SHAWN M. SWEENY, PHD
HEALTH RESEARCH ALLIANCE
MEETING
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What is GENIE?

- AACR Project Genetics Evidence Neoplasia Information Exchange is an international cancer registry
  - Conceptualized by Charles L. Sawyers, MD (MSKCC)

- The project goals are to:
  - Improve clinical decision-making by linking clinical genotypes with clinical phenotypes
  - Catalyze clinical and translational research

- The data will be made publicly accessible after defined periods of exclusivity

www.aacr.org/genie
The GENIE Registry

Aggregate tumor-only NGS data & limited clinical data set from project Participants into registry

Clinical question asked

Necessary clinical information linked to data within the registry

Data made publicly available after defined periods.

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Why An International Cancer Registry?

CANCER is largely a disease of acquired genetic mutations

| ~13 | Genes are mutated in >5 % of patients |
| ~78 | Genes are mutated in 1-2 % of patients |
| 100s | Genes are mutated in <1% of patients |
| 🌈 | Most tumors have multiple mutations occurring even less frequently |
| 🚫 | No single institution is likely to have enough data to make informed clinical decisions |

SHARING DATA IS THE SOLUTION

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Based on Lawrence, MS, et al., Nature. 2014
Why Now?

Cost per Genome

Moore's Law

National Human Genome Research Institute
genome.gov/sequencingcosts

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Why Now?

Growth of DNA Sequencing

- Recorded growth
- Double every 7 months (Historical growth rate)
- Double every 12 months (Summa Estimate)
- Double every 18 months (Moore's Law)

Successful Data Sharing

- GENIE harmonizes the participants’ datasets through bioinformatics without the need for a common platform.

- Germline data is excluded in order to facilitate sharing fully de-identified data.

- Each participant retains exclusive access to their data prior to consortium access and public access.

- Each participant retains exclusive access to its medical records and only the necessary clinical data elements are shared.

- Three subcommittees manage essential processes:
  - Data Use and Membership
  - Concept Proposals and Publication
  - Data Standards

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Phase I Participants

New Participants Will be Accepted in Late Phase I Once Financial Milestones Are Met!

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Governance

AACR Leadership

Member Centers 1-7

AACR Special Committees

External Advisory Board

GENIE Steering Committee

Coordinating Center (AACR)

Project Lead

Project Manager

Staff Associate

Strategic Partner

SAGE

Scientific Advisor

Data Scientist

cBioPortal

Software Engineer/Bioinformatician

- Data standards
- Concept proposals & publications
- Membership and data access

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Data (All Types) Workflow

- HIPPA compliant
- Cloud & Local Instances
- 7 Members

- Limited PHI will be deleted following de-identification, and certification provided.
- Data will be updated quarterly
  - Prior datasets will be overwritten by new submissions.

De-identified data

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Longitudinal Clinical Data Availability Timeline

**Long-term Clinical Data**
- Prior treatment
- Other disease-specific questions
- Most recent therapy
- Exposure to standard-of-care therapies
- Relevant past medical history

- Post-sequencing treatments
- Off-label therapeutics
- Assignment to clinical trials
- Date of last follow-up or death
- Therapeutic response data
- Others as appropriate

**Diagram**
- Manual Curation of Medical Records
- Institution-only Access
- Consortium-only Access
- Manuscript preparation
- Community access

Time to access:
- 3 months
- 9 months
- 15-24 months

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AACR Project GENIE is an international cancer registry composed of CLIA/ISO-certified tumor sequences from patients treated at:

- The Center for Personalized Cancer Treatment (CPCT), The Netherlands
- Dana Farber Cancer Institute
- Institut Gustave Roussy, France
- Kimmel Cancer Center, Johns Hopkins University
- Memorial Sloan Kettering Cancer Center
- Princess Margaret Cancer Center, Canada
- Vanderbilt-Ingram Cancer Center
- New participants will be added in the next phase of the project

The goal is to improve clinical decision-making, and catalyze clinical and translational research by linking longitudinal outcomes of patients treated at these centers with their genotypes in response to specific clinical questions.

The data will be made publically available after defined periods of exclusivity.
Conclusion

- Thank You
- Questions?
- Comments?
- Observations?