

The Genotype-Tissue Expression (GTEx) Project Update

**National Advisory Council
for Human Genome Research
February 11, 2013**

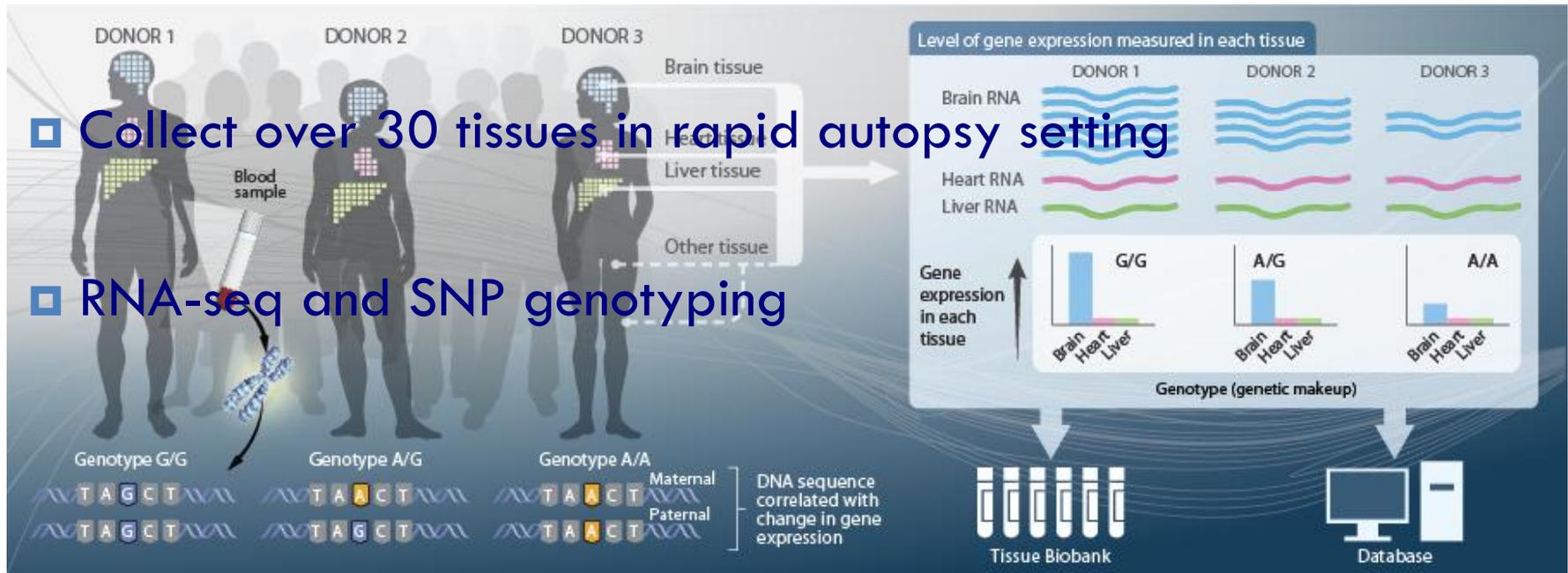
**Simona Volpi
Division of Genomic Medicine
NHGRI**

The Genotype-Tissue Expression (GTEx)

- NIH Common Fund (commonfund.nih.gov/gtex)
- Co-chairs – Eric Green, NHGRI & Tom Insel, NIMH
- Lead ICs (Program Officers)
 - ▣ NHGRI (Jeff Struewing, Simona Volpi)
 - ▣ NIMH (Su Koester, Roger Little)
 - ▣ NCI (Jim Vaught, Chana Rabiner)
- Active participation from many other ICs

