Translational Bioinformatics Definitions:

PLOS Computational Biology:

Translational bioinformatics is an emerging field that addresses the current challenges of integrating increasingly voluminous amounts of molecular and clinical data.

Its aim is to provide a better understanding of the molecular basis of disease, which in turn will inform clinical practice and ultimately improve human health

Main Goal of Translational Bioinformatics: to enable Translational Research

"A major challenge for translational medicine is to connect the molecular/cellular world with the clinical world"

Dr. Russ B. Altman
Department of Genetics, Stanford University

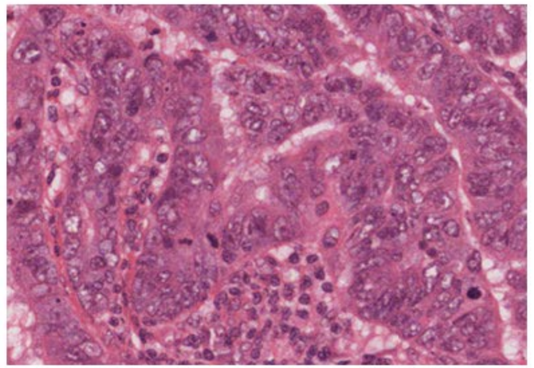
Focusing on Translational Cancer Research and Precision Medicine

- What Translational Bioinformatics can do for Cancer Research?
- G-DOC – a new platform for data management, integration and analysis in Translational Cancer Research and Precision (Personalized) Medicine

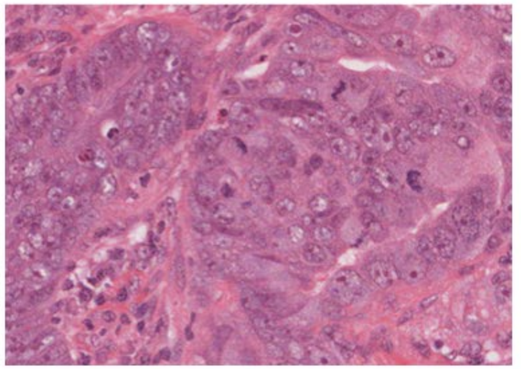
Can genome-wide molecular profiles help diagnose cancer?

Colorectal Adenocarcinoma under the microscope

Pathology sees ONE cancer



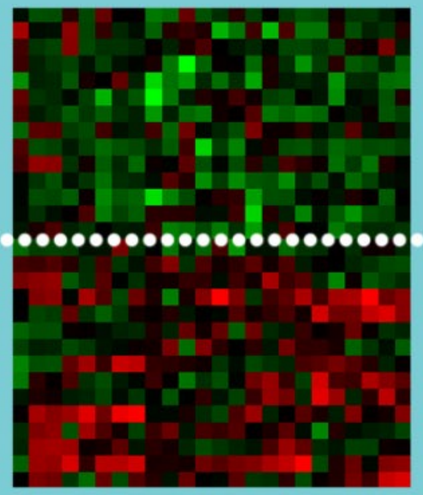
Patient 1



Patient 2

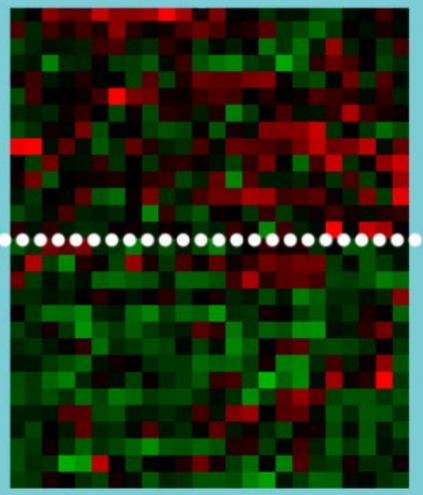
Colorectal Adenocarcinoma after genome-wide profiling

Molecular diagnosis sees TWO cancers

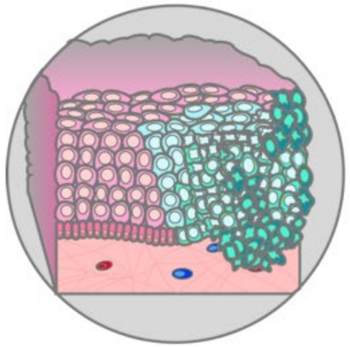


Region A

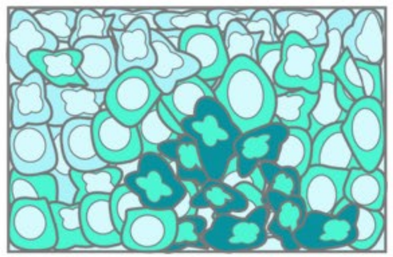
Region B



Can genome-wide molecular profiles **inform** cancer prognosis?

