.863)

„Some unimportant thought runs through my mind and bothers me”

OR = 0.557 (0.324 – 0.960)

(Governmental supported project on the periconceptional folate status and on attitude towards different supplementation programs)
Screening

Existing programs:
1) Compulsory screening during pregnancy by gynecologists
2) Compulsory neonatal metabolic disorders (25) screening
3) Compulsory screening of children by pediatricians and public health visitors

Proportion of prenatal diagnosis of Down syndrome in Hungary

Proportion of induced abortion of Down syndrome in Hungarian districts

75% <
50% - 75%
25% - 50%
< 25%
Newborn screening program

- arginosuccinic aciduria (arginosuccinate lyase deficiency, ASL)
- beta-ketothiolase (oxothiolase) deficiency
- biotinidase deficiency
- Carnitin transport deficiency (CT)
- Carnitin-palmytoil transferase deficiency (CPT-I)
- Carnitin-palmytoil transferase deficiency (CPT-II)
- citrullinaemia I (argininosuccinát synthase deficiency, ASS)
- GA II
- GA I
- galactosaemia
- homocystinuria
- hypothyreosis
- IVA
- LCHAD
- MCAD
- MCC
- MMA
- MSUD
- multiplex carboxylase deficiency
- PA
- phenylketonuria
- SCAD
- tyrosinaemia I
- tyrosinaemia II
- VLCAD
Availability and accessibility of accurate diagnostic/genetic tests

At present:
Many genetic laboratories
  With small capacities, without quality management programs
  Many missing services – Existing services expensive – Limited reliability

Genetic counseling centers are sometimes far from the patient
  Less capacities then needed - Difficulties in access

Proposal of the National Network of Medical Genetics
  elaborated by the National Health Council
  6 qualified labs (concentration)
  increased density of genetic counseling centres
Regional distribution of age distribution of RD patients – indirect measure of delay
Best practices on RD care

1. Almost every national board of the medical professions has a nominated expert on rare diseases

2. Protocol development in the Ministry of Health
   - RD related guidelines and protocols released
   - Cystic fibrosis
   - Myasthenia gravis
   - Autism spectrum
   - Diagnosis of the inherited metabolic diseases

3. National Institute of Healthcare Improvement and Inspection to develop audits
Development of national/regional centres of expertise/reference

At present:

• Expensive therapies available in centres only
• The centres are assigned by National Health Insurance Foundation case-by-case
• No systematic accreditation, and service development of CE

Ongoing:

• Project on database building of RD providers:
• Conferences on centers of excellence (2008, 2009)
• Preparation of accreditation system
Concentration of outpatient care for RD

50% of patient in 25 outpatient centers

80% of patient within 50 km
Regional differences in availability of RD outpatient care

Down syndrome

Cystic fibrosis
Hungarian rare diseases mortality study on the underlying cause of death declared by death certification

![Graph showing the trend of rare diseases mortality over different age groups and time periods.](image)

- **Graph Description**:
  - The graph illustrates the trend of rare diseases mortality from 1980-84 to 2005-06.
  - Each data point represents the probability of death from rare diseases within a specific age group (5-9, 10-19, 20-29, etc.) across different time periods.
  - The probability is measured on the y-axis, while the age groups are plotted on the x-axis.
  - Separate lines are used for different time periods (1980-84, 1985-89, 1990-94, etc.), each color-coded for easy distinction.

- **Key Observations**:
  - There is an overall increasing trend in the probability of death from rare diseases across all age groups.
  - The probability is highest in the elderly age groups (70-79, 80-89, and 90+).
  - The probability is lowest in the age groups below 20 years.

- **Statistical Significance**:
  - The trend is statistically significant with a P-value of <0.001.
  - The coefficient of determination (R^2) is 0.821, indicating a strong relationship.

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Fourth Eastern European Conference for Rare Diseases and Orphan Drugs
“Together for Integrative Approach to Rare Diseases”
13-14 June 2009 - Plovdiv, Bulgaria