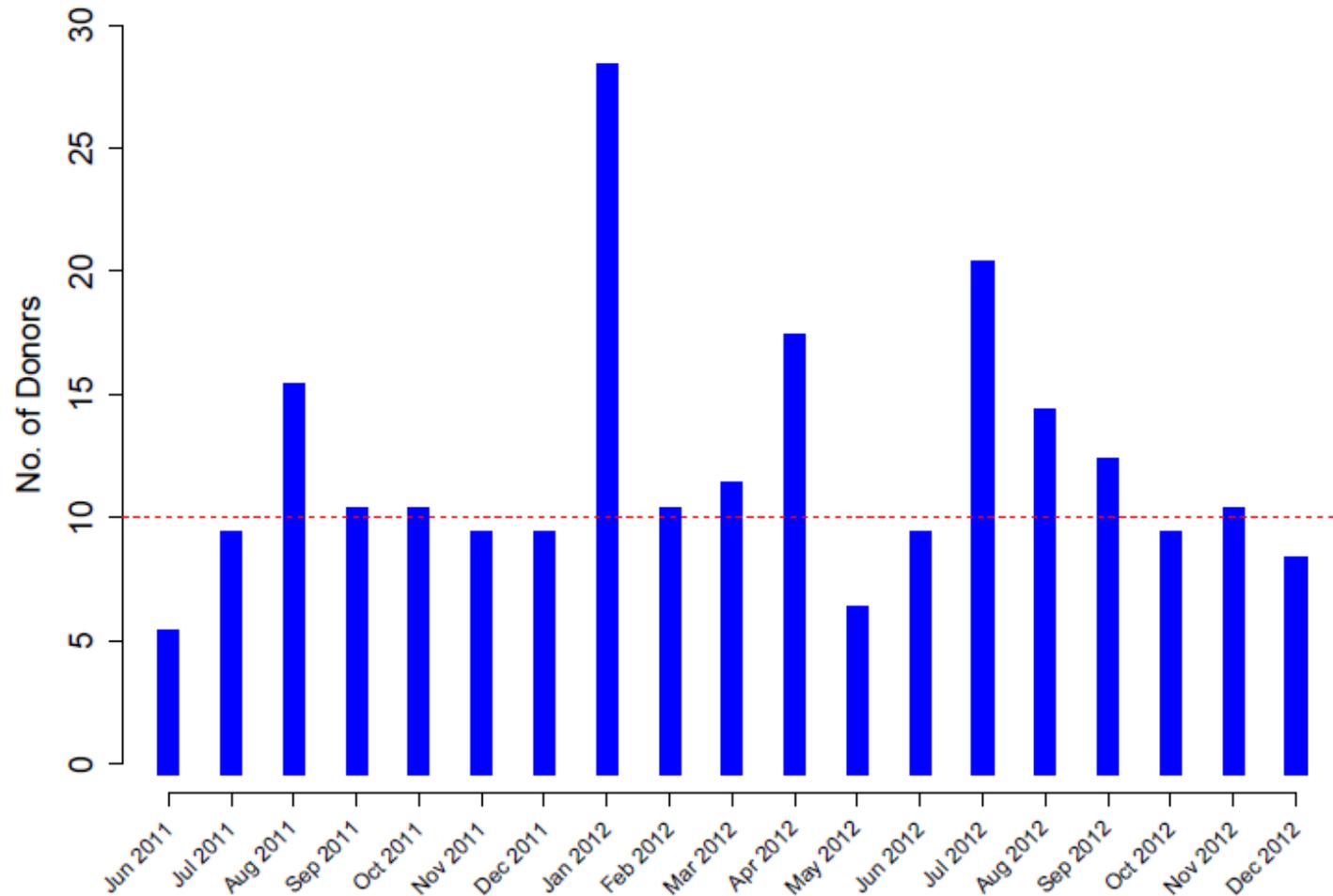
GTEx

- Goal – Establish a resource database and tissue bank in which to study the relationship between genetic variation and gene expression in reference/non-diseased human tissues.



GTEx Pilot Phase: 2010-2012

- Recruitment and RNA quality goals met



GTEx Scale up

- “Human Gene Expression Atlas”
- Comprehensive *cis*- and *trans*-eQTL results for each tissue
 - 900 post-mortem donors, completely genotyped
 - Over 25,000 tissues
 - Gene expression (RNA-Seq) for >20,000 tissue samples
 - Associated clinical and histopathological information
 - Access system for data and samples
 - ELSI study of donor families

GTEEx components

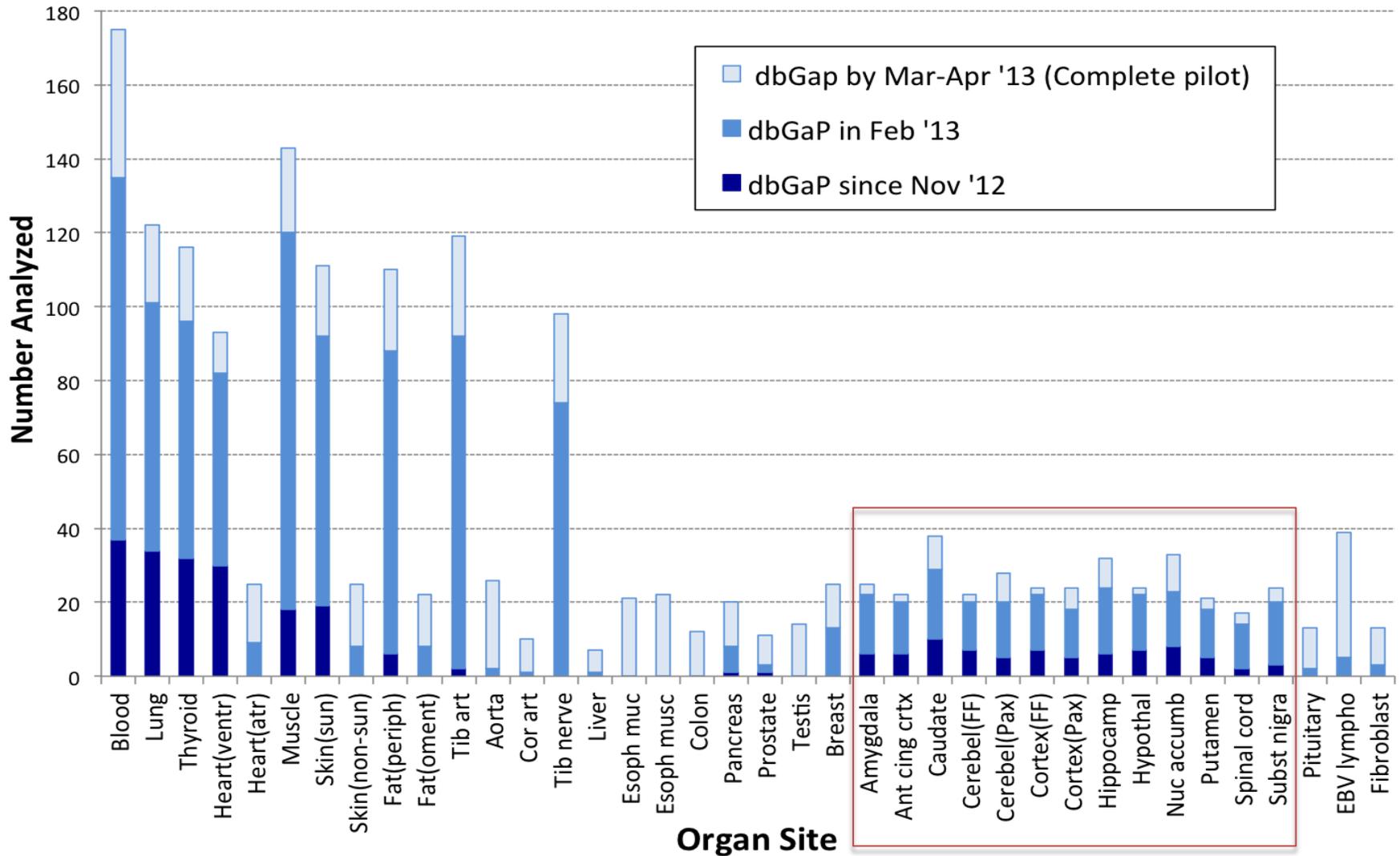
- Organ Procurement Organizations and Tissue Banks
- Sample and data repository
- Pathology review
- Brain Bank
- Laboratory, Data Analysis & Coordinating Center
- Statistical methods development grants
- ELSI study
- GTEEx database and browser
- Enhanced GTEEx (eGTEEx)

