

Day one-Keynote address
Jonathan Moreno

Advancing rare disease research: ethical dimensions

A little historical orientation to the field of bioethics, esp. re values of privacy/confidentiality and patient/subject self-determination

- ❖ Where the field is now RE facilitating clinical trials vs patient/subject's rights
- ❖ Benefits of registries
- ❖ Risks of registries
- ❖ GINA and HIPAA
- ❖ Relationships between pharma and patients with rare diseases
- ❖ Institutional innovations--Looking ahead re rare diseases
- ❖ Cloud computing, social networking
- ❖ Prediction markets
- ❖ Implications of more powerful microarray technology for research resource allocation



Uniting Rare Diseases

*Advancing Rare Disease Research:
The Intersection of Patient Registries, Biospecimen Repositories
and Clinical Data*

Keynote Address

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*Advancing Rare Disease Research:
Ethical Dimensions*

Mom



Bioethics, Then and Now

- Traditional medical ethics – professional virtues, trade rules, confidentiality
- Modern bioethics – truth telling, informed consent
 - Prophetic bioethics
 - Regulatory bioethics
- Clinical ethics
- Research ethics
- Public health ethics
- Science ethics

Clinical trials: Protection and access

- Early focus was on protecting potential subjects from high risk/no benefit studies
- HIV/AIDS era created more interest in access to trials; also women's health movement
- Special issue for rare disease research
 - Often close relationships between investigators, sponsors, patients, families
 - Can blur roles, confuse communication

Rare Disease Registries (1)

- Rare disease research can especially benefit from data sharing (E.g., open data base for DMD)
- Protect donor privacy by limiting phenotypic information to alterations for that disorder
- Key held by trustee
- NIH and Wellcome Trust (2003) require grantees to make data available to other investigators for collaborative analysis

Rare Disease Registries (2)

- But risk of identifiability is growing due to proliferation of genetic data sets
- This could trigger HIPAA rule and trump open access
- GINA is a partial protection against discrimination but does not cover life, disability, or long-term care insurance
- Burden of proof would be on donor

Innovative Technologies

- Need to look ahead to the way technology will change research environment
 - Cloud computing/Social networking
 - Prediction markets
 - DNA microarrays and resource allocation

Implications

Change meaning of "disease" and "patient", more truly participatory research?

General awareness that we all have rare diseases