



*Telling Alabama's*  
**GENOMIC STORY**

ALABAMA GENOMIC HEALTH INITIATIVE



*Alabama Genomic*  
**HEALTH INITIATIVE**



UAB Medicine/HudsonAlpha  
Institute for Biotechnology



\$2 million allocation from  
state FY17



Genomic analysis of 10,000  
citizens over five years



Return of clinical data



Research database and  
biobank

### Oversight Committee



**Mitchell B. Cohen, MD**  
Chair of UAB Department  
of Pediatrics



**Edward Partridge, MD**  
Director of UAB  
Comprehensive Cancer Center

*\*Upon Dr. Partridge's retirement in  
July 2017, Dr. Cohen stepped into this  
role on the oversight committee.*



**Rick Myers, PhD**  
President and Science Director  
at HudsonAlpha Institute  
for Biotechnology

### Additional leaders in the UAB School of Medicine on the Oversight Committee



**Etty Benveniste, PhD Senior**  
Associate Dean for  
Research Administration  
and Development



**Robert Kimberly, MD**  
Senior Associate Dean  
for Clinical and  
Translational Research



**Toni Leeth, MPH**  
Associate Dean for Strategic  
Planning and Administration

### Principal Investigators



**Bruce R. Korf, MD, PhD**  
Chief Genomics Officer  
of UAB Medicine



**Greg Barsh, MD, PhD**  
Faculty Investigator and  
Chair of HudsonAlpha Institute  
for Biotechnology



**Matthew Might, PhD**  
Director of UAB Hugh Kaul  
Precision Medicine Institute

# Population Cohort

Genotyping array  
Variant analysis  
Return of results of actionable variants  
Genetic counseling  
Supportive care

# Affected Cohort

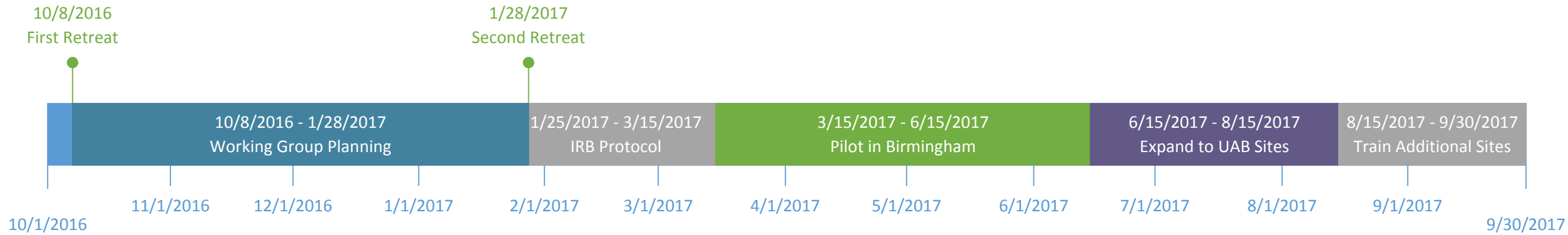
Whole genome sequencing  
Variant analysis  
Return of results of pathogenic variants  
Genetic counseling  
Supportive care

DNA/Tissue Bank  
Genomic Database  
Medical Records (i2b2)

# ACMG Secondary Findings

Type	Genes
<b>Tumor Predisposition</b> Breast/ovarian, Li-Fraumeni, Peutz-Jeghers, Lynch, Polyposis, Von Hippel-Lindau, MEN1/2, Medullary thyroid cancer, PTEN hamartoma syndrome, Retinoblastoma, Paraganglioma/pheochromocytoma, Tuberous sclerosis complex, WT1-related Wilms' tumor, NF2	BRCA1/2, TP53, STK11, MLH1, MSH2, MSH6, PMS2, APC, MUTYH, BMPR1A, SMAD4, VHL, MEN1 RET, PTEN, RB1, SDHD, SDHAF2, SDHC, SDHB, TSC1, TSC2, WT1, NF2
<b>Connective Tissue Dysplasia</b> Ehlers-Danlos vascular type, Marfan, Loeys-Dietz, Familial aortic aneurysms and dissections	COL3A1, FBN1, TGFB1, TGFB2, SMAD3, ACTA2, MYH11
<b>Cardiac</b> Hypertrophic cardiomyopathy, dilated cardiomyopathy, Arrhythmia	MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA, MYL2, LMNA, RYR2, PKP2, DSP, DSC2, TMEM43, DSG2, KCNQ1, KCNH2, SCN5A
<b>Metabolic</b> Hypercholesterolemia, Wilson disease, Ornithine transcarbamylase deficiency	LDLR, APOB, PCSK9, ATP7B, OTC
<b>Pharmacogenetic</b> Malignant Hyperthermia	RYR1, CACNA1S

# Timeline FY17



The public launch of the AGHI represented a collaborative effort among UAB Health System Marketing Communications, UAB University Relations, UAB Department of Genetics, UAB School of Medicine Communications, AGHI Leadership, and HudsonAlpha Communications and Marketing. Through combined tactical communications efforts, the media relations potential reach was over 3.5 million.



