

Disclosure Slide

Financial Disclosure for:

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We have nothing to disclose

Section 1

Imputation and the Server



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MICHIGAN
IMPUTATION SERVER

Learning objectives

Participants will

1. Understand the principles of genotype imputation and the Michigan Imputation Server

Genotype imputation

Key method used in GWAS to

- Increase the number of tested variants
- Fine-mapping becomes more complete
- Meta-analysis using different arrays

0. Imputation setting

GWAS Haplotypes

```
... . . . A . . . . . . . A . . . . . A . . . .  
... . . . G . . . . . . . C . . . . . A . . . .
```

Reference Haplotypes (e.g. 1000G)

```
C G A G A T C T C C T T C T T C T G T G C  
C G A G A T C T C C C G A C C T C A T G G  
C C A A G C T C T T T C T T C T G T G C  
C G A A G C T C T T T C T T C T G T G C  
C G A G A C T C T C C G A C C T T A T G C  
T G G G A T C T C C C G A C C T C A T G G  
C G A G A T C T C C C G A C C T T G T G C  
C G A G A C T C T T T C T T T G T A C  
C G A G A C T C T C C G A C C T C G T G C  
C G A A G C T C T T T C T T C T G T G C
```

1. Identify match among reference

GWAS Haplotypes

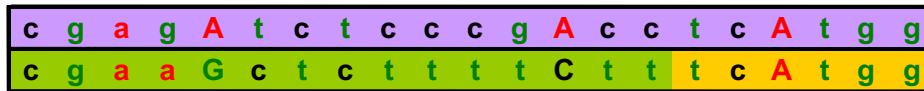
. . . . A A A
. . . . G C A

Reference Haplotypes (e.g. 1000G)

C	G	A	G	A	T	C	T	C	C	T	T	C	T	T	T	C	T	G	T	G	C
C	G	A	G	A	T	C	T	C	C	C	G	A	C	C	T	C	A	T	G	G	
C	C	A	A	G	C	T	C	T	T	T	T	C	T	T	T	C	T	G	T	G	C
C	G	A	A	G	C	T	C	T	T	T	T	C	T	T	T	C	T	G	T	G	C
C	G	A	G	A	C	T	C	T	C	C	G	A	C	C	T	T	A	T	G	C	
T	G	G	G	A	T	C	T	C	C	C	G	A	C	C	T	C	A	T	G	G	
C	G	A	G	A	T	C	T	C	C	C	G	A	C	C	T	T	G	T	G	C	
C	G	A	G	A	C	T	C	T	T	T	T	C	T	T	T	T	G	T	A	C	
C	G	A	G	A	C	T	C	T	C	C	G	A	C	C	T	C	G	T	G	C	
C	G	A	A	G	C	T	C	T	T	T	T	C	T	T	T	C	T	G	T	G	C