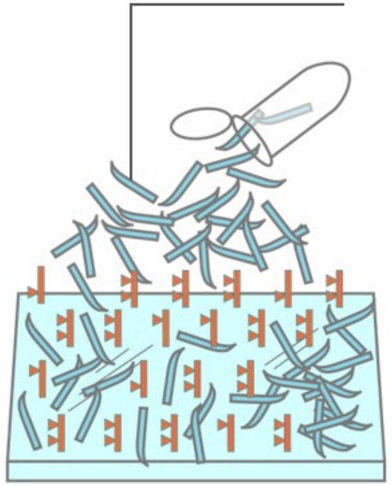


Patient's tissue sample



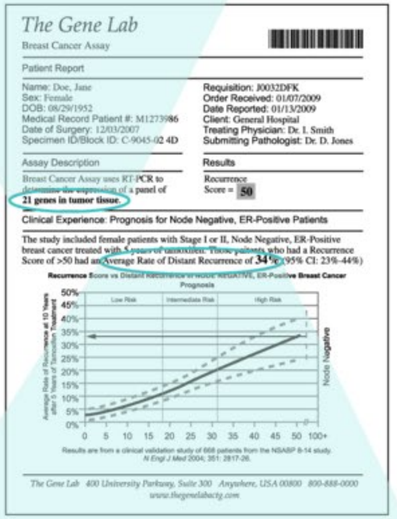
Paraffin block

Patient DNA



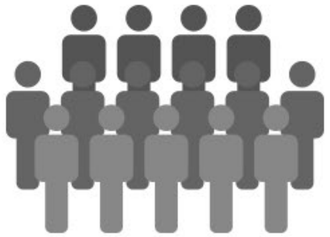
21-gene panel linked to breast cancer recurrence

