The Haystack is Made of Needles: A Global View of Rare Diseases

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President, EspeRare Foundation
Executive Committee, IRDiRC
What are We Doing Here?

People are dying.

Are we going to talk for two days?

Spend tens of thousands of Euro on yet another meeting?

What will we have to show for it?
“You never change things by fighting existing reality. To change something, build a new model that makes the existing model obsolete.”

Buckminster Fuller

Are we ready to risk?
Because if we cannot risk what families risk everyday, then we will not make a difference.
This meeting is about action, not talk

200 new therapies

Genetic tests for most rare diseases

Let’s go...
Needles in Haystacks
But the haystack is made of needles...

There is no lack of participants!
The Irish Version

Like Trying to Find a 4-leaf Clover
## Culture Shift

<table>
<thead>
<tr>
<th>Industrial Age (old)</th>
<th>Information Age (new)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Control means of production</td>
<td>Open means of production</td>
</tr>
<tr>
<td>Based on scarcity</td>
<td>Based on abundance</td>
</tr>
<tr>
<td>Hierarchical / Command</td>
<td>Network / Collaboration</td>
</tr>
<tr>
<td>Linear / Sequential</td>
<td>Organic</td>
</tr>
<tr>
<td>Win / Lose</td>
<td>Win / Win</td>
</tr>
<tr>
<td>Materials</td>
<td>Information</td>
</tr>
</tbody>
</table>

...but biomedical research and healthcare lag
Maturing Participation

- **1950s-1960s – Medical Models**
  - Voluntary Health Organizations

- **1970s – Nascent Patient Movement – Missing Services**
  - Self-organized Disease Specific Organizations

- **1980s – Maturing Patient Movement – IS & IT Technology**
  - New Alliances and New Strategies Emerge

- **1990s – Powerful Momentum “Patient Power” – Websites & Email**
  - Institutionalized Advocacy Coalitions
  - Patient Organized Networked Research Organizations
  - Effecting Broad Change of Public Policy

- **2000s – Successful Models “Research Advocacy” – BioBanks**
  - Active Engagement in the Research Enterprise
  - Breaking Conventional Boundaries of the Medical Model
  - Demand for Quality, Services, Choice, & Personalized Delivery
  - Patient Rights Public Policy – Changing the Status-Quo

- **2010s – Smart Networks in the Commons – Translation & Delivery**
  - Participatory, dynamic, long tail, precise
### The world has changed

<table>
<thead>
<tr>
<th>Change</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Single Condition</td>
<td>Multi-condition; cross-disease</td>
</tr>
<tr>
<td>Siloed collections</td>
<td>Models for shared infrastructure</td>
</tr>
<tr>
<td>Individual governance</td>
<td>Group governance</td>
</tr>
<tr>
<td>Limited standardization</td>
<td>Increased standardized protocols for data</td>
</tr>
<tr>
<td>Limited data</td>
<td>Robust data from multiple sources</td>
</tr>
<tr>
<td>Limited role of ‘subjects’</td>
<td>Dynamic involvement of public / advocates/ citizen scientists</td>
</tr>
</tbody>
</table>
People hold the keys, how about we also participate in solving the puzzles?

Visionary leadership
Shared infrastructure
Higher power
Lower costs
Dirty data gets cleaner

Current fragmentation
Redundant infrastructure
Low power
High costs
What do personal data collection tools look like in 2013?
The Public

Network effects for tools and analysis

Conversion

This needs support: financial, tech solutions, leadership

Participants Drive Collection and Sharing
In this **new age**: we have the **tools** to address the problem of **fragmented systems**, and still we **lose opportunities** every day.

Data can accelerate **research and services**, help us to get **better patient outcomes**, and **save money**...
This team crystalized this model – which integrates some things that are already built, some that are offered and some we need to find.
Join Reg4ALL

13,000 disease pages
Hundreds of thousands of visitors
High SEO

Pharma and Biotech analyzing data to choose diseases
This team crystalized this model – which integrates some things that are already built, some that are offered and some we need to find.
<table>
<thead>
<tr>
<th>Privacy Setting</th>
<th>Discover My Data</th>
<th>Contact Me</th>
<th>Use My Data</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Advocacy &amp; Support Groups</strong></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>D3B-listed organizations serving your condition</td>
<td>✓ Allow</td>
<td>✓ Allow</td>
<td>✓ Ask Me</td>
</tr>
<tr>
<td>Non-profit organizations serving your condition</td>
<td>✓ Allow</td>
<td>✓ Ask Me</td>
<td>✓ Ask Me</td>
</tr>
<tr>
<td><strong>Medical Researchers</strong></td>
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</tr>
<tr>
<td>Researchers recommended by a D3B-listed organization serving your condition</td>
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<td>✓ Allow</td>
<td>✓ Allow</td>
</tr>
<tr>
<td>IRB-approved research addressing your condition</td>
<td>✓ Allow</td>
<td>✓ Ask Me</td>
<td>✓ Allow</td>
</tr>
<tr>
<td>All researchers</td>
<td>✓ Allow</td>
<td>✓ Ask Me</td>
<td>✓ Ask Me</td>
</tr>
<tr>
<td><strong>Data Analysis</strong></td>
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<td></td>
</tr>
<tr>
<td>&quot;Compare with others&quot; feature</td>
<td>N/A</td>
<td>N/A</td>
<td>✓ Allow</td>
</tr>
<tr>
<td>&quot;Show related content&quot; feature</td>
<td>N/A</td>
<td>N/A</td>
<td>✓ Allow</td>
</tr>
<tr>
<td>Genetic Alliance Transnational Research Network</td>
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<td>✓ Allow</td>
</tr>
<tr>
<td>Oracle Health Sciences Network</td>
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<td>N/A</td>
<td>✓ Ask Me</td>
</tr>
<tr>
<td>Newly-Released Data Analysis Platforms</td>
<td>✓ Ask Me</td>
<td>N/A</td>
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The EspeRare Foundation: Advancing treatments for Rare Diseases -together

Patient-centricity & pharma R&D expertise to reposition drugs to address underserved needs of Rare Disease Patients

**Status**

Esperare is a nonprofit private organisation established in Switzerland in March 2013.

**Mission**

In collaboration with patient groups and other key stakeholders, we uncover the potential of existing drugs to address severe therapeutic unmet needs in rare diseases.

- **Identify (re)positioning opportunities** with high therapeutic value potential & clear path to reach rare disease patients
- **Focus on preclinical & early clinical validation** by bridging the existing translational gap
- **Define clear clinical & commercial path forward**

**Goals**

**Activities**

Our first effort focuses on developing Rimeporide, a Merck Serono molecule with the potential to slow muscle degeneration in children with Duchenne muscular dystrophy, a genetic and fatal rare disease.
Jon Baumhauer, Chairman of the
Family Board and of the Executive Board of E. Merck KG
Operating model & initial program

ACCESS
Disease Advocacy Organizations

EXECUTION
Preclinical & Clinical Reference Centers

FOUNDATION
- Drive R&D activities
- Alliances mgmt
- Funding strategy

STRATEGY
Clinical & Regulatory Advisory Board

First Program
1. Rimeporide in Duchenne's muscular dystrophy
   - Disease modifying agent

2. Identification of new opportunities
   - Creating partnerships (pharma, biotech, academia...)

For more: www.esperare.org