EVALUATION OF THE QUALITY OF HEALTH CARE SERVICES FOR PATIENTS WITH RARE DISEASES

Most of the RDs are chronic, have a severe evolution and are associated with a poor quality of life (Nogales, 2004). People with rare diseases often experience the same problems: failure or delay in diagnosis, limited health information, lack or delay of referral to specialised professionals, poor co-ordination of in and outpatient care, poor social support including integration into working, social and educational environment.

The PMG, EWG and other partners held 2 meetings (December 16-17, 2002: “The role and contribution of patient’s organisations” – Rome, Istituto Superiore di Sanità (ISS) and December 19, 2002: “Sociological aspects of RDs and Quality of Life” – Rome, Istituto Superiore di Sanità (ISS)) discussing the social aspects and related needs of patients with RDs. The discussion emphasised that RDs have an impact on different aspects of life that need to be properly studied and understood in order to provide the appropriate support to patients. RDs undermine the health of the patients; often the physical or mental impairment is exacerbated by the poor understanding of the disease and it creates difficulties in the everyday life including school attendance for children or working capacity for adults.

During the meetings it was agreed that was important to better explore the different needs of patients with RDs as many gaps still exist in this area. Accordingly, the PMG agreed to undertake a pilot study on the accessibility to and quality of health, social and educational services for patients with RDs. The study was undertaken in collaboration with patients’ associations. Patients’ association are essential to provide the right answers, to reach patients and/or care givers willing to participate in the study. In addition, patient’s association can play a major role in identifying the priority research questions and in developing the appropriate tools for undertaking the survey.

3.1 Pilot study on the quality of and accessibility to health and social care in RD management

A pilot study was carried out to assess the quality of and accessibility to health and social care. The study focused on 4 of the 7 diseases studied by the NEPHIRD: Myasthenia Gravis, Neurofibromatosis, Prader Willi syndrome and Rett Syndrome. The diseases were selected because of their major physical involvement (Myasthenia Gravis), mental impairment (Prader Willy and Rett Syndrome) or because of the impairment of different systems and/or organs (Neurofibromatosis). The pilot study was undertaken in the following NEPHIRD countries: Italy, Spain, France, and UK. It is worth mentioning that because of the increasing interest on RDs and the better networking at the EU and not EU level, additional countries such as Romania and Turkey expressed their interest in being involved in the study and they were welcomed to participate. For each of the 4 diseases, patients’ associations were identified among the different countries as follows: Prader Willi Syndrome
A self-filled questionnaire was elaborated and validated by the expert on accessibility and quality of health care in collaboration with the clinical experts of the specific diseases selected (Appendix 4).

The self-filled questionnaire is addressed to patients and caregivers. The latter are the most appropriate in reporting opinions of patients with major mental impairments such as those affected by Rett Syndrome and Prader Willi syndrome.

The questionnaire asks respondents (patients or caregivers) how often they had had both negative and positive experiences about accessibility to and quality of health and social services. The frequency is measured with a 5-level scale: “every time”, “most of the times”, “sometimes”, “seldom”, “never”.

The questionnaire includes questions about the following items:

1. negative opinions on: access to and quality of health care (diagnostic exams, pharmacological therapies, rehabilitation, psychological support); access to and quality of social services (vocational training, work, education); lack of kindness and empathy by professionals; poor quality and quantity of information on health, social, educational services, policies and legal provisions; coordination and integration of care; amount of public financial support to health and social services.

2. positive opinions on: availability of and readiness to help by professionals, good quality of information on health, social, educational services, policies and laws, improvement in health conditions.

The questionnaire includes also some questions to capture patients’ opinion on the improvements of public services in the last three years. The improvement is measured with a 5-level intensity scale (“a lot”, “something”, “little”, “no improvement”, “deterioration”). The questionnaire is available in Annex 4.

The first draft questionnaire was in Italian and was tested in a focus group conducted with a panel of patients’ associations (Accessibility to and quality of health social services in Italy for the patients with rare diseases: the opinion of associations. Agazio et al. Ann Ig 2005; 17:121-128). The final version of the questionnaire was translated into English.
The English and Italian versions of the self-filled questionnaire were sent to the chairmen of the aforementioned patients’ associations. Each association translated the questionnaire into the local language and disseminated it to the members. The data were collected from August 2004 to July 2005.

