Rare Disease Day 2010
Bridging Patients and Researchers
to Build the Future Agenda for
Rare Disease Research in Europe

“The E-Rare Network”: Results from survey on scientists’ and policy-makers’ research priorities in the field of rare diseases

Manuel Posada*, Alejandro Ramírez# and Rafael de Andrés$

Research Institute for Rare Diseases (IIER)# & CIBERER* – Agency for Funding Health Research (FIS) $
Instituto de Salud Carlos III. (Madrid, Spain)
Aim

- To develop and influence research policies on rare diseases based on,
  - Experience from national and European funding agencies
  - Consultation with relevant Experts and Stakeholders

Deliverable # 4.3

- Strategy paper on future themes and needs of rare disease research funding
- Priorities for E-Rare funding
E-Rare: Strategy Paper

• Strategy paper on future themes and needs of rare diseases research funding

  • To gain knowledge of the current status in research funding in rare diseases
  • To define strategic priorities in research funding
  • To develop and influence common research policy on rare diseases
  • To propose future programmes and research activities on national, transnational and European levels
Methods

- **Questionnaire**
  - To assess the interest of the rare diseases community in a number of proposed topics related to research
  - Responses ranged from “No interest” to “High Interest”
  - Key feature: respondents only had to respond to those areas where they had declared at least “High” interests (in the first table)

- **1568 stakeholders** (researchers, clinicians, research funding agencies, policy makers, patients associations, etc.)
## I. Priority Areas

<table>
<thead>
<tr>
<th>Priority Areas</th>
<th>Priority</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No Interest</td>
</tr>
<tr>
<td><strong>1. Health Systems Research</strong></td>
<td></td>
</tr>
<tr>
<td><strong>2. Human and Social Science Research</strong></td>
<td></td>
</tr>
<tr>
<td><strong>3. Epidemiology/Natural History of diseases</strong></td>
<td></td>
</tr>
<tr>
<td><strong>4. Research in Genetics and Pathophysiology</strong></td>
<td></td>
</tr>
<tr>
<td><strong>5. Pre-Clinical Therapeutic Research</strong></td>
<td></td>
</tr>
<tr>
<td><strong>6. Therapeutic Research</strong></td>
<td></td>
</tr>
<tr>
<td><strong>7. Research Platforms/Infrastructures</strong></td>
<td></td>
</tr>
<tr>
<td><strong>8. Human Resources</strong></td>
<td></td>
</tr>
<tr>
<td><strong>9. Development and availability of tools needed to accelerate research on rare diseases</strong></td>
<td></td>
</tr>
<tr>
<td><strong>10. Others</strong>*</td>
<td></td>
</tr>
</tbody>
</table>
# Research in Genetics and physiopathology

<table>
<thead>
<tr>
<th>Research in Genetics and physiopathology</th>
<th>Priority</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No interest</td>
</tr>
<tr>
<td>a. Gene identification and Molecular Mechanisms</td>
<td>☐</td>
</tr>
<tr>
<td>b. New database method developments to be applied to omics research</td>
<td>☐</td>
</tr>
<tr>
<td>c. Biobanking research</td>
<td>☐</td>
</tr>
<tr>
<td>d. High throughput genomics/sequencing</td>
<td>☐</td>
</tr>
<tr>
<td>e. New bioinformatic methods and developments</td>
<td>☐</td>
</tr>
<tr>
<td>f. New technologies (e.g., atomic microscopy...)</td>
<td>☐</td>
</tr>
<tr>
<td>g. Omics technology (e.g., proteo, metabolo...)</td>
<td>☐</td>
</tr>
<tr>
<td>h. Development of new animal models</td>
<td>☐</td>
</tr>
<tr>
<td>i. New Imaging/Microscopy methods</td>
<td>☐</td>
</tr>
<tr>
<td>j. Development of new therapeutic tools</td>
<td>☐</td>
</tr>
<tr>
<td>k. Others*</td>
<td>☐</td>
</tr>
</tbody>
</table>

* If your answer is “Others”, please specify:
List of Priority Areas

1. Health Systems Research
2. Human and Social Science
3. Epidemiology/Natural History of diseases
4. Research in Genetics and Pathophysiology
5. Pre-clinical Therapeutic Research
6. Therapeutic Research
7. Research Platforms/Infrastructures
8. Human Resources
9. Development and availability of tools needed to accelerate research on Rare Diseases
Results
Research Priority Areas

- Research in Genetics and Pathophysiology
- Therapeutic Research
- Development & availability of tools to accelerate research on RD
- Epidemiology/Natural history of diseases
- Pre-Clinical Therapeutic Research
- Health Systems Research
- Human Resources
- Human and Social Science Research

[Bar chart showing percentage of interest in each area]
The Five Categories with the highest “No interest” rate

- Assessment of surgery procedures
- Transplants
- Bioproduction centres
- Drug Libraries
- High throughput drug screening

Values:
- 10.47
- 8.48
- 5.24
- 4.99
- 4.49
The Ten categories with the highest “Low Interest” rate

- Omics technology
- Development of new animal models
- New database method developments to be...
- Medical Devices
- Transplants
- Assessment of surgery procedures
- New bioinformatics methods and...
- New imaging/microscopy methods
- Bioproduction centres
- New technologies
The Five categories with the highest “High Interest” rate

