The Cancer Genome Atlas (TCGA)

National Human Genome Research Institute

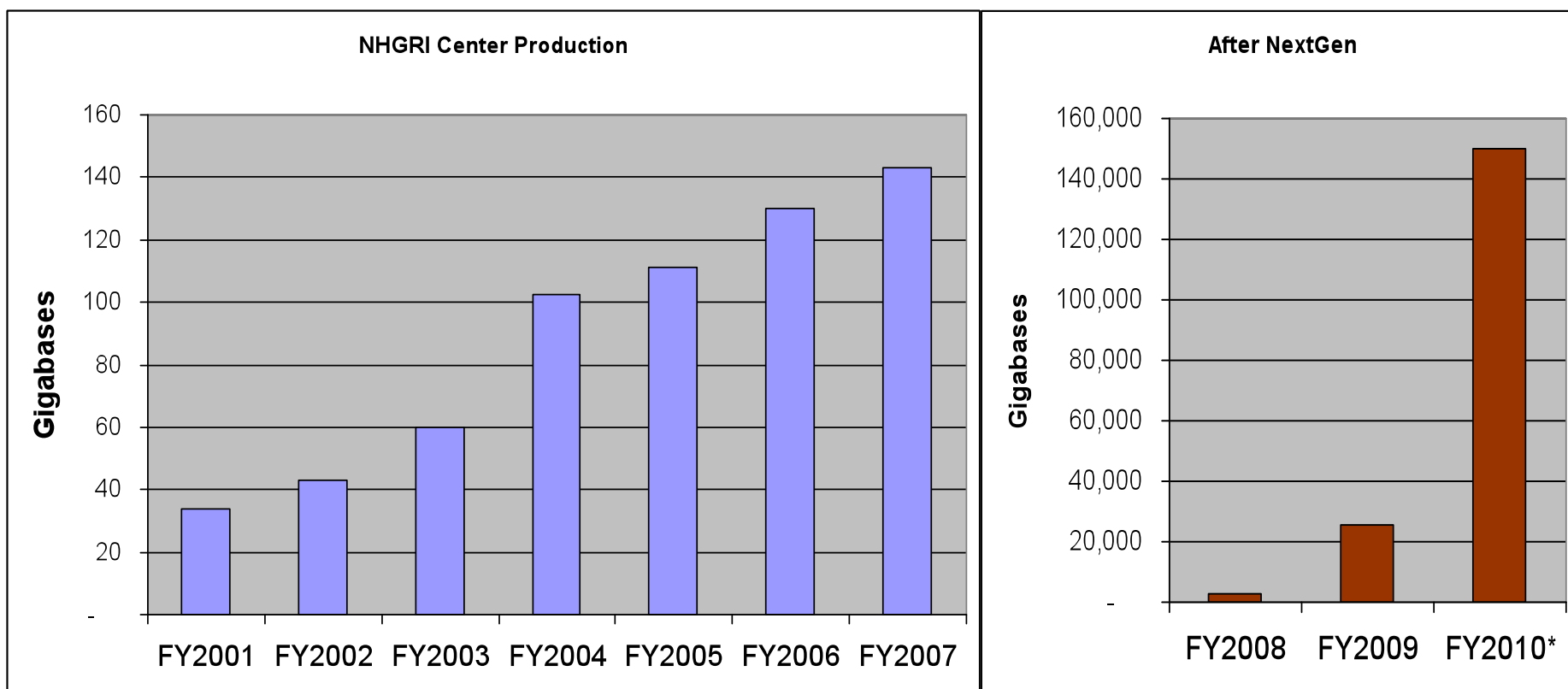## National Cancer Institute

Brad Ozenberger, PhD

Program Director, NHGRI

Cancer in the U.S.

- \>10,000,000 in the US have cancer (1 in 30 people)
- ~1,400,000 people will be diagnosed this year
- ~700,000 will die from cancer this year (a death every 45 seconds)