21 genes in tumor tissue

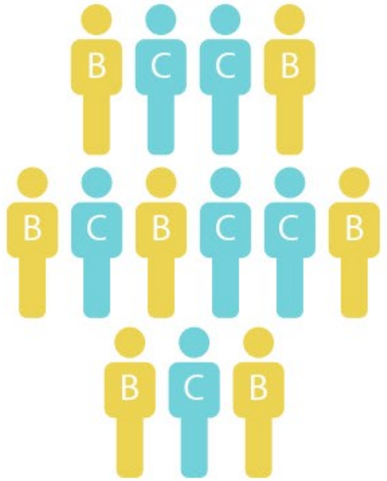


Average rate of distant recurrence is 34%

Colon cancer patients



Individual genome-wide profiles are sorted

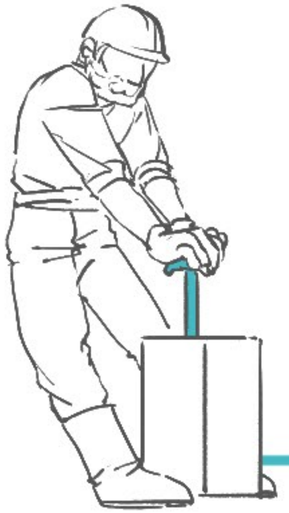


Have mutated K-ras gene

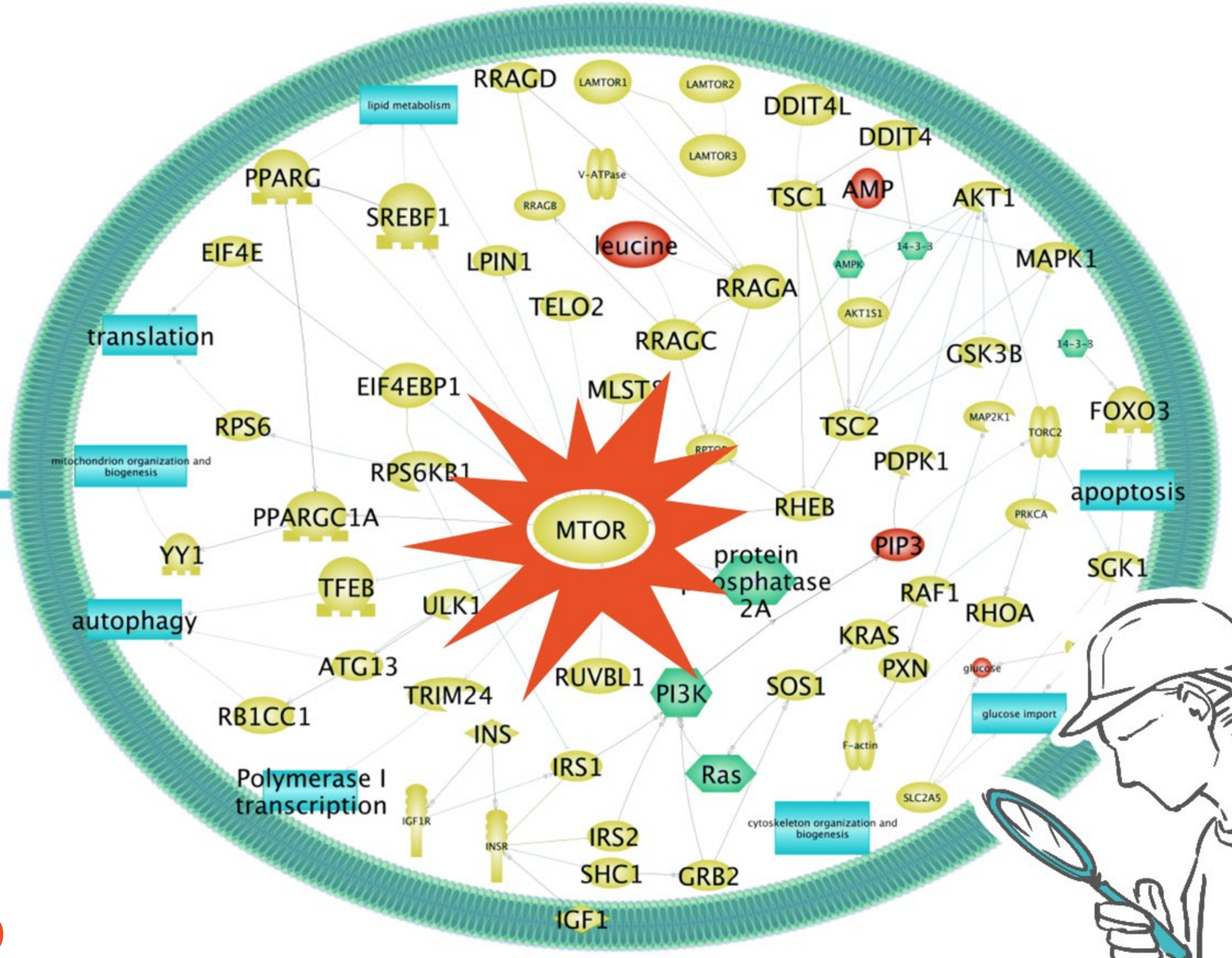
Do not have mutated K-ras gene

Respond to targeted therapy A

Can genome-wide molecular profiles **inform** cancer treatment planning?