- Gene identification and molecular mechanisms: 66.83%
- Development of new therapeutic tools: 82.29%
- Transnational calls for project funding: 70.32%
- Network development: 70.32%
- Creation of transnational research consortiums: 70.82%
The Five categories with the highest Patients Interest

- Phase I/II Clinical trials
- Drug Development
- Development of new therapeutic tools
- Assessment of surgery procedures
- Network development

Number vs Percentage
The Five categories with the highest Patients’ Interest

- **1st.** Phase I/II clinical Trials (100%)
- **2nd.** Drug development (91%)
- **3rd.** Development of new therapeutic tools (82%)
- **4th.** Assessment of surgery procedures (82%)
- **5th.** Network development (82%)

For the whole sample 22nd position with 53% of interest
For the whole sample 29th position with 49% of interest
For the whole sample 2nd position with 71% of interest
For the whole sample 61st position with 20% of interest
For the whole sample 5th position with 67% of interest
Bottleneck Analysis
Bottlenecks: Some examples

- **Funding issues and particularly,**
  - Limited budget
  - Lack of funding for promoting international cooperation projects
  - Lack of RD programmes research at national level
  - Lack of support for long-term projects (registries; cohort studies)
  - Comprehensive Biobanks in Europe
  - Specific infrastructures/platforms (developing and accessing)
  - Proof of concept studies funding
  - Clinical trails phase I/II
  - Align priorities and/or funding instruments at different levels and between them

- **Specific inequities/differences among type of diseases investigated**
  - Geographic inequalities on existing neonatal screening
  - Low priority for certain issues – congenital anomalies
  - Tailored solutions for some specific questions
  - Clinical aspects are usually disregarded
  - Chromosomopathies
Bottlenecks: Some examples

- Policy and ethical issues
  - Harmonization, sharing values and moral principles
  - Orphan drugs accessibility should be compulsory
  - International ethic rules for registries
  - Ethics rules and European legislation for clinical trials
  - Different ethics rules on biobanks
  - Fear to run new drugs due to side effects
  - Patients participation on priority decisions
  - No interest by hospital managers
  - Lack of Rotational issues and Training programmes
  - Project evaluators are not so well prepared
  - Compassionate tendency instead scientific approach
Bottlenecks: Some examples

- **Methodology issues pointed out as bottlenecks**
  - Epidemiological research
    - HTA methods do not fit with RD
    - Higher rates of false positives on neonatal screening
    - Lack of technical assistance for long-term studies
    - European registries
    - Limitations on developing a good work on large networks
    - Specific clinical trials methods
  - Private registries
  - Databases for Omics analysis
  - Difficult to get a good diagnosis
  - Common approach for some rare and common diseases
  - Basic methods
    - Animal models
    - Getting live cells from patients
    - In-vitro systems
    - New drugs and lack of knowledge of molecular mechanisms
    - Evaluation cell at morphologic level
    - Vectors production

- **Public/private cooperation**
- People working either at hospitals or non-profit research institutions
Messages to E-RARE

• ERARE- No limits in cooperation among countries
• ERARE- Enlarging this initiative
**Bottlenecks by Priority Areas**

Percentage

- Human and Social science research
- Human resources
- Health Systems research
- Development and availability of tools needed...
- Preclinical therapeutic research
- Criteria for prioritising
- Therapeutic diseases
- Research in Genetics and physiopathology
- Ep/Natural history of diseases
### Future themes and needs for rare diseases research funding: Recommendations

<table>
<thead>
<tr>
<th>To the EC</th>
<th>To E-Rare</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Increase funding</td>
<td>• Open calls (no thematic restriction)</td>
</tr>
<tr>
<td>• Need for a common (MS, EC) road map (led by E-Rare?)</td>
<td>• Focus on « translational » projects</td>
</tr>
<tr>
<td>• Promote RD as model for common diseases</td>
<td>• Promote interdisciplinary interactions (societal &amp; psychological issues)</td>
</tr>
<tr>
<td>• Ease mobility of clinicians (« protected time »)</td>
<td>• Promote social &amp; human sciences</td>
</tr>
<tr>
<td>• Decrease discontinuity of funding elements of translational research</td>
<td>• Address methodology bottlenecks</td>
</tr>
<tr>
<td>• Funding proof-of-concept</td>
<td>• Develop/support decentralized Techn. Platf.</td>
</tr>
<tr>
<td>• Promote (build-up and maintenance) of EU-funded research projects</td>
<td>• Fund possible rotation for clinicians into projects</td>
</tr>
<tr>
<td>• Adress current gaps in research on policies &amp; ethical regulations</td>
<td>• Maximize collaborative outputs (other EC pgr.)</td>
</tr>
<tr>
<td>• Adress inequalities (neglected RD)</td>
<td>• Work out funding rules to maximize the real commitment of MS available funds</td>
</tr>
</tbody>
</table>
List of Contributors

- Alejandro Ramírez (ISCIII; IIER)
- Ana Villaverde Hueso (ISCIII; CIBERER)
- Verónica Alonso (ISCIII; CIBERER)
- María José Carroquino (ISCIII; WHO-CC)
- Rafael de Ándres (ISCIII; FIS)
- Manuel Posada (ISCIII-IIER; WHO-CC; CIBERER)
- E-RARE partners
Congratulations for this Rare Diseases Day

Thank you for your attention!

www.e-rare.eu