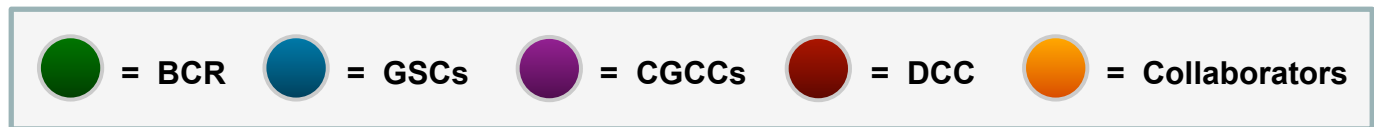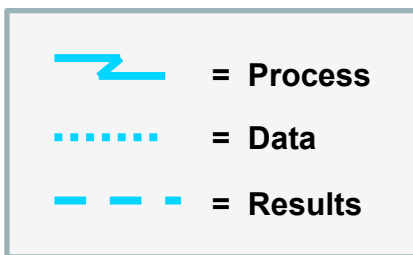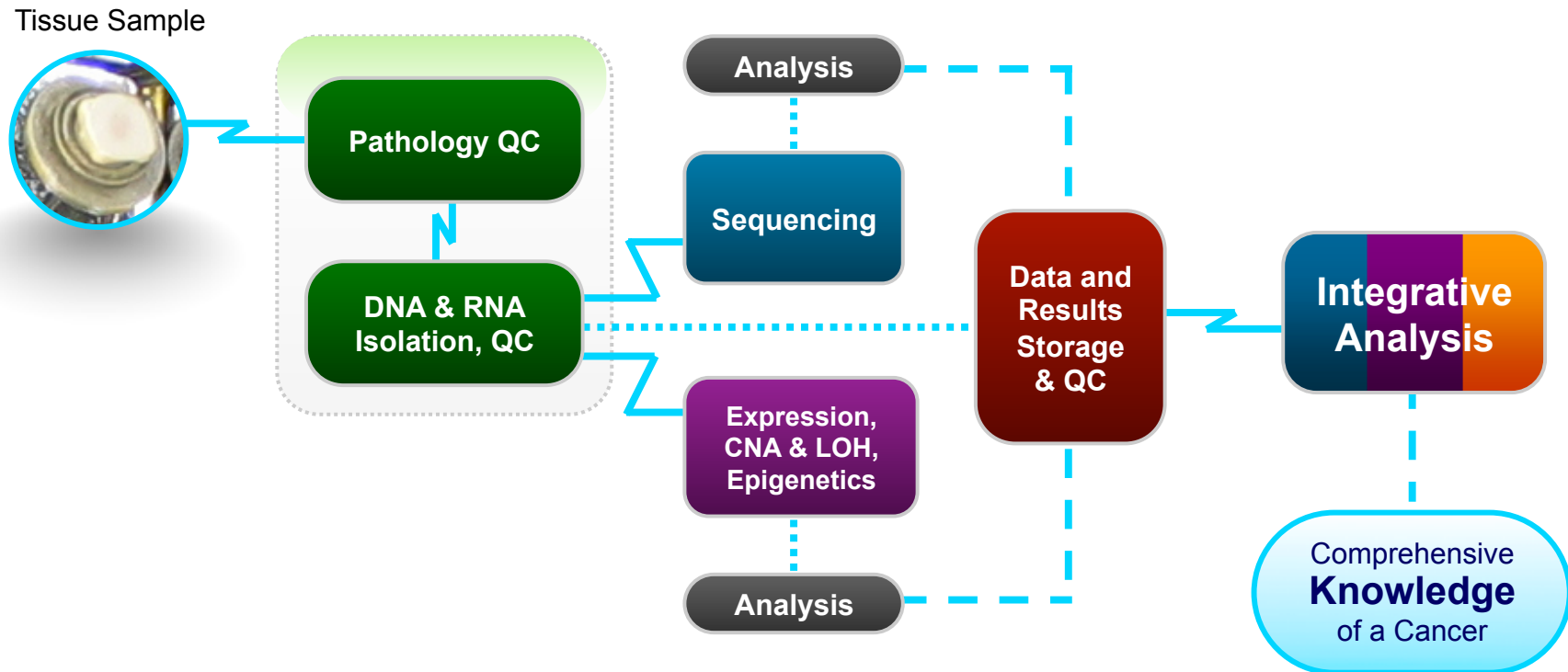# Scale of sequencing and data production

THE CANCER GENOME ATLAS

Comprehensive genomic analysis in large numbers of cancer specimens is now technically and economically feasible.

TCGA Project Pipeline

THE CANCER GENOME ATLAS

OVARIAN

Coverage(T/N)	Callable	Purity	Ploidy
31x / 30x	**81%**	**90%**	**2.8**

Name   **TCGA-13-0751**
Alias   **OV-0751**
Issued By   **Broad Institute**
Issue Date   **July 8, 2009**

Point Mutations

Rate/Mb	**0.75**
Total	**1786**
Coding	**9**

MIS   5
STOP 1
INDEL ---

HIGHLIGHTS

GENE	MUTATION	FUNCTION
TP53	Insertion	Tumor suppressor
EXOC6B	Missense	protein transport, exocytosis
ANKRD6	Missense	ankyrin
AHNAK	Missense	CNS development
C11orf52	Nonsense	?
GABRB3	Missense	GABA receptor

