



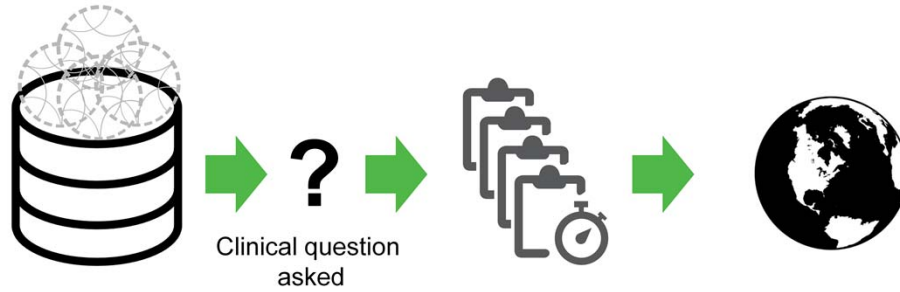
**PROJECTGENIE**  
Genomics Evidence Neoplasia Information Exchange

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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



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

Necessary clinical information linked to data within the registry

Data made publicly available after defined periods.



**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

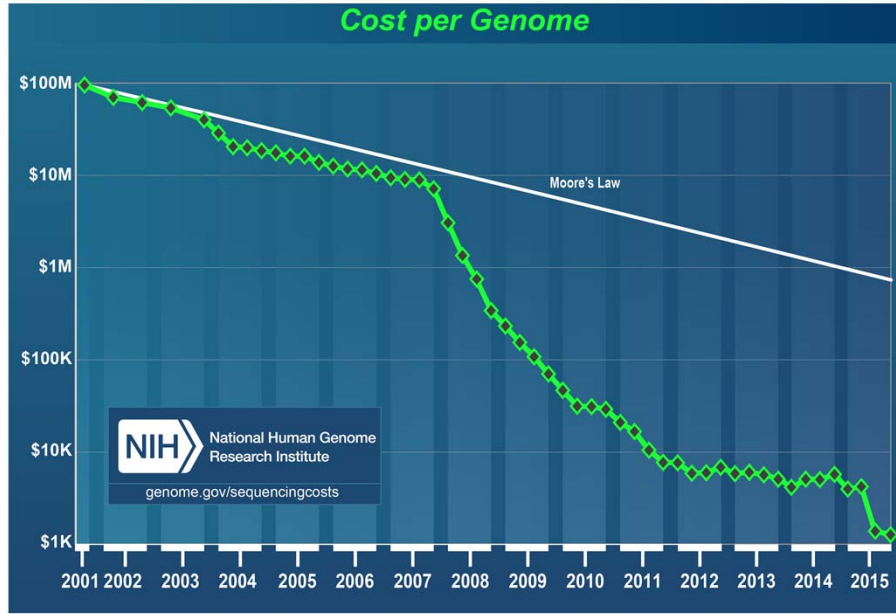


No single institution is likely to have enough  
data to make informed clinical decisions

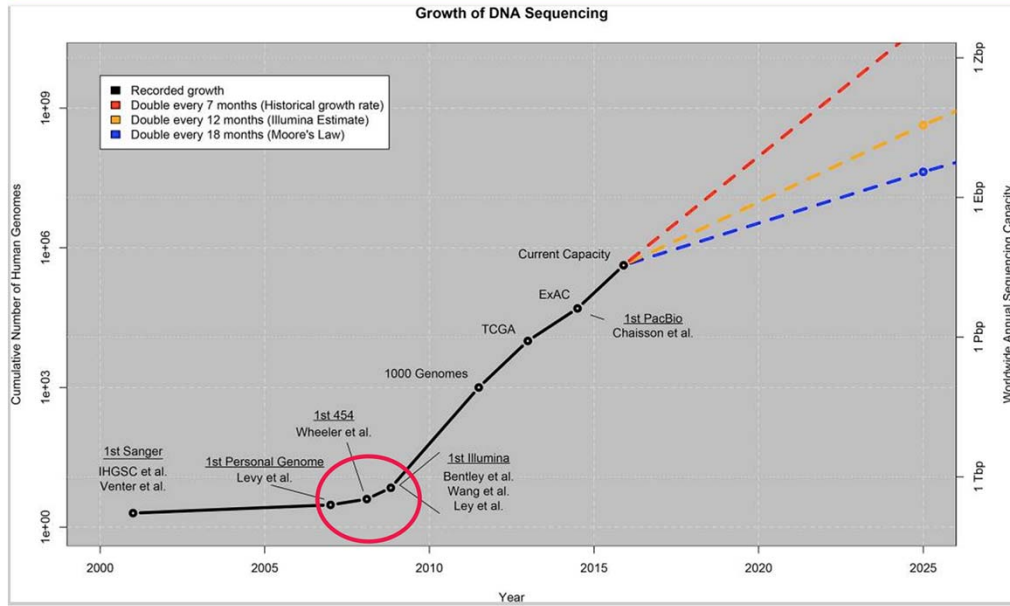


**SHARING DATA IS THE SOLUTION**

# Why Now?



# Why Now?



[www.aacr.org/genie](http://www.aacr.org/genie)

Stephens, Z.D., et al., PLoS Biol. 2015 Jul; 13(7): e1002195.