**10/08**  
1st AGHI planning retreat\*

**03/01**  
@uabprecisionmedicine Twitter account goes live

**03/09**  
AGHI Press Conference

**05/02**  
UAB Institutional Review Board grants approval for AGHI Recruitment and Engagement Protocol

**05/18**  
Pilot recruitment phase begins at The Kirklin Clinic of UAB Hospital in Birmingham

**06/15**  
First whole genome sequence participant enrolled at North Alabama Children's Specialists, part of the Children's of Alabama network

**07/11**  
AGHI presented at UAB Cancer Center Community Dissemination Institute

**07/25**  
First whole genome sequence participants enrolled at Children's of Alabama

**09/01**  
First participant results received. Genetic counselors begin to return results to participants and providers\*\*\*

**09/14**  
First whole genome sequence participants enrolled at The Kirklin Clinic

UAB awarded the grant from the state of Alabama



OCTOBER 2016

JANUARY 2017

MARCH 2017

MAY 2017

JUNE 2017

JULY 2017

AUGUST 2017

SEPTEMBER 2017

OCTOBER 2017

**01/28**  
2nd AGHI planning retreat\*

**03/08**  
www.aghi.org website launched in preparation for press conference

**05/03**  
AGHI Town Hall led by Sara Knight, PhD, at UAB Health Disparities Research Symposium, Birmingham

**06/01**  
100 participants enrolled\*\*

**06/22**  
Enrollment opens at Medical Towers

**07/28**  
250 participants enrolled

**08/14**  
500 participants enrolled

**09/04**  
750 participants enrolled

**09/29**  
1,000 participants enrolled

**03/09**  
UAB Institutional Review Board grants approval for AGHI Administrative Protocol

**05/08**  
AGHI presented at UAB-HudsonAlpha Center for Genomic Medicine Symposium in Huntsville

**07/11**  
Open/Public enrollment begins

\*Planning retreats were essential steps toward executing a comprehensive strategy for the AGHI and to clarify procedures moving forward.

\*\*At this point, AGHI recruitment implemented a quick pause to evaluate efforts and materials, and to make improvements prior to launching a full-scale public recruitment effort.

\*\*\*Providers are notified only if participants consented to have results shared with their chosen health care provider.



## BY THE NUMBERS

### CORE VALUES

TRANSPARENCY/HONESTY • RESPECT  
CONFIDENTIALITY • FAIRNESS

 **59**   
ACTIONABLE GENES

POWER OF MORE  
THAN 75 PHYSICIANS,  
SCIENTISTS, HEALTH  
CARE ADMINISTRATORS,  
& PATIENT NAVIGATORS  
900+ BIOBANK  
SPECIMENS FOR  
FUTURE RESEARCH

**1,013**

TOTAL PARTICIPANTS ENROLLED AS OF SEPTEMBER 29, 2017

Data includes first five months of enrollment of AGHI.

**\$2**  
MILLION  
FROM THE STATE OF  
ALABAMA

 **45**  
ALABAMA  
COUNTIES

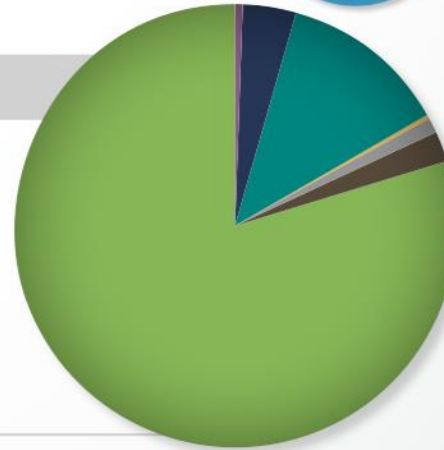
PARTICIPANTS FROM  
45 OF 67 ALABAMA  
COUNTIES

