

RARE 



Research for Rare Diseases Challenges and Opportunities

Summary of Discussions from Break-out Group

Data Sharing

Barriers

- Lack of common data elements
- Lack of policy
- Limitations in consent forms
- Academic disincentives to sharing
- Lack of guidelines and best practices (e.g. publication policies)
- Multiple IRB reviews in multi-site studies

Opportunities

- Existing infrastructure (e.g., H3 Africa)
- International partnerships (e.g., ICORD)
- Patients support data sharing
- Other stakeholders (governments, industry) need data

Patient Engagement

Barriers

- Lack of information/pathways
- Stigmatization
- Cultural/language/religious barriers
- Lack of funding/support
- Uncertainty how to find accurate and reliable information online
- Fragmentation of patient communities, geographic dispersion of patients

Opportunities

- Forming umbrella organizations, unified voice, “lumping rather than splitting”
- Social media, mobile technology such as smart phones
- Active partnership throughout the research process (e.g. protocol development groups, IRBs)

Sustainable/Scalable Models

Barriers

- Misalignment of stakeholder pathways and incentives
- Lack of national and international coordination of priorities
- Lack of harmonized policies
- Duplication of efforts and re-inventing the wheel
- Rare disease “myth” = not a priority in developing countries, not a significant public health concern

Opportunities

- Sharing of resources
- Benefit of studying indigenous populations
- Involving patients as disease experts
- Social media and mobile technologies
- Cutting-edge bioinformatics and technological solutions for data management
- Re-phrase the goal: Not a way to study rare disease only, but to make progress on common diseases

Thank you

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