GTEx data

- Data
 - ▣ dbGaP + GTEx portal
 - ▣ SNP genotyping, RNA-seq, results etc.

The screenshot displays the GTEx Genotype-Tissue Expression Portal. At the top, the text "Search eQTLs" is visible. The main header features the "GTEx" logo and the URL "http://www.broadinstitute.org/gtex/". Below the header is a navigation bar with links for "Home", "Search", "Datasets", "Samples", and "Documentation". The "Search" dropdown menu is open, showing a "Search eQTLs" option. Below the navigation bar, there are two search forms. The first form is titled "Search eQTLs" and includes a "Find eQTLs for a Gene" button. The second form is titled "Lookup Significant eQTLs for a SNP" and includes a text input field for "SNP Id...", a dropdown menu for "Adipose", and a "Find eQTLs for a SNP" button.

Expression data production and release



GTEx samples

- Available Biospecimens
 - ▣ PAXgene fixed, frozen tissue; PAXgene Fixed, Paraffin Embedded Tissue; RNA; DNA
 - ▣ Flash frozen brain
 - ▣ Lymphoblastoid and fibroblast cell lines

Sample Search

Please Note: The list of samples returned by this search is inclusive of all samples collected for GTEx. Many of these samples, however, have not undergone primary molecular analyses for use in the GTEx resource. As a result, sample availability and amount are subject to change without notice.

Material Type

Volume Greater Than μL

Tissue Site

Concentration Greater Than $\text{ng}/\mu\text{L}$

Gender

View Columns

- Material Type
- Original Material Type
- Tissue Site
- Gender
- Autolysis Score

- RIN Number
- GTEx Public Sample ID
- GTEx Public Participant ID
- Genotype

- Volume μL
- Concentration $\text{ng}/\mu\text{L}$
- Mass
- Expression

RFA-RM-12-009: Enhancing GTEx with molecular analyses of stored biospecimens (U01)

- Receipt date: March 28, 2013
- Summer review and September NHGRI council
- ~ \$10,000,000 (total costs) over three years, with \$3,800,000 in FY2014
- Open to the most compelling assays and ideas

GTEx Acknowledgments

Cancer Human Biobank (caHUB)

Biospecimen Source Sites (BSS)

- National Disease Research Interchange
 - Gift of Life Donor Program
 - LifeNet Health
 - Albert Einstein Medical Center
 - Virginia Commonwealth University (ELSI)
- Roswell Park Cancer Institute
 - UNYTS

Comprehensive Biospecimen Res (CBR)

- Van Andel Institute

Comprehensive Data Res (CDR)

- SAIC-F

Pathology Resource Center (PRC)

- NCI/SAIC-F

Brain Bank

- University of Miami

Laboratory, Data Analysis, and Coordinating Center (LDACC)

- The Broad Institute

Statistical Methods Development R01s

- Harvard University
- University of Chicago (2)
- University of Geneva
- University of North Carolina - Chapel Hill

GTEx Database and Browser

- NCBI

NIH WG Members

commonfund.nih.gov/GTEx/members.aspx

Questions ?