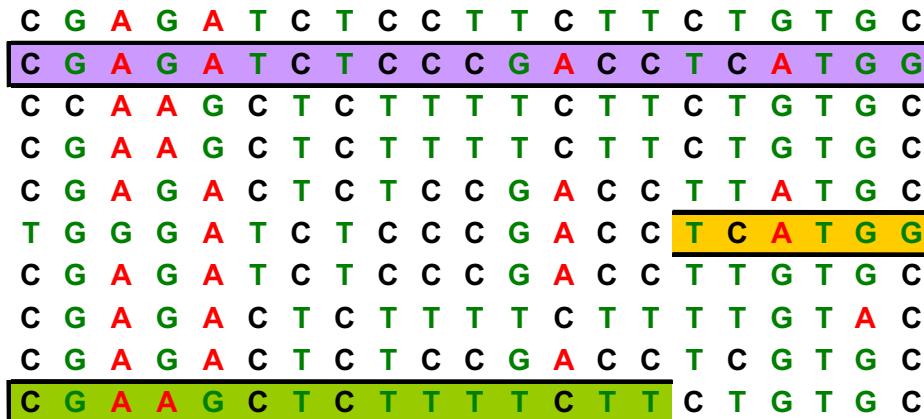
2. Impute

GWAS Haplotypes



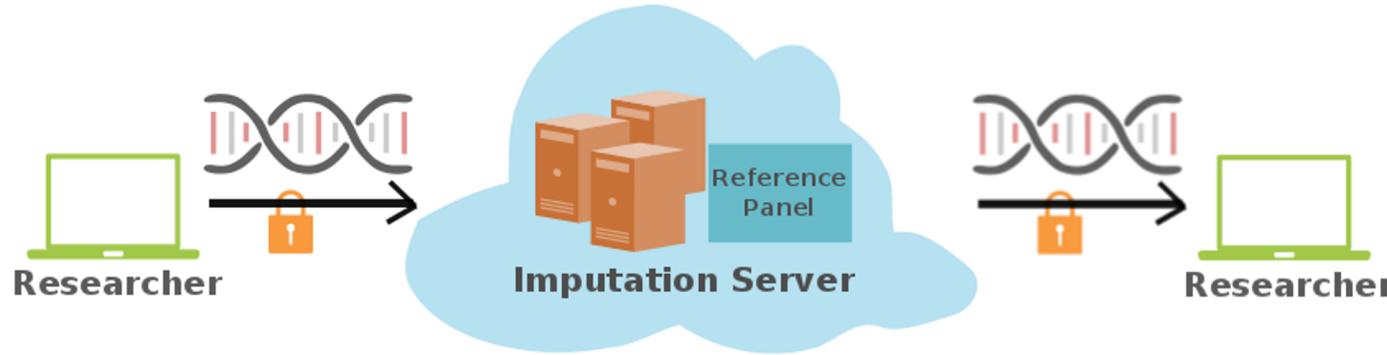
c g a g A t c t c c c g A c c t c A t g g
c g a a G c t c t t t C t t t c A t g g

Reference Haplotypes (e.g. 1000G)



C G A G A T C T C C T T C T T C T G T G C
C G A G A T C T C C C G A C C T C A T G G
C C A A G C T C T T T T C T T C T G T G C
C G A A G C T C T T T T C T T C T G T G C
C G A G A C T C T C C C G A C C T T A T G C
T G G G A T C T C C C G A C C T C A T G G
C G A G A T C T C C C G A C C T T G T G C
C G A G A C T C T T T T C T T T T G T A C
C G A G A C T C T C C C G A C C T C G T G C
C G A A G C T C T T T C T T C T G T G C

ASHG 2014: imputation web service



1.

Upload GWAS data

2.