In collaboration with
## EurordisCare3 survey in Hungary

<table>
<thead>
<tr>
<th></th>
<th>EurordisCare3</th>
<th>Hungary</th>
<th>Hun/EC3</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Number of medical services used per patient over the last 2 years</strong></td>
<td>9,4</td>
<td>9,2</td>
<td>0,98</td>
</tr>
<tr>
<td><strong>Number of different care used per patient over the last 2 years</strong></td>
<td>2,4</td>
<td>1,9</td>
<td>0,81</td>
</tr>
<tr>
<td><strong>Rejection by health professionals</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Rejection linked to the disease</td>
<td>17,8%</td>
<td>15,1%</td>
<td>0,85</td>
</tr>
<tr>
<td>Rejection linked to the patients physical aspect</td>
<td>14,7%</td>
<td>12,6%</td>
<td>0,86</td>
</tr>
<tr>
<td>Rejection linked to the patients behaviour</td>
<td>1,8%</td>
<td>1,3%</td>
<td>0,69</td>
</tr>
<tr>
<td>Rejection linked to the patients communication difficulties</td>
<td>1,9%</td>
<td>1,1%</td>
<td>0,60</td>
</tr>
<tr>
<td><strong>Need for social assistance</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Access to social assistance impossible</td>
<td>29,0%</td>
<td>21,0%</td>
<td>0,72</td>
</tr>
<tr>
<td>Access to social assistance difficult</td>
<td>4,8%</td>
<td>7,5%</td>
<td>1,58</td>
</tr>
<tr>
<td>Access to social assistance easy</td>
<td>27,4%</td>
<td>34,2%</td>
<td>1,25</td>
</tr>
<tr>
<td><strong>Reduction in professional activity as patient</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Reduction in professional activity to take care of a relative</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Need for moving</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>to a more adapted house</td>
<td>17,8%</td>
<td>25,1%</td>
<td>1,41</td>
</tr>
<tr>
<td>to a specialised care centre</td>
<td>10,8%</td>
<td>18,3%</td>
<td>1,70</td>
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<tr>
<td>nearer to specialists</td>
<td>2,3%</td>
<td>3,0%</td>
<td>1,33</td>
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<tr>
<td>closer to a relative</td>
<td>2,5%</td>
<td>1,2%</td>
<td>0,47</td>
</tr>
</tbody>
</table>

*In collaboration with* EurordisCare3 survey in Hungary
Orphan drugs

• Few of them is available in Hungary
• The cost is very small for patients can get the OD

• The National Institute of Pharmacy has no distinct protocol for orphan drugs’ authorization

• There is system of National Health Insurance Foundation for special drug import if a patient need orphan drug not commercialized in Hungary (case-by-case expert group opinion needed + significant budgetary constrains)
Empowerment of patients organizations

- Patients’ representatives are in the National RD Advisory Board
- There is no regular, direct governmental support for RD self help groups
- There are many indirect financing mechanism of the government: ¼ of the civil budget is from governmental sources
- Study on social capital of families with RD child
- Study on the sociological features of RD self-help groups

<table>
<thead>
<tr>
<th>Source of Funding</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normative from national budget</td>
<td>3.5%</td>
</tr>
<tr>
<td>Non-normative from national budget</td>
<td>9.9%</td>
</tr>
<tr>
<td>1% of personal taxes</td>
<td>0.7%</td>
</tr>
<tr>
<td>National foundations</td>
<td>3.0%</td>
</tr>
<tr>
<td>VAT-refund</td>
<td>0.5%</td>
</tr>
<tr>
<td>Normative from local governments’ budget</td>
<td>0.4%</td>
</tr>
<tr>
<td>Non-normative from local governments’ budget</td>
<td>4.2%</td>
</tr>
<tr>
<td>Governmental institutions budget</td>
<td>1.4%</td>
</tr>
<tr>
<td>Governmental together</td>
<td>23.6%</td>
</tr>
</tbody>
</table>
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Survey on methods of breaking bad news in hospitals

(89 hospitals; 01/11/2005-01/02/2006)

Do you have protocol on informing patients?

![Bar chart showing yes, no, and no answer categories with percentages]
Conferences on multidisciplinary care

Multidisciplinary care of Down-children (2006.)
- systematic review of the contribution of different professions
- benefit of collaborations

Case studies on Down-families (2007.)
- focusing on the crucial problems of the Down-children and their families
- identifying intervention targets

Rare disease patients’ organizations role in life quality improvement (2008)
- discussion on patient groups as organizations
- structure, operation, impact, non-utilized opportunities

Every child is unique (2009)
- Montessori pedagogy for handicaped children

Rare Disease Day (2006, 2007, 2008)
- for health care professionals

Rare Disease Awareness Day (2007, 2008)
- participation in HUFERDIS meetings

Lectures in conferences on genetics, public health, sociology

Establishing RD health policy elements by

- Utilization of the existing organizations, legal opportunities, expertise

- Building capacities, networks

-Considering the EU recommendation

Keeping on agenda the necessity of national program