Day two-breakout session B.1
Chairs: Amy Farber, Kate McCurdy & Paul Harris
Note takers: Kate McCurdy & Jennifer Farmer

B.1 Patient Participation / Outreach Activities & Patient Advocacy

Patient registries may serve as a catalyst for new scientific hypotheses, lower the burden of identifying study participants and ultimately play a significant role in speeding and advancing disease-specific and cross-disease translational research. What is the role of advocacy organizations and patient and patient family member participation in disease-specific and cross-disease registries? How do we maximally incentivize participation and recruitment? What are some of the critical lessons learned and areas of collective strength that we might further consider in mapping out next steps?

Registries are only as valuable as the information they contain. As a result, the success of each registry depends on the patients involved. Patient participation is the fundamental requirement for any registry, and so this breakout session will focus on the very important issues related to the possible benefits to, motivations for and means of achieving optimal patient participation.

Similarly, the success of a network of registries, such as the one being contemplated here, and the vast power that can come from future communication between these registries, is dependent on the integrity of the registries that are linked and standardization of data among them. This breakout session will also include discussion about contributions of registries to scientific and medical advances and therefore their value to advocacy groups focused on improving the lives of those affected by rare diseases.

Some of the issues to be discussed and questions to be addressed will include:

1. How can patients with rare diseases, their family members and advocacy organizations benefit from patient registries?

2. How can we incentivize patients and advocacy groups to participate in rare disease registries?

3. How can we incentivize all key stakeholders (for this discussion, especially patients, their physicians, advocacy groups and researchers) to participate in and utilize rare disease registries?

4. Can we envision ways to include patients with diseases for which there are neither existing advocacy organizations nor patient registries?
5. What must each of the advocacy groups do individually and collectively to help successfully establish a coordinated umbrella rare disease patient registry?

6. How can we best communicate information (research findings, new initiatives, existence of support groups) back to registry participants, families and advocacy groups to keep them interested and informed?

7. How can we maximize recruitment of patients into clinical research studies?

8. How can we ensure that recruitment of patients is effective and reaches the maximum number of patients?

9. How can we ensure that data, test results and research findings are properly and effectively delivered and communicated to the patient and the patient advocacy groups?

10. What can be done to increase trust and responsiveness between patients, physicians and research teams?

11. What can we, the patient advocacy groups, do to be better informed about recent developments within the research and medical arena?

12. What are the pros/cons of “patient-initiated research” versus “investigator-initiated research”?

13. What should we do to increase tissue and specimen procurement in rare disease patients?

14. What are the main obstacles and/or challenges from a patient and rare disease group point of view that must be addressed in order to ensure the success of this project?
Breakout Session B1

*Patient Participation/ Outreach Activities and Patient Advocacy*

**Chairs:** Amy Farber, Ph.D., Kate McCurdy & Paul Harris, Ph.D.  
**Note taker:** Kate McCurdy, Jennifer Farmer, M.S.  
**Discussion Panel:**  
Ron Bartek (FARA)  
Jennifer Farmer, M. S., (FARA)  
Amy Farber, Ph.D., (LAM Treatment Alliance)  
Leslie Gordon, M.D., (Progeria Research Foundation)  
Lynn Etheredge (Rapid Learning Project, George Washington University)  
Paul Harris, Ph.D., (Vanderbilt Institute for Clinical and Translational Research)

**Caveats:**

1. Topic was *Patient Participation/ Outreach Activities and Patient Advocacy* but scope drifted, important and somewhat bounded ways:
2. We were cognizant of who was *not* in the room at this meeting and tried to be aware of selection bias that might make the profiles of the orgs represented in this meeting context more homogenous and less representative of the rare disease patient and advocacy group community at large;
3. We were also aware of groups and patients not tapped into advocacy groups and registries and not necessarily motivated by the things that make recruitment relatively easy for the vast majority of advocacy groups represented at this meeting who find patient identification a far greater challenge. In other words, we need to remember that unmet medical need within the rare disease community often also impacts those who qualify as under-represented and underserved minorities in other ways.

**Who was in our session?**
- Approximately 50% patients/patient advocates; approx. 20% clinicians; 20% researchers and approximately 2 ethicists; 2 regulatory folks; 1 person from pharma and we estimate a few IT/engineer/platform development folks.