## AGHI Recruitment By the Numbers (as of September 29, 2017)



### AGHI ENROLLMENT BY RACE

American Indian or Alaska Native	0.8%
Asian	4.0%
Black/African American	13.1%
Native Hawaiian or Other Pacific Native	0.1%
More than one race	1.3%
Unknown	2.3%
White	78.4%



**\* 5.6% OF PARTICIPANTS ARE HISPANIC/LATINO**

## AGHI: CURRENT & FUTURE SITES



### Current Sites



The Kirklind Clinic of UAB  
Hospital, Birmingham

UAB Medical Towers,  
Birmingham



Children's of Alabama,  
Birmingham

North Alabama Children's  
Specialists, Huntsville

### Planned Future Sites for FY18

Huntsville

Montgomery

Selma

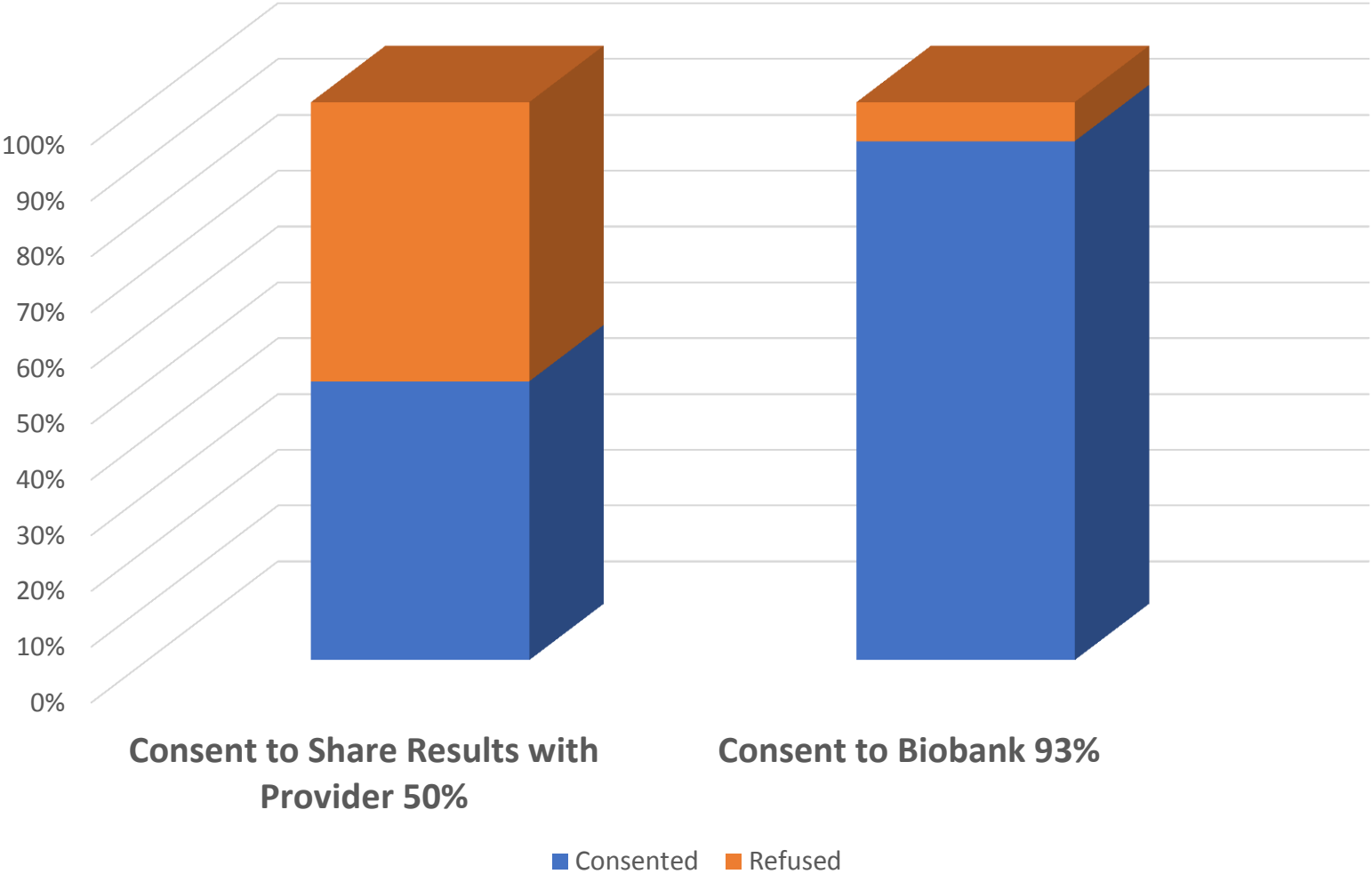
Tuscaloosa

Mobile

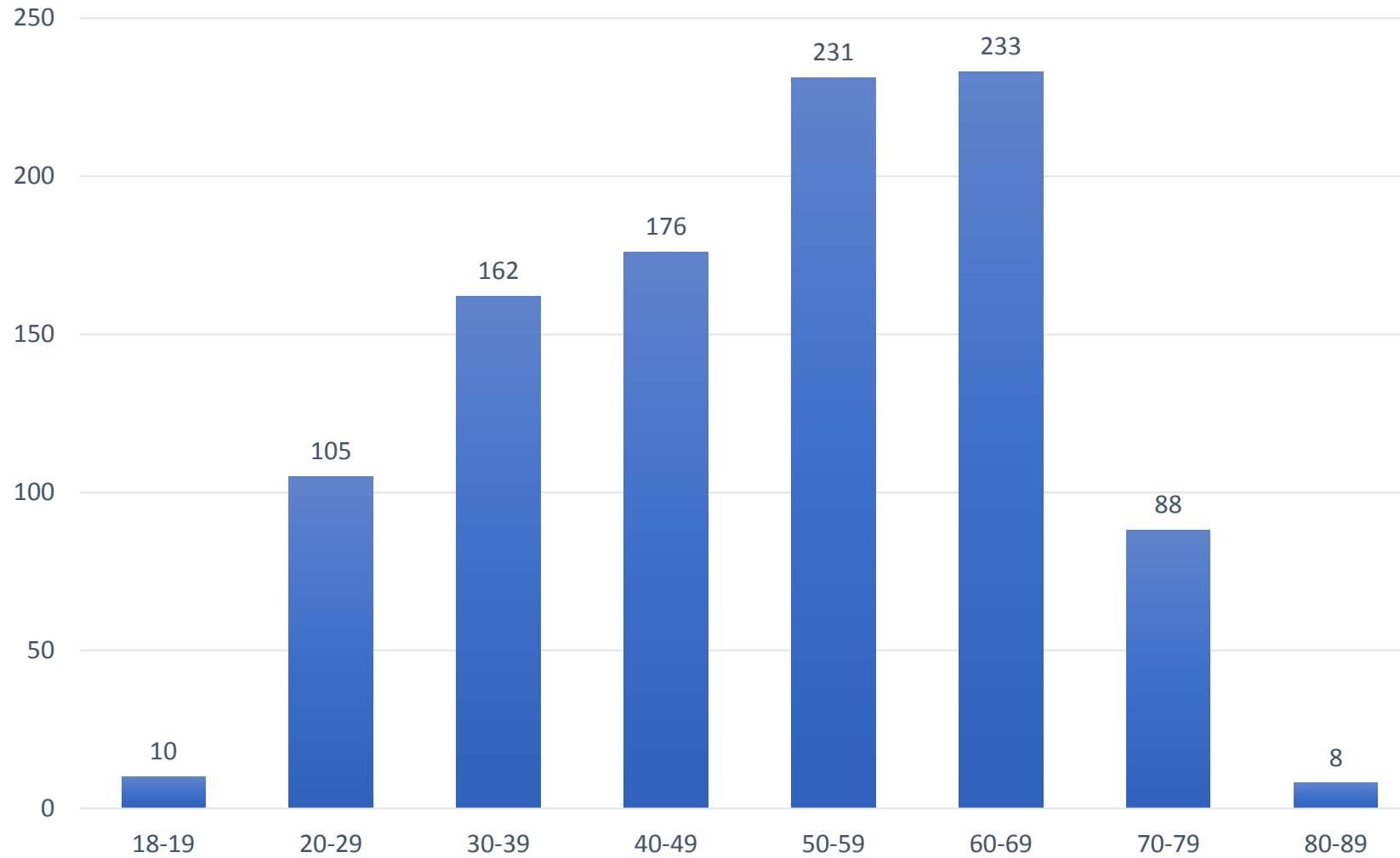


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AGHI Participant Consent FY 17