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- 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

## Phase I Participants

**AACR** American Association  
for Cancer Research  
FINDING CURES TOGETHER™



Memorial Sloan Kettering  
Cancer Center.



DANA-FARBER  
CANCER INSTITUTE

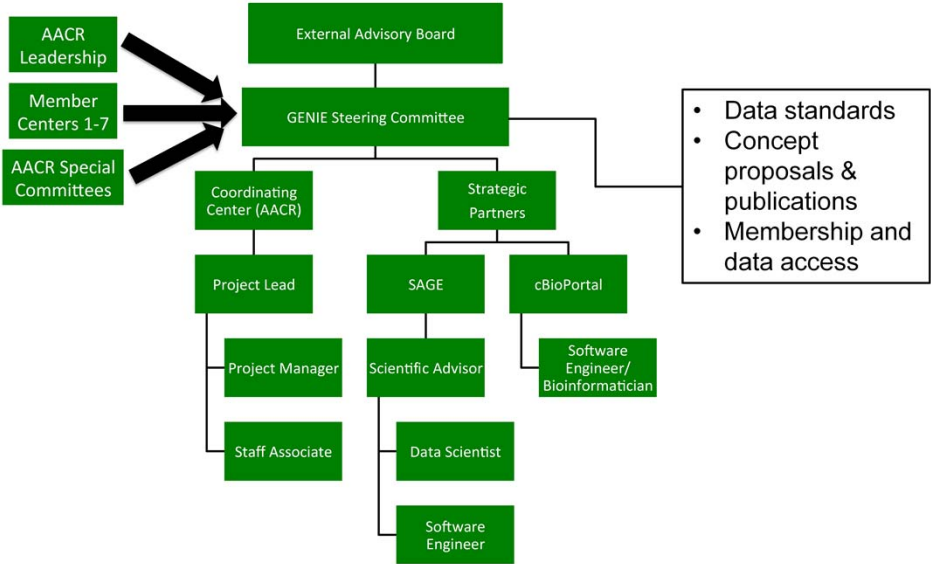


VANDERBILT UNIVERSITY

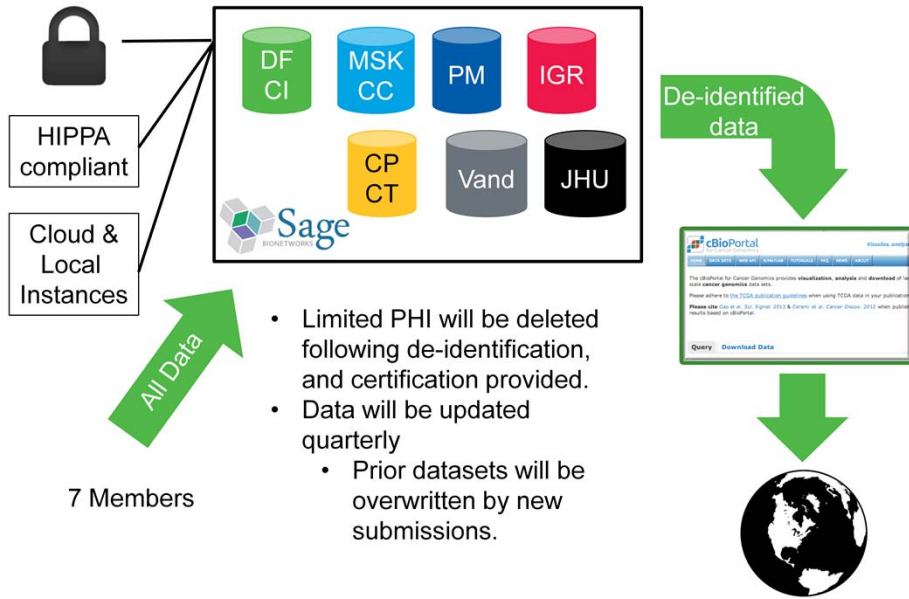
New Participants Will be Accepted in Late Phase I  
Once Financial Milestones Are Met!







## Data (All Types) Workflow



AACR Project GENIE Query this study  
AACR Project GENIE (Genomics Evidence Neoplasia Information Exchange) 10656 samples from 10347 patients.