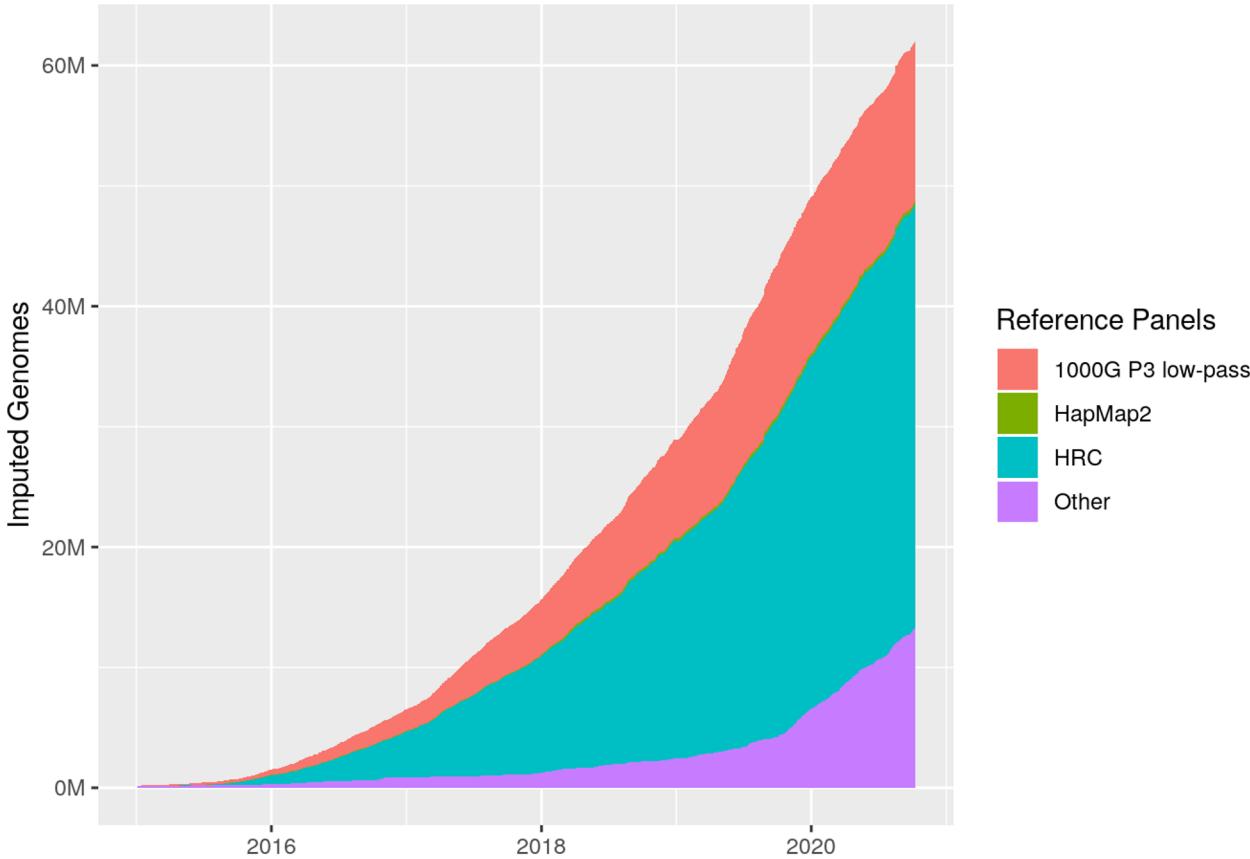
Server performs

- Quality checks
- Pre-phasing
- Imputation
- Encryption

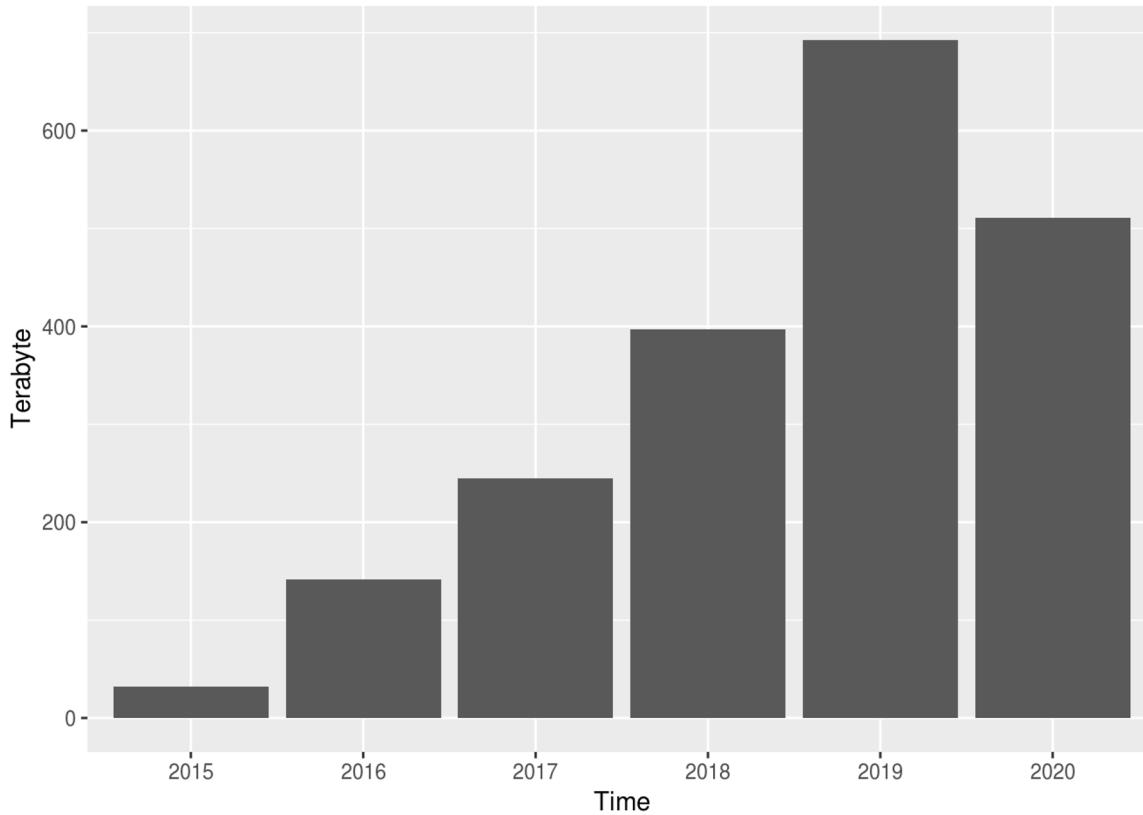
3.