# FY17 AGHI Participants by Age



■ For first 1013 participants enrolled in FY17

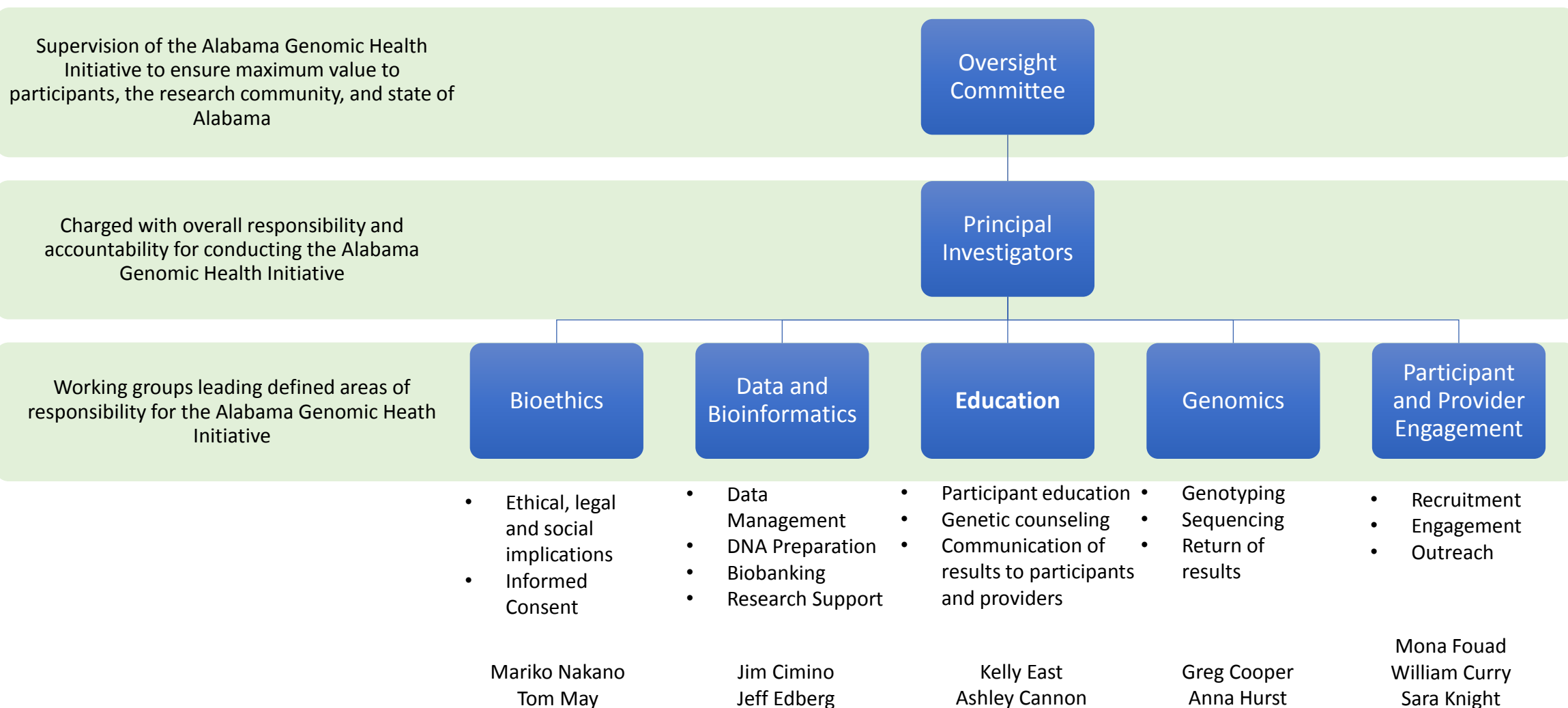


## On the Horizon for FY18

- Expand enrollment to include Selma, Montgomery, Huntsville, Tuscaloosa
- Enroll additional participants to mirror state population of Alabama
- Utilize the UAB MD Learning Channel and [www.aghi.org](http://www.aghi.org) to distribute a series of videos and podcasts focusing on genomic medicine
- Continue community engagement research to gain feedback from diverse voices across the state
- Continue to develop biobank and AGHI data to support precision medicine research for years to come



# Alabama Genomic HEALTH INITIATIVE



# Some Lessons and Issues

1

## Adding Value

- Ancestry
- Carrier Status
- Pharmacogenetics

2

## Concerns

- Diversity
- Clinical vs. Research
- Numbers

3

## Impact

- Awareness
- Education
- National Model



# *Alabama Genomic* HEALTH INITIATIVE

## *Contact Information*

[www.aghi.org](http://www.aghi.org)



@uabprecisionmed

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