Statistical analysis

For each item of the questionnaire average, standard deviations, median and the proportion of very negative and very positive answers (every time + most of the times) with 95% confidence interval, were computed. To assess the internal validity of the questionnaire we studied the association between negative and positive opinions of few topics (accessibility to and quality of health and social care, physician attitudes toward patients, health and laws information), using the non-parametric Spearman’s correlation coefficient. To assess the statistical significance of differences in averages, the non-parametric-Kruskall-Wallis test was used. Descriptive and comparative analyses were performed using Stata for windows and Epi Info 2000.

3.2 Results

In total 302 questionnaires were filled in: 66 for Prader Willi Syndrome, 89 for Myasthenia Gravis, 99 for Neurofibromatosis, 48 for Rett Syndrome. There were no statistically significant differences in the demographic characteristics of the respondents for the same disease among the different countries. For all diseases, more than 50% of the respondents were females.

Table 3.1 Size and main demographic features of the respondents’ by countries and diseases.

<table>
<thead>
<tr>
<th>Prader Willi Syndrome</th>
<th>Italy</th>
<th>Romania</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of respondents</td>
<td>30</td>
<td>36</td>
</tr>
<tr>
<td>% of females</td>
<td>60%</td>
<td>59%</td>
</tr>
<tr>
<td>median age (years)</td>
<td>10</td>
<td>10</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Myasthenia Gravis</th>
<th>France</th>
<th>Italy</th>
<th>UK</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of respondents</td>
<td>20</td>
<td>37</td>
<td>32</td>
</tr>
<tr>
<td>% of females</td>
<td>85%</td>
<td>73%</td>
<td>78%</td>
</tr>
<tr>
<td>median age (years)</td>
<td>53</td>
<td>48</td>
<td>63</td>
</tr>
</tbody>
</table>
Neurofibromatosis

<table>
<thead>
<tr>
<th></th>
<th>Italy</th>
<th>Spain</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of respondents</td>
<td>79</td>
<td>20</td>
</tr>
<tr>
<td>% of females</td>
<td>53%</td>
<td>55%</td>
</tr>
<tr>
<td>median age (years)</td>
<td>26</td>
<td>27</td>
</tr>
</tbody>
</table>

Rett syndrome

Respondents were all female.

<table>
<thead>
<tr>
<th></th>
<th>Italy</th>
<th>Turkey</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of respondents</td>
<td>26</td>
<td>22</td>
</tr>
<tr>
<td>median age (years)</td>
<td>16</td>
<td>13</td>
</tr>
</tbody>
</table>

For Prader Willi and Rett syndromes, almost all questionnaires were filled in by caregivers due to patients’ mental retardation.

Figure 3.1 summarizes the major areas of care on which patients/caregivers reported negative opinions.

**Figure 3.1: Negative opinions on provision of care**

The most negative experiences are reported on coordination of health services (58%), quality of information (44%) and social care (27%).

Eighteen percent of respondents reported a negative opinion on financial support and only 16% on the relational quality including lack of kindness and empathy of professionals.
Figure 3.2 shows that the most frequent negative opinions on health care are reported on quality of and accessibility to psychological support (31%) while the most frequent negative opinions on social care are related to the quality of and accessibility to work (29%).

**Figure 3.2: Negative opinions on health and social care**

![Bar chart showing negative opinions on health and social care](image)

Other negative opinions were reported on the following: health information on the diseases provided by professional (41%), quality and quantity of information on laws and rights (48%) and quality of vocational training (44%) (Figure 3.3).

**Figure 3.3: Information about the disease and laws and rights**

![Bar chart showing information about disease and laws and rights](image)

With regard to the positive opinions, for all diseases the most frequent positive experiences reported by patients and caregivers were kindness and availability (readiness to help) of health professionals (Figure 3.4).
Figure 3.4: Positive opinions reported by patients/caregivers

The following analysis aims at comparing responders’ opinions among countries.

**Neurofibromatosis**

Two countries (Italy and Spain) distributed the questionnaire to Neurofibromatosis patients’ association. Both countries showed the same pattern of responses: the highest complaints concerned quality of information (52%) and social care (36%) (Figure 3.5).

**Myasthenia gravis**

Myasthenia gravis patients’ associations were identified and involved in Italy, France and UK. All countries showed the same pattern of responses: the highest complaints concerned quality of information (54%) and social care (35%) (Figure 3.6).
Prader Willi and Rett Syndrome

Romania and Italy distributed the questionnaires to Prader Willi patients’ associations. The comparison between the 2 countries are relevant and interesting. For instance, in Romania the most negative experiences reported for Prader Willi syndrome were accessibility to and quality of diagnostic examination, drug therapies and rehabilitation intervention, while in Italy the major complaints were related to health and legal information, quality of and accessibility to vocational and educational training and work availability. Romanian patients placed access to and quality of health care at the top of their list of what makes them dissatisfied, probably due to the difficulty in diagnosing PWS. Italian patients had negative opinions mainly on information and social care.