Download results

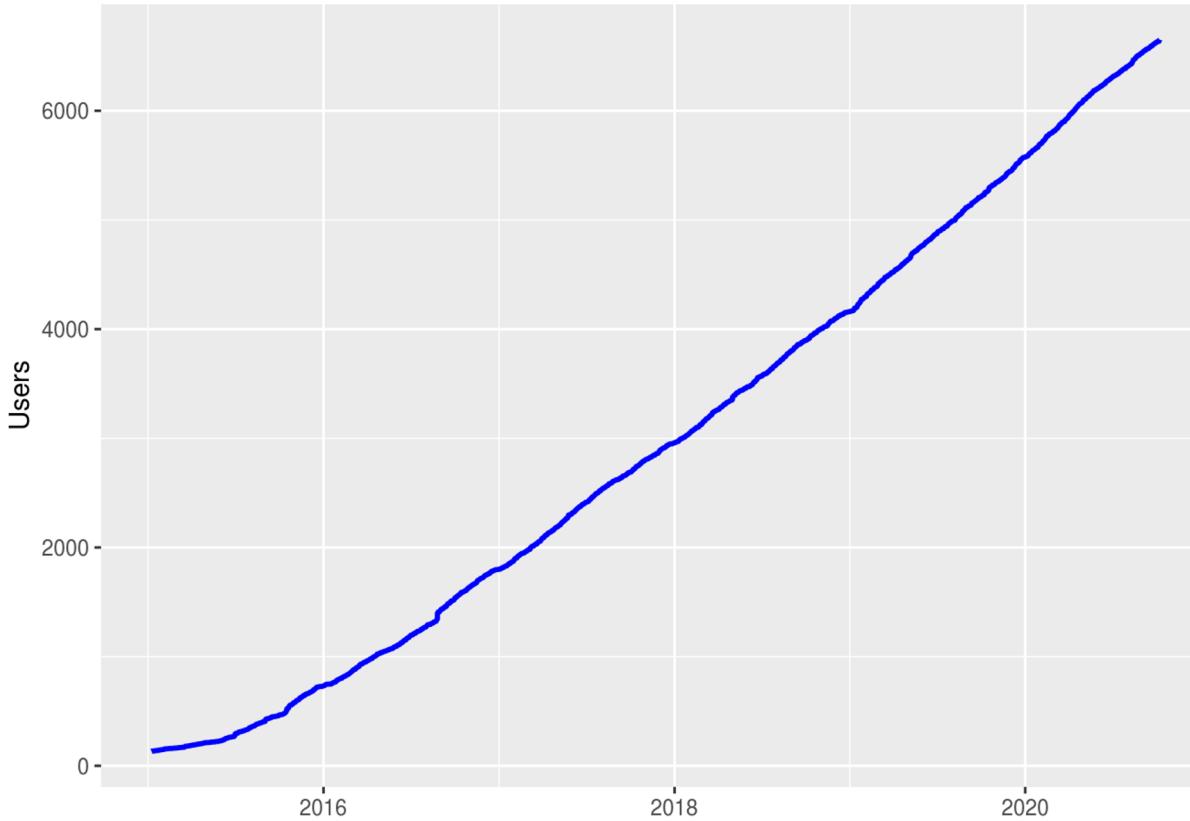
>60M imputed genomes



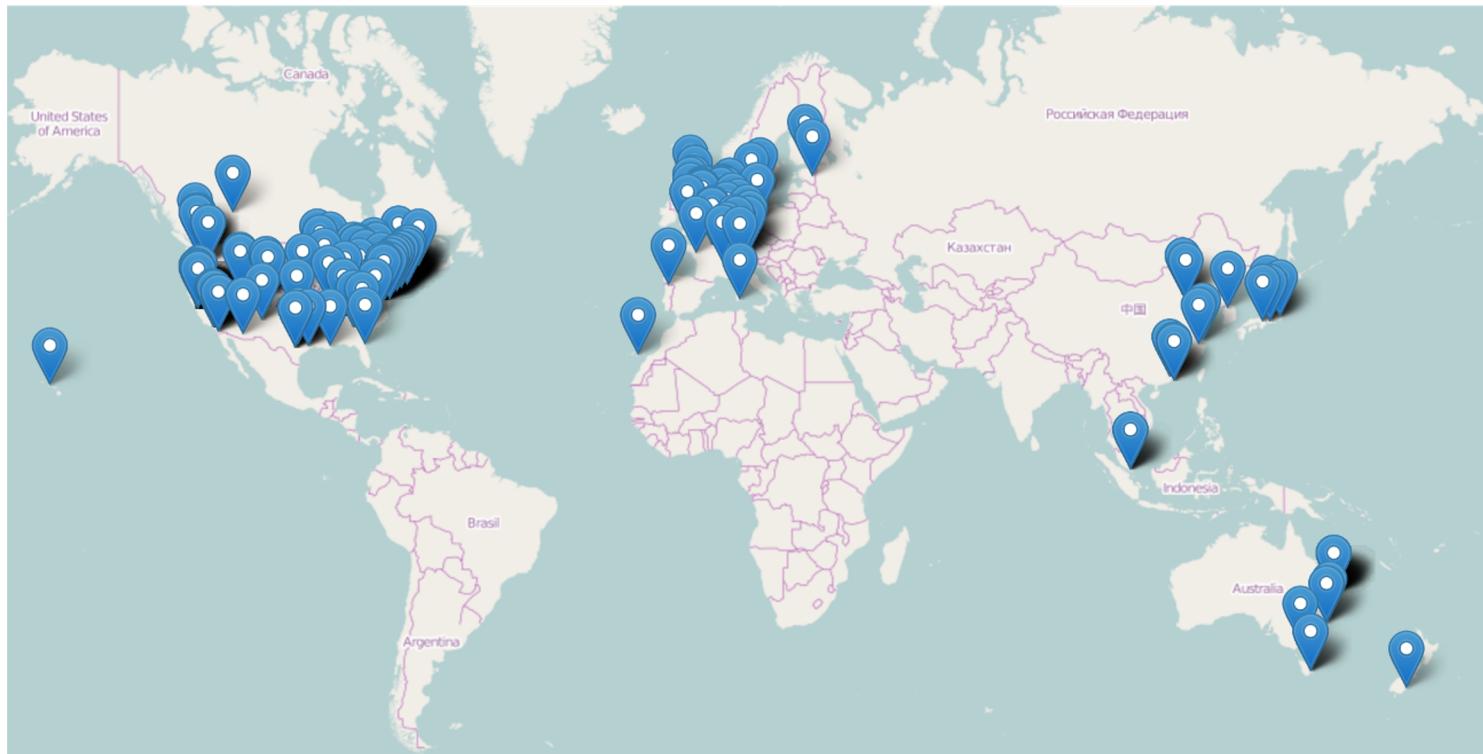
>2 PB data delivered



>6,500 users



Used by researcher world-wide



European General Data Protection Regulation (GDPR)

Added language to MIS to make it GDPR compliant

Currently, unable to sign study-specific agreements

Keep working on this challenge

Summary

Genotype imputation key method in GWAS

Michigan imputation server is easy to use and ensures high quality imputation

Cloud-services will accelerate genetic research so we can devote our time to more interesting tasks

More info and FAQ can be found here:
<https://imputationserver.readthedocs.io>