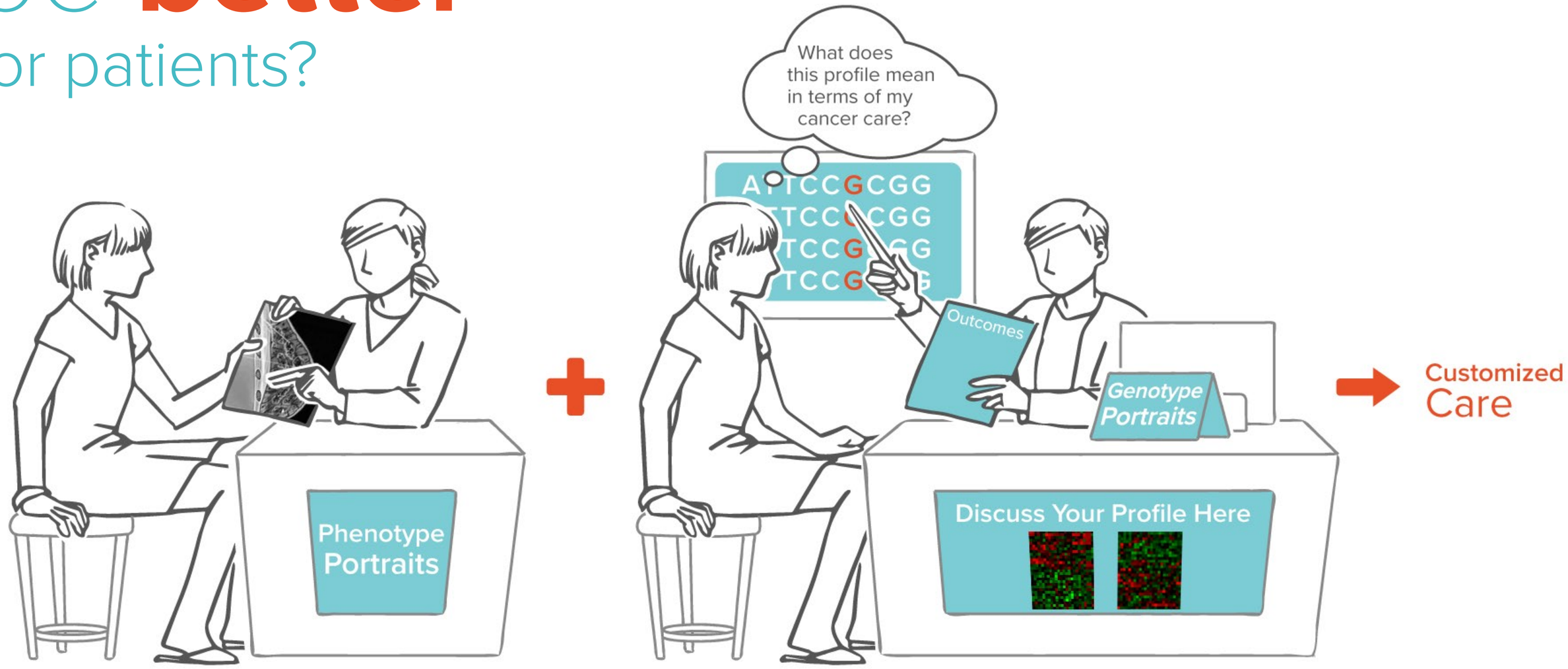


Can
genome-wide
molecular
profiles
monitor
the response to
cancer treatment?