Study Summary Clinical Data Mutated Genes

Samples selected: 10656 query genes - click to expand Query Select cases by IDs Add Chart

Cancer Type	#	Freq
Non-Small Cell Lung Cancer	1539	14.4%
NA	1312	12.3%
Breast Carcinoma	1081	10.1%
Colorectal Cancer	811	7.6%
Glioma	592	5.6%
Prostate Cancer	551	5.2%
Bladder Cancer	393	3.7%
Soft Tissue Sarcoma	353	3.3%
Renal Cell Carcinoma	296	2.8%
Esohooeoastric Carcinoma	293	2.7%

Cancer Type Detailed	#	Freq
NA	1312	12.3%
Lung Adenocarcinoma	1251	11.7%
Breast Invasive Ductal Card...	683	6.4%
Prostate Adenocarcinoma	535	5.0%
Colon Adenocarcinoma	530	5.0%
Bladder Urethelial Carcinoma	308	2.9%
Pancreatic Adenocarcinoma	170	1.6%
Lung Squamous Cell Carcinoma	166	1.6%
Colorectal Adenocarcinoma	163	1.5%
Glolioblastoma Multiforme	155	1.5%

Gene	# Mut	#	Freq
TP53	7452	4404	41.3%
KRAS	1679	1544	14.5%
PIK3CA	1673	1403	13.2%
APC	2023	1209	11.3%
TERT	1002	946	8.9%
KMT2D	1171	819	7.7%
PTEN	2006	809	7.6%
ARID1A	893	729	6.8%
ATM	877	664	6.2%
EGFR	919	651	6.1%

Gene	Cytoband	CNA	#	Freq
CDKN2B	9p21	DEL	543	5.1%
CDKN...	9p21	DEL	450	4.2%
CDKN...	9p21	DEL	445	4.2%
CCND1	11q13	AMP	341	3.2%

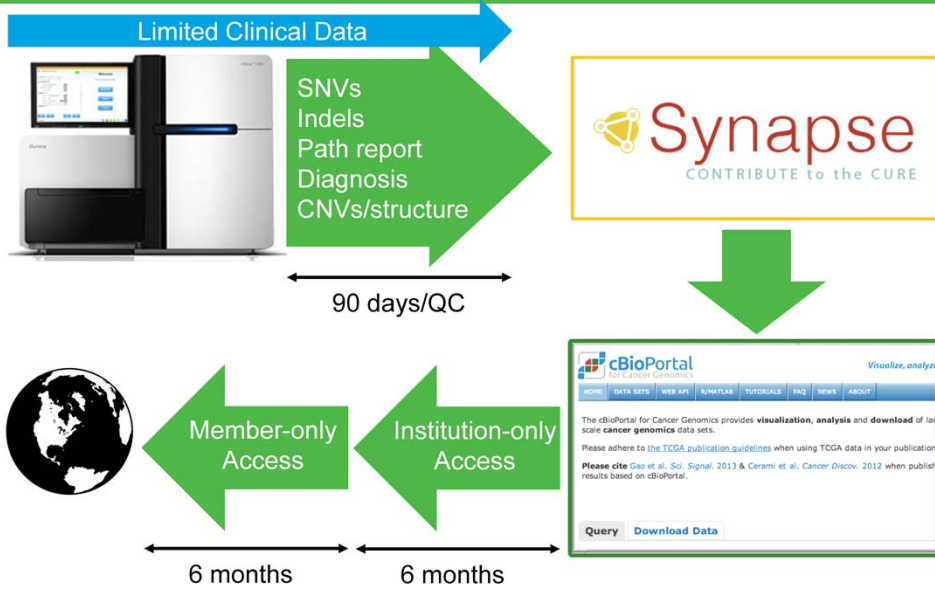
Sex

Sample Type

Primary Tumor Site

Metastatic Site

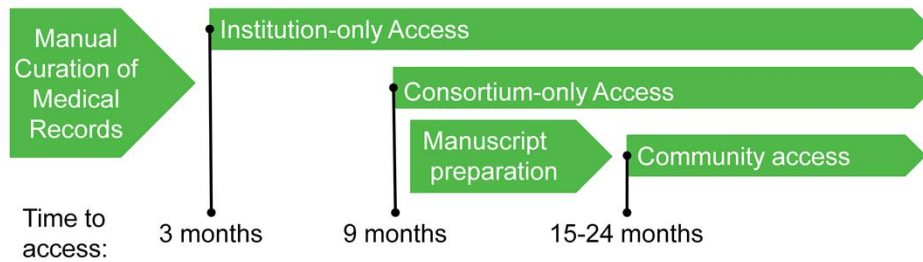
# Genomic/Limited Clinical Data Data Sharing Workflow



## 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



## Summary

- 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?