**Where do the registry projects stand among those who participated in this session?**
- Approximately ½ of patient advocacy groups had a registry; a little fewer than half had a registry in development; a few had no registry at all; we didn’t
hear from patients without advocacy orgs or registries but we suspect they exist, even if not in the room; there was one researcher without an advocacy org;

**Session Format:**
We divided our time into two parts: a) the first focused on disease-specific registries and related concerns and successes on the Patient Participation/Outreach Activities and Patient Advocacy front and part b) the second part focused on the Network of Registries and felt needs and concerns as they relate to Patient Participation/Outreach Activities and Patient Advocacy.

**Disease specific registries:**

**Registry Scope:**
- Defining what “IT” / “the registry” in question was, even at the disease-specific level, was an ongoing out-the-gate focus as we delved into our subject.
- We asked people to describe the scope of their registry projects when they discussed their struggles and successes with patient participation/outreach activities and patient advocacy at the disease-specific registry level to be able to better understand the goals behind the various patient outreach efforts.
  
  **Findings:**
  - Of existing registries, only one or two have demographics info only;
  - Other existing registries covered some continuum of patient natural history and clinical data; many are curated; many have at least one patient entered data component; many enable hypothesis generation; and many focus on and were initiated in order to facilitate trial recruitment;
  - Some are struggling to incorporate clinician-entered data/clinical data due to costs of data entry and curation;
  - Most had biorepositories;
  - Most covered basic disease characterization and history;

**Concerns:**
- Problem with patient identification;
- Less problem with recruitment to their registry for initial contact;
- Challenge with follow-up over time;
- Need more carrots for burned out patients and patient families;
- Maintaining trust;
- Hard work of ongoing communication to remain trust and build community;
- Language and cultural barriers;
- Incentivizing clinicians/care teams to be involved and to refer patients to registries;
- Frustration – we all have things in our databases that could help each other at this disease specific level;
- No list or inventory of registries, their scope, what resources exist from that that could be shared and some quality assessment – to facilitate sharing at disease specific level (NORD said they would provide); connections and best practices; Need a one stop – well known and legitimate space where for listing these advocacies; dissemination of that... ;
- Getting clinical data into registries is expensive;
- Still need for registry-building for dummies;

**Successes:**
- Recruitment: because seems recruitment is a form of communication that helps build community; cultivates buy-in in communicating direct value of self report; benefits of participation address lifestyle issues; helps with treatment guidelines; relatively easy to get people in; some caveat, however, in re to Social and Economic Status (SES): computer literacy; Internet access;
- Many have started small and have been able to evolve in scope and value; build in a way that can morph/ evolve;

**Exciting potential prospects at both disease specific and Network of Registries level:**
- Prospect EMR facilitating patient identification and recruitment (?);
- Ways of capturing information that isn’t otherwise captured: modules such as diaries;
- Value and rewards of self report data: patient empowerment - patients as participants in own healing and research; patients when children...establishes a tradition of participation early on);
- Good continuity where feed info back; nurture relationship;
- Empowers patients and gives sense of community;
- Economies of scale;
- Venn diagram – we are all intersections of multiple rare diseases and some common diseases;

**Middle level existing registry networks** - clustering already underway between diseases that are understood to be related in some way;

**Uber Registry: Network of Registries**

**Desires:**
- People don’t have one disease; they have many:
- To drive awareness – an advocacy tool / means of answering questions pertinent to all or many rare diseases;
- A place to learn share across diseases;
- All want some clinical information;
- Power in numbers;
• Economy of scale;
• Absence of overlaps is telling but otherwise difficult to explore empirically;

Concerns:
• What is “it”? Defining goals and scope!
  o How much data/ info to go in?
  o Timing – can we wait? Should we share and implement at the disease specific level instead and in an organized way? We only have so many hours in the day... Timing; limited time in which we can invest;
  o Cross disease sharing? Some already clustering; some just want to leverage or borrow modules and link into existing disease specific registries;
  o How to work with registry we have and do more/ do what we can
  o Governance: Access, control ownership;
  o Cost?
  o Regulatory compliance/ ethics
  o Longevity
  o Credit for advocacy orgs