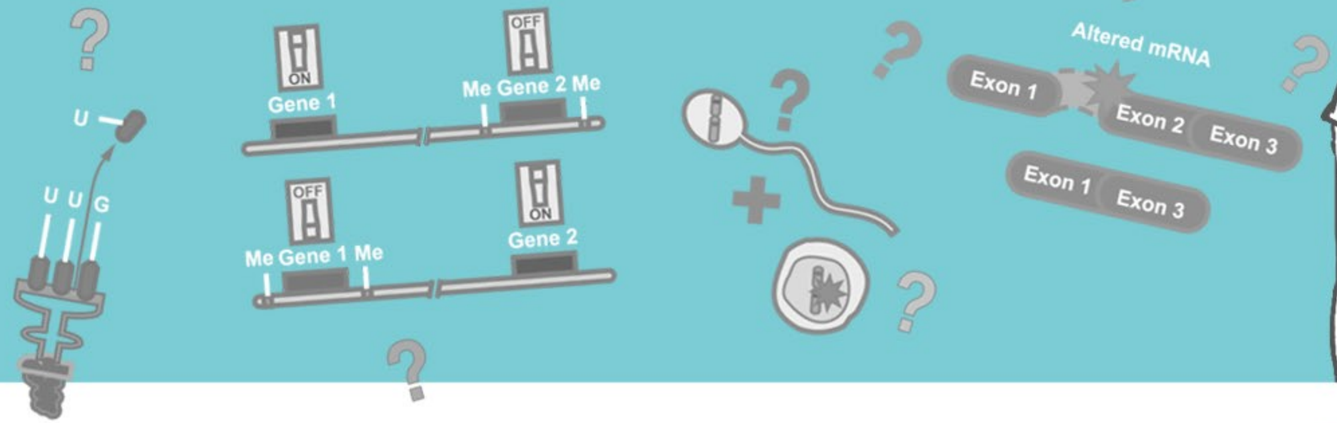
Goal: verify that pathway
is destroyed

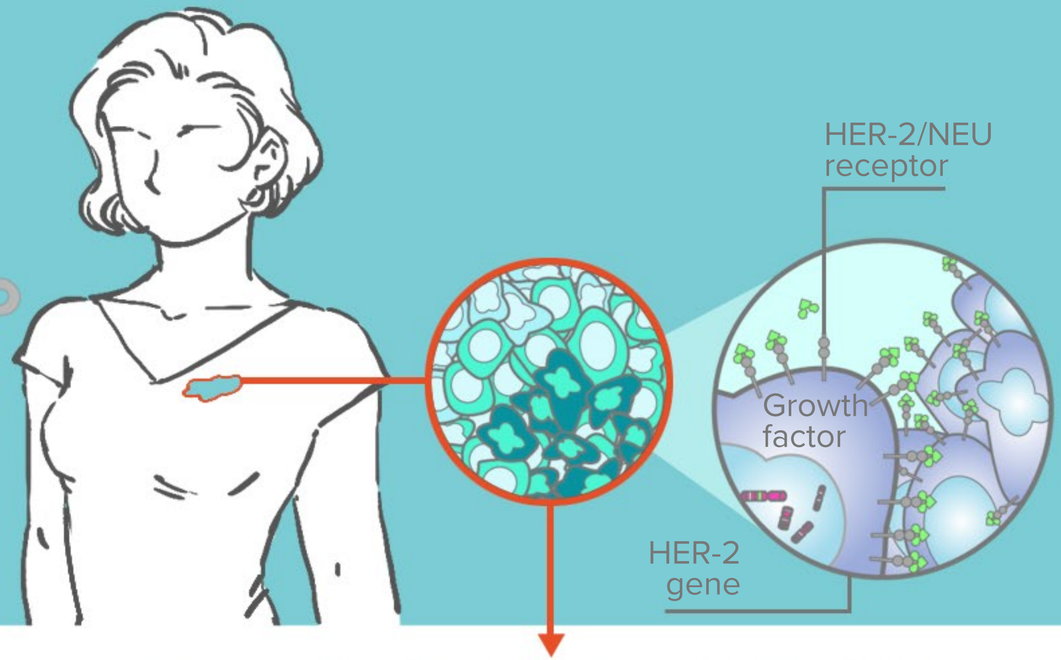
Will genomically informed cancer care be **better** for patients?



Personalized medicine:

A daunting challenge





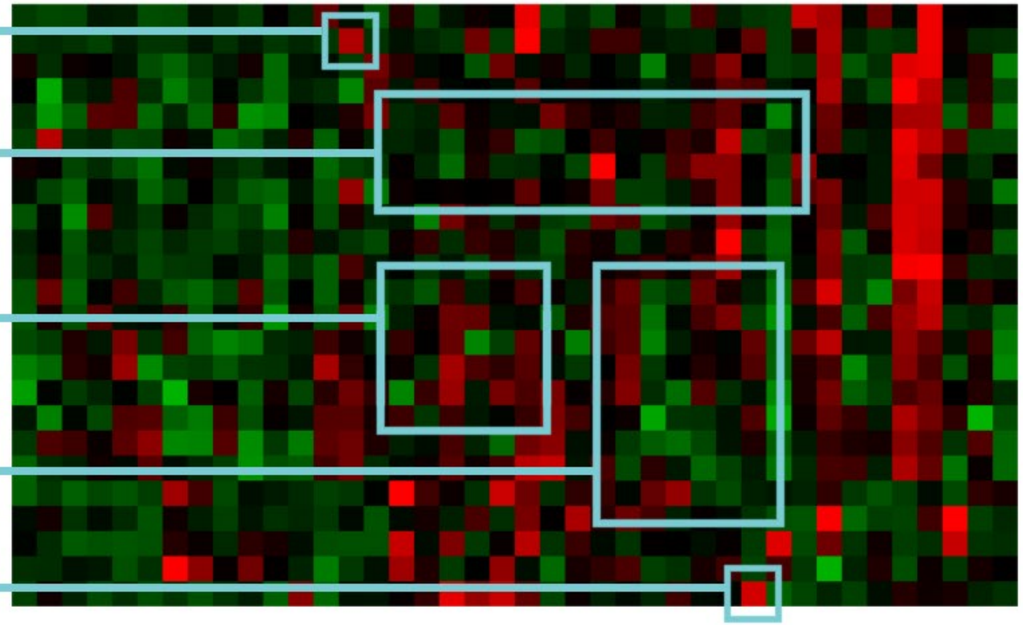
Growth receptor present

High risk of relapse

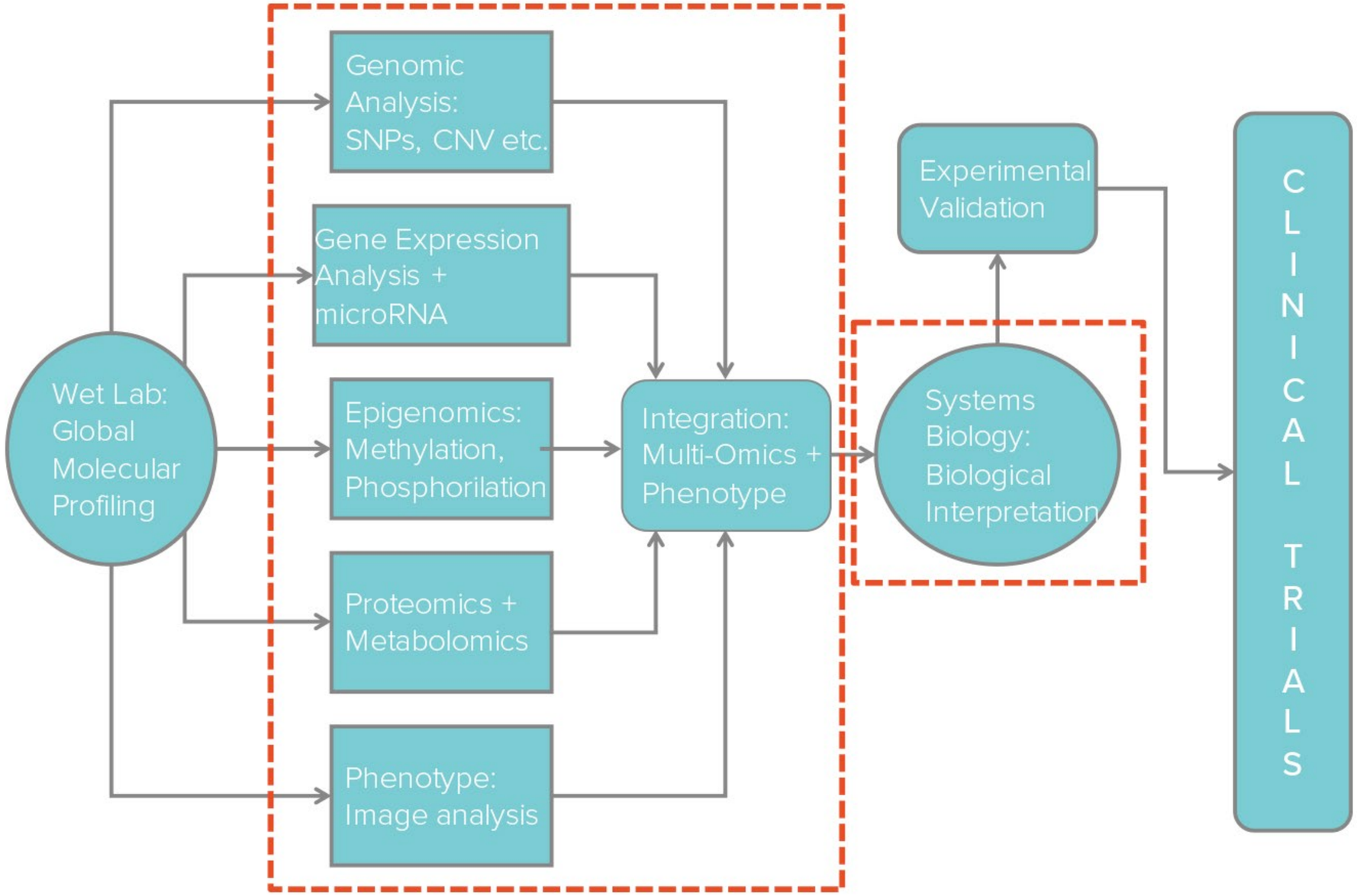
Not good candidate for
conventional therapy

Mutated susceptibility genes

Vascular markers present



Global Molecular Profiling of Diseases: Role of Translational Bioinformatics



Georgetown Lombardi launches

G-DOC®:

A unique cancer database

by Renee Twombly

Georgetown Lombardi Director Louis M. Weiner, MD, has long been frustrated. Despite the molecular revolution that has been taking place in medicine for decades, the way many physicians evaluate their patients dates back to the early part of the last century.