Considering the similar demographic characteristics of respondents, possible explanation for such a difference could be the different socio-economic status of the 2 countries specially with regard to the health system and services. However, this is only a hypothesis and further studies should be done to better explore and understand differences among countries.
Rett syndrome patients’s associations were identified in Italy and Turkey. Turkish respondents complained more about educational services, Italian ones focused more on vocational training and information.

**Improvements areas**

Opinions on the public services improvement in the last 3 years, were generally negative. Health care and health information were the areas where more improvement was reported (Table 3.3).

**Table 3.3. Means value (with SD) of the opinions about the improvement made by public services in the last 3 years (2002-2005); (2 = no improvement; 3 = little improvement)**

<table>
<thead>
<tr>
<th>Fields</th>
<th>Means (SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Social support</td>
<td>2.9 (1,1)</td>
</tr>
<tr>
<td>Health care</td>
<td>3.2 (1,1)</td>
</tr>
<tr>
<td>Rehabilitation</td>
<td>3.0 (0,9)</td>
</tr>
<tr>
<td>Scientific research</td>
<td>3.1 (0,9)</td>
</tr>
<tr>
<td>Health information</td>
<td>3.2 (0,9)</td>
</tr>
<tr>
<td>Laws and patients’ rights</td>
<td>2.9 (0,9)</td>
</tr>
</tbody>
</table>
Conclusions

This was a pilot study aimed at assessing the needs of patients with RDs.

The assessment encompassing clinical, rehabilitative, social, educational and legal dimensions identified and confirmed the multidimensional aspect of the needs of RDs patients.

The study has some limitations due to the sampling method and to the limited validation of the questionnaire. In any case, even with these limitations some of the results appear to be of great interest.

1. The overall rate of negative and positive experiences are relatively similar for the four diseases, suggesting that rare conditions have major common problems: 1) social services (educational and work); 2) lack of health information and 3) inadequate provision of legal support.

Results have to be carefully interpreted. More social then medical problems were reported, however it could be due to the different availability of social and health care services within each country. Health care services even with limitations and problems are much more available than the social services across countries. In countries where this is not the case (Romania), the lack of health care services was identified as a major issue. The questionnaire asked about positive and negative opinions on different services thus respondents may have focused their opinions more on what is not available at (social services) all than on problems of what is somehow available (health services).

2. This study clearly stresses the importance of considering the different needs of RDs patients, needs that go clearly beyond the clinical side of the disease.

3. Considering the limitations of the study, it also points out interesting differences between countries with a different socio-economic situation. Further studies should be conducted to better explore such differences.

4. The study confirmed the two fold advantage of carrying out surveys engaging patients and/or their families: 1) it helps healthcare providers to improve the quality of and accessibility to their services and 2) it empowers patients. (Clerly PD, 1997, 1999).

In conclusion, it seems that health care and social services for RDs patients need to be improved. Immediate actions must be taken by Government to properly address the different needs of patients and also to provide better support to families.

People do not understand and appreciate diversity, persons with RDs are often marginalized and stigmatised instead of being fully integrated within the society. They deserve support from the society and government to study and work. The social network that should help and support families in the everyday management of patients is missing almost everywhere and families have to cope with the burden of the disease on their own.
**Recommendations**

In order to better plan and provide comprehensive support to patients with RDs and their families the following priority actions are suggested:

- Further explore social care needs and differences among EU countries. Validated tools to assess quality of care on the basis of patients’ opinions/health care users are needed. The questionnaire used in this study was only piloted and it would be very important to improve it. Rigorous methods should be used to elicit the patients’ experiences (Fitzpatrick R, 1991); responses are subjective and difficult to interpret since they depend also on expectations that vary greatly among patients (Acquadro C, 2004). The best way to collect patient’s opinions should be further discussed (general evaluation categories versus detailed description of the experience);
- Promote the assessment of patient’s and caregiver’s opinions to properly plan health and social service. Patient’s associations play a key role in promoting a culture on RDs, a social culture to defend rights related to diversity and health. They can deliver knowledge and information to doctors and families, a knowledge which is based on families’ experiences. Strategies should be developed bearing in mind such experiences;
- Advocate for the importance of a new, multidisciplinary and multidimensional approach to tackle RDs;
- Plan for an appropriate social support to patient with RDs and their families;
- Develop and disseminate information for patients, health professionals and the general population about RDs;
- Strengthen links and co-ordination between social and health services.

**Bibliography**