“The Patient’s Perspective”

Lorna Speid, Ph.D., M.R.Pharm.S.  
Founder and President  
Putting Rare Diseases Patients First!

ASCPT 2016
Outline

• The World of the Rare Diseases Patient
• Patient Engagement and Empowerment
• Making an Impact: Your Role as Researchers and Drug Developers
The World of the Rare Diseases Patient
The World of the Rare Diseases
Patient

Diagnosis
The World of the Rare Diseases
Patient

Treatment
The World of the Rare Diseases
Parent
The World of the Rare Diseases Patient

Help
Help

Pharma Biotech
Academia Government
Foundations Patient Organizations

Very little output
Help

Pharma Biotech

Academia Government

Foundations Patient Organizations

Very little output
Patient Engagement and Empowerment
Engagement and Empowerment
Patient Empowerment

- PRDPF! Formed in January 2014
- 501 (c ) (3) organization, charity status
- Tax exempt status in California and from the IRS
- Global outreach
- Free educational content
  - Webinar 001 - *The Seven Things You Need to Know about the Drug Development Process For Rare Diseases*, Dr. Lorna Speid, PRDPF!
  - Webinar 002 - “Will Someone Please Tell Me What is Wrong With Me?” – Dr. Timothy McDaniel, now with TGEN
  - Webinar 003 - “The Rare Diseases Patient at the Center of it All” – Dr. John Whyte, FDA
  - Webinar 004 – “The Rare Diseases Patient at the Center of it All – Part 2” – Patient Engagement – Dr. Sangeetha Jethwa, Roche
  - Webinar 005 - “Natural History Studies – Making a Contribution” - Part 1 – Laying a Foundation - Dr. Nuria Carrillo, NIH-NCATS
  - Webinar 006 – “Natural History Studies and Your Rare Disease Child” – Part 2 –Dr. Ann Barbier, Agios Pharmaceuticals and Professor Morton Cowan, UCSF
Patient Empowerment

PRDPF! Facebook page: https://www.facebook.com/puttingrarediseasepatientsfirst

PRDPF! Twitter: https://twitter.com/PuttingRDPF

PRDPF! Blog: https://rarediseases123.wordpress.com/
Engagement: Risk Benefit Assessment
### Engagement: Risk Benefit Assessments

#### Patient Scenarios

<table>
<thead>
<tr>
<th>Adult male or female</th>
<th>Adult patient</th>
<th>The disease afflicts children</th>
</tr>
</thead>
<tbody>
<tr>
<td>Recent diagnosis</td>
<td>Diagnosed some years ago</td>
<td>Numbers of children impacted are small</td>
</tr>
<tr>
<td>No known treatment</td>
<td>Treatment is suboptimal or only treats symptoms</td>
<td>No effective treatments</td>
</tr>
<tr>
<td>No natural history studies</td>
<td>Disease is slowly progressing</td>
<td>Rapidly progressing</td>
</tr>
<tr>
<td>Rapidly progressing</td>
<td>Lifespan may or may not be shortened</td>
<td>Shortened lifespan</td>
</tr>
<tr>
<td>Shortened lifespan</td>
<td>Natural history of the disease is known</td>
<td></td>
</tr>
</tbody>
</table>

- Adult male or female
- Recent diagnosis
- No known treatment
- No natural history studies
- Rapidly progressing
- Shortened lifespan

- Adult patient
- Diagnosed some years ago
- Treatment is suboptimal or only treats symptoms
- Disease is slowly progressing
- Lifespan may or may not be shortened
- Natural history of the disease is known

- The disease afflicts children
- Numbers of children impacted are small
- No effective treatments
- Rapidly progressing
- Shortened lifespan
Making an Impact
<table>
<thead>
<tr>
<th>Drug Discovery</th>
<th>Preclinical R&amp;D</th>
<th>Clinical Studies</th>
<th>Post Marketing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Screening</td>
<td></td>
<td>Phase 1</td>
<td>Phase 4 Studies</td>
</tr>
<tr>
<td>Chemistry</td>
<td></td>
<td>Phase 2</td>
<td>REMs</td>
</tr>
<tr>
<td>Biology</td>
<td></td>
<td>Phase 3</td>
<td>PSURs</td>
</tr>
<tr>
<td>DMPK</td>
<td></td>
<td>Pediatric Development</td>
<td>New Indications</td>
</tr>
<tr>
<td>Formulation Development</td>
<td>Process Development</td>
<td>Accelerated Approval</td>
<td>505(b) (2)</td>
</tr>
<tr>
<td></td>
<td>Synthesis</td>
<td></td>
<td>Generics</td>
</tr>
<tr>
<td></td>
<td>Scale Up of Active Substance</td>
<td></td>
<td>Biosimilars</td>
</tr>
<tr>
<td></td>
<td>Formulation Development</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Animal Efficacy Studies</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Tox Testing</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Short Term</td>
<td>Long Term</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Health Outcome Studies</td>
<td></td>
</tr>
</tbody>
</table>

Sponsor / FDA & EMA meetings encouraged for scientific advice
FDA / EMA Time
Start diagnostic/biomarker development

IND/CTA Submission
NDA/MAA Submission
Review Decision
Sponsor Answers
Review questions

Pediatric Development
Health Outcome Studies

Start diagnostic/biomarker development
Patient Involvement
Pharma, Biotech, and Academia
Engaging with Patients

• Engage with Patients
  – Targeted Product Profile
  – Global Regulatory Strategy – *Use regulatory mechanisms to speed up access to commercialization for patients*
    – Well designed clinical trials
    – Well designed development programs
  – Right patients – Representative
    – Using the technologies available to us – right sized studies
    – Re-engineering our industry

• Reimbursement

• Get it Right First Time – Fastest Time to Market
Patient Involvement – What Does it Look Like?

• Risk benefit assessment programs
• Natural History Studies – design and participation
• Patient Registries
• Legislation
• Regulatory Authorities
• Contribution to the design of clinical trials
• Ensuring there is sufficient support for patients in the trenches
Regulatory Environment

• FDA www.fda.gov
  – Patient Liaison Program
  – Patient Network Website
  – Patient Reported Outcome
  – Patient Focused Drug Development Meetings
Regulatory Environment

www.ema.Europa.eu
Guidance for Best Practices

• No guidelines from FDA or EMA at this time
  – Be compliant with guidelines – when they are issued

• Care
  – Conflicts of interest
  – Vulnerability
  – Representative patients
  – Biases

• Templates
  – Don’t reinvent the wheel
  – Learn from other disease areas
  – Engage with organizations that are already going through the process
  – Speak to real patients and parents of children with the disease in question

• Specially Trained Personnel
  – Pharmacists
  – Physicians
  – Nurses