G-DOC – more than a database

Integrated Computational Environment Enabling Translational Research: Clinical Data + Molecular Data + Analysis Tools

Major Goals:

Finding molecular correlates of clinical outcome
Enabling population based analysis as well as
Individual patient-level comparisons of molecular profiles
Identification of most informative molecular “players”
i.e. candidate biomarkers
Mapping these biomarkers to specific pathways /networks
Exploring “dragability” of these biomarkers

G-DOC Plus Launched a year ago

The next-generation research platform for translational research and precision medicine

- More than 1500 users
- > 10000 samples: patient samples and cell line data

Data types in G-DOC Plus

G-DOC Plus Launched a year ago

Tools and workflows

Precision medicine

Explore medical images

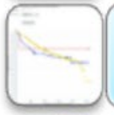
Explore somatic mutations

Translational research

Heat map viewer

Principal component analysis

KM plot using clinical data



KM plot using gene expression data

Group comparison

Plot single gene expression

Chromosomal instability index analysis

Population Genetics

Explore 1000 genomes data

Other tools

Explore clinical data and create groups

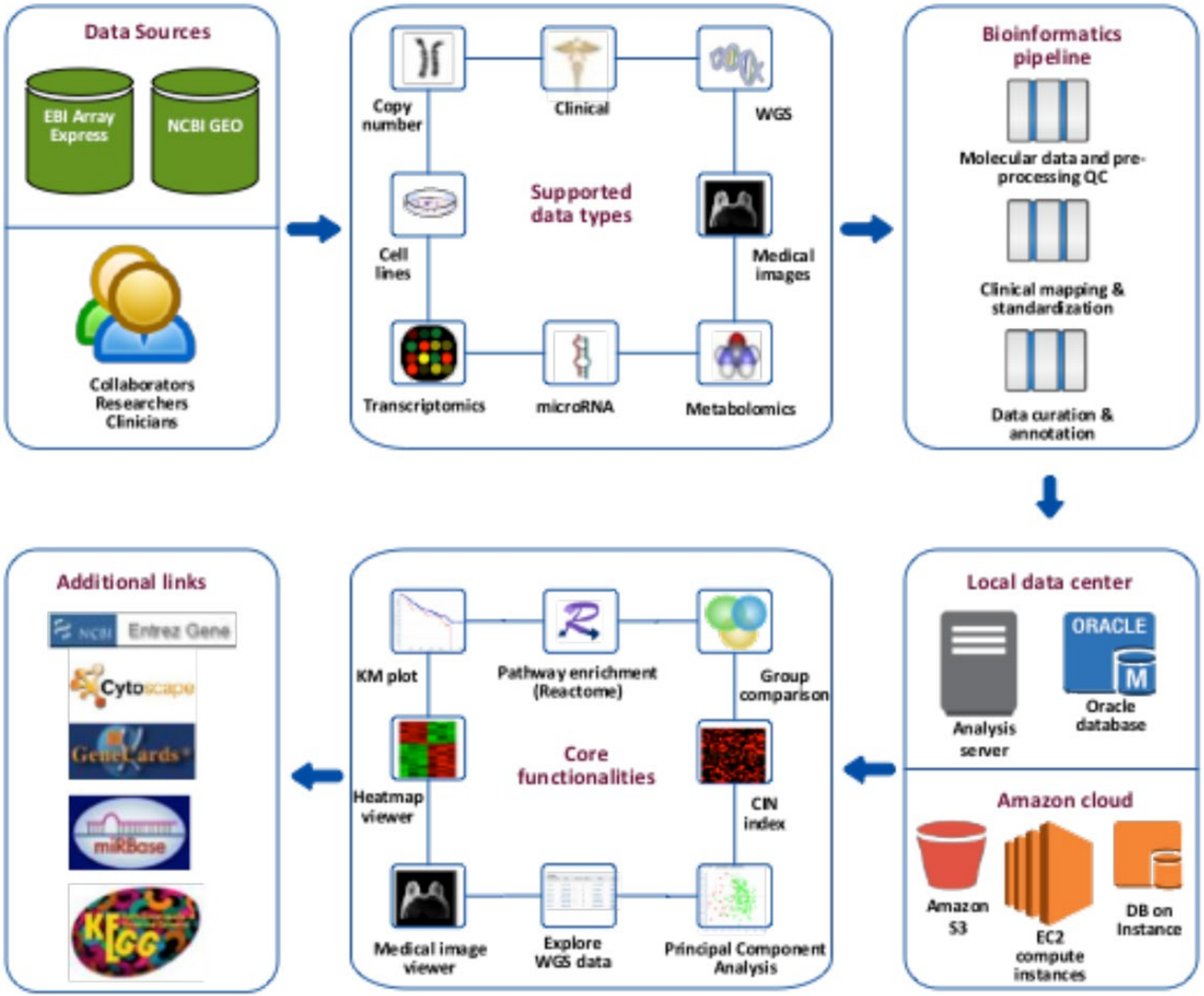
Pathway Enrichment Using Reactome

Create Venn diagrams

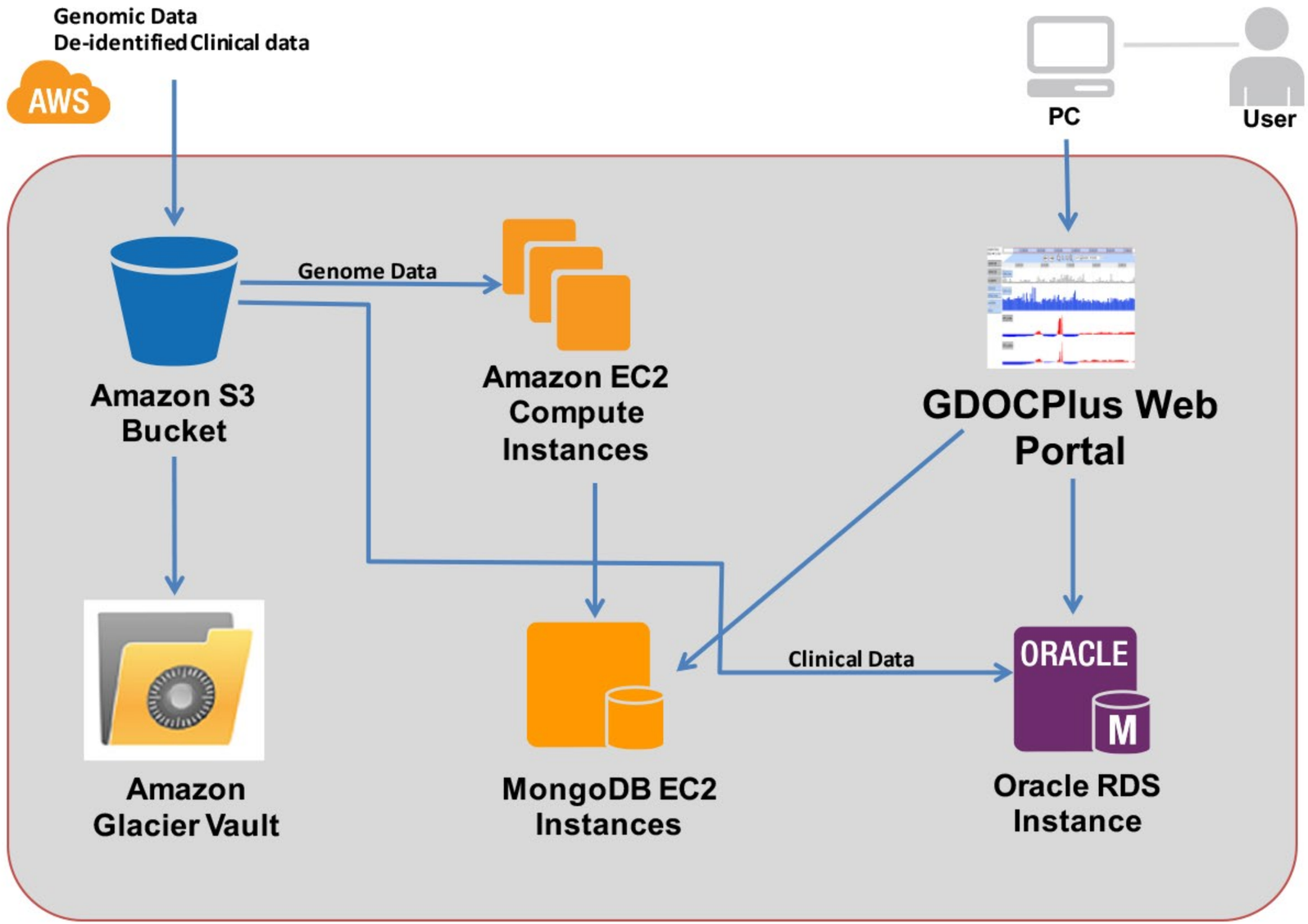
Molecular target viewer

Genome browser

G-DOC Data Processing and Analysis workflow



Next Generation Sequencing Big Data Analytics on the Cloud: powered by GDOC Plus



What’s next for GDOC?

New disease studies to be added to GDOC: Immunology

New Data Types: RNAseq

Additional Workflows: Precision Medicine

IMPORTANT: tutorials are available in public folder
https://gdoc.georgetown.edu/tutorials

Next: Live demo session with G-DOC

Tutorials and webinar recordings are available in public Box folder: https://gdoc.georgetown.edu/tutorials

1. Register at login page: gdoc.georgetown.edu
2. Log In with your username and password
3. Look around – become familiar with the G-DOC interface