Lost BRCA1 germline indel

Chr. Aberrations

CNA Breaks	**---**
TX-Inter	**9**
TX-Intra	**15**

HIGHLIGHTS

**NF1-EFCAB5 fusion gene
probably inactivating
validated by RNA-seq**
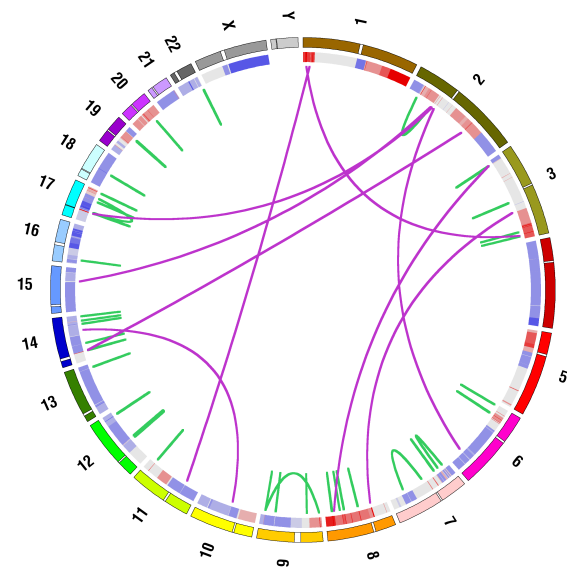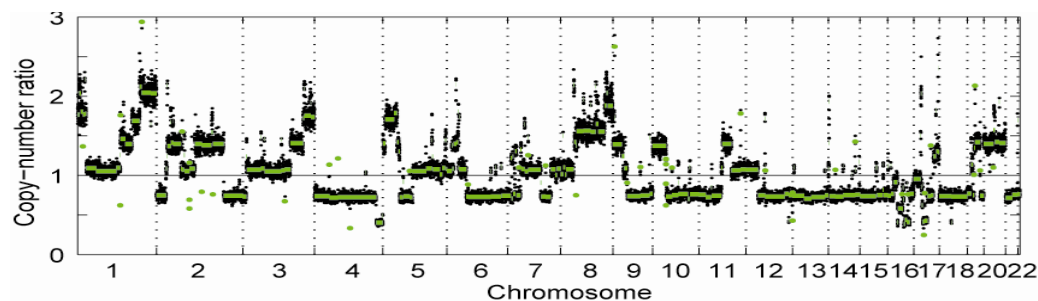
Courtesy of Gaddy Getz – Unpublished, Analysis in Process

TCGA Tumor Projects

THE CANCER GENOME ATLAS

Projects with comprehensive data available

➢ Glioblastoma (brain)
➢ Ovarian

Projects in progress (partial data sets available)

➢ Acute Myeloid Leukemia
➢ Colon Adenocarcinoma
➢ Rectal Carcinoma
➢ Lung Adenocarcinoma
➢ Lung Squamous Cell Carcinoma
➢ Renal Clear Cell Carcinoma

Projects anticipated to begin in 2010-2011

➢ Breast (multiple types)
➢ Bladder
➢ Cervical
➢ Head and neck
➢ Hepatacellular (liver)
➢ Lymphoma
➢ Melanoma
➢ Multiple myeloma
➢ Pancreatic
➢ Prostate
➢ Sarcoma
➢ Stomach
➢ Thyroid
➢ Uterine (endometrial)

International Cancer Genome Consortium Projects (June, 2010)

- USA
  - TCGA projects
- Canada
  - pancreatic*
  - prostate
- Australia
  - pancreatic*
  - ovarian
- China
  - stomach
- EU/France
  - renal carcinomas
- EU/United Kingdom
  - breast cancers*

- France
  - breast cancers*
  - hepatic (alcohol-associated)
- Germany
  - pediatric brain cancers
- India
  - oral
- Italy
  - rare pancreatic types
- Japan
  - hepatic (virus-associated)
- Spain
  - chronic lymphocytic leukemia
- United Kingdom
  - breast cancers*

*collaborative project

Cancer Research and Treatment in the Future

THE CANCER GENOME ATLAS

❑ TCGA can reveal all of the mutated genes that are functionally important for a specific tumor type (even if that gene is mutated in <5% of patients)

❑ TCGA will reveal new tumor biomarkers, new therapeutic targets

❑ Individualized approaches to therapy will be enabled; customized treatment based on the genetics of the tumor

❑ TCGA is breaking ground for new approaches to genomic and cancer research; methods to analyze and make sense of exceedingly massive and complex genomic data sets

❑ Cancer patients soon, and forever, will benefit