



MICHIGAN IMPUTATIONSERVER



Christian
Fuchsberger



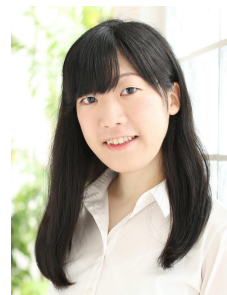
Sebastian
Schönherr



Xueling Sim



Lukas Forer



Saori Sakaue



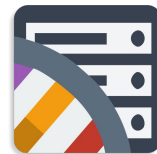
Albert Smith

Section 1

Imputation and the Server



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MICHIGAN
IMPUTATIONSERVER

Setup

- 6 Sessions:
(1) Intro, (2) Use the server, (3) GWAS, (4) Imputation Bot and PGS Server, (5) HLA Imputation, (6) TOPMed
 - Lectures
 - Demos
 - Interaction
 - Our question: Raise your hand or add to the chat
 - Your questions: please add to Q & A box
- Question & Answer session at the end (or email us)

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	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	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	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	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	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	C	T	G	T	G	C

2. Impute

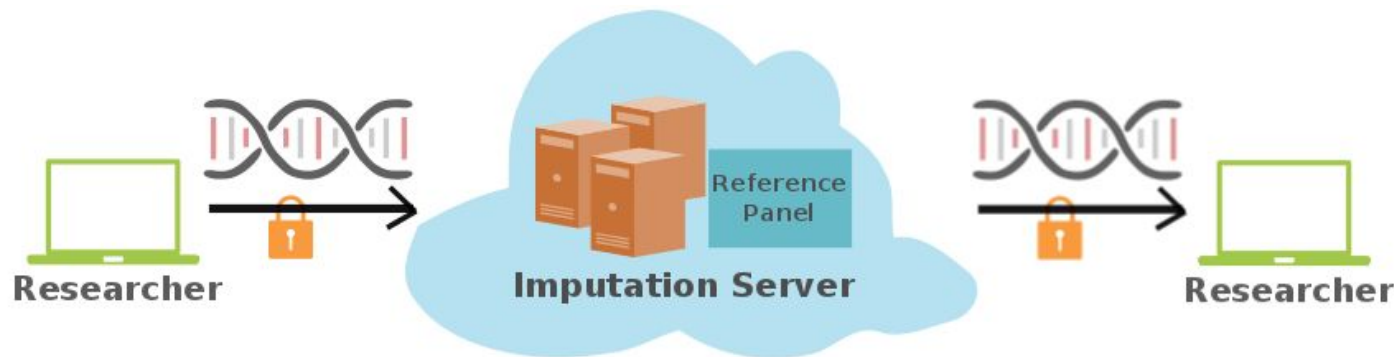
GWAS Haplotypes

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	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	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	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.

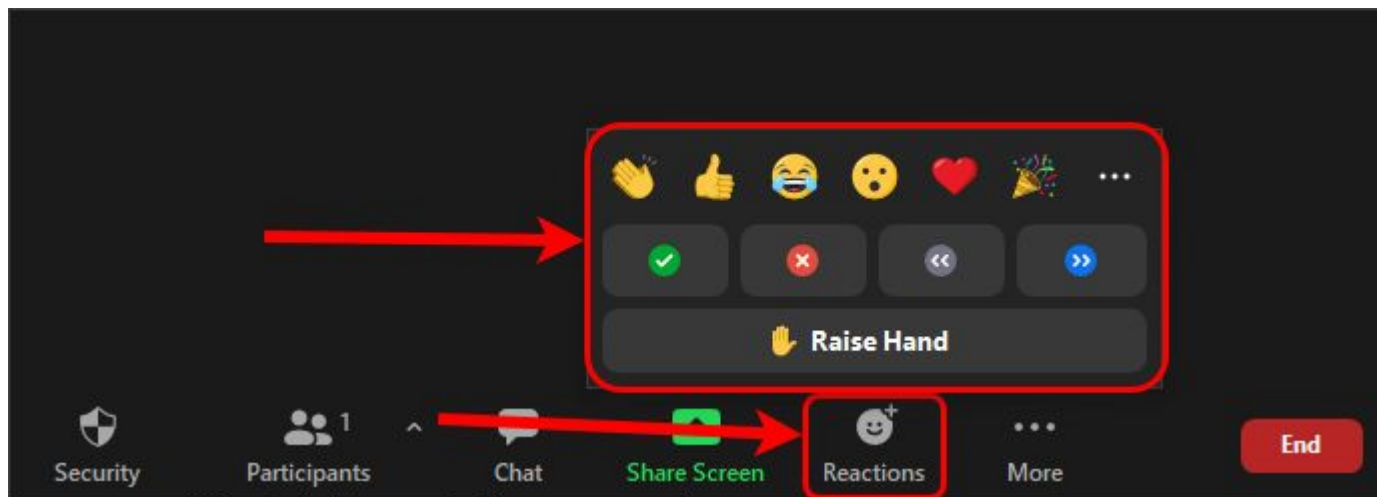
Download results

Have you used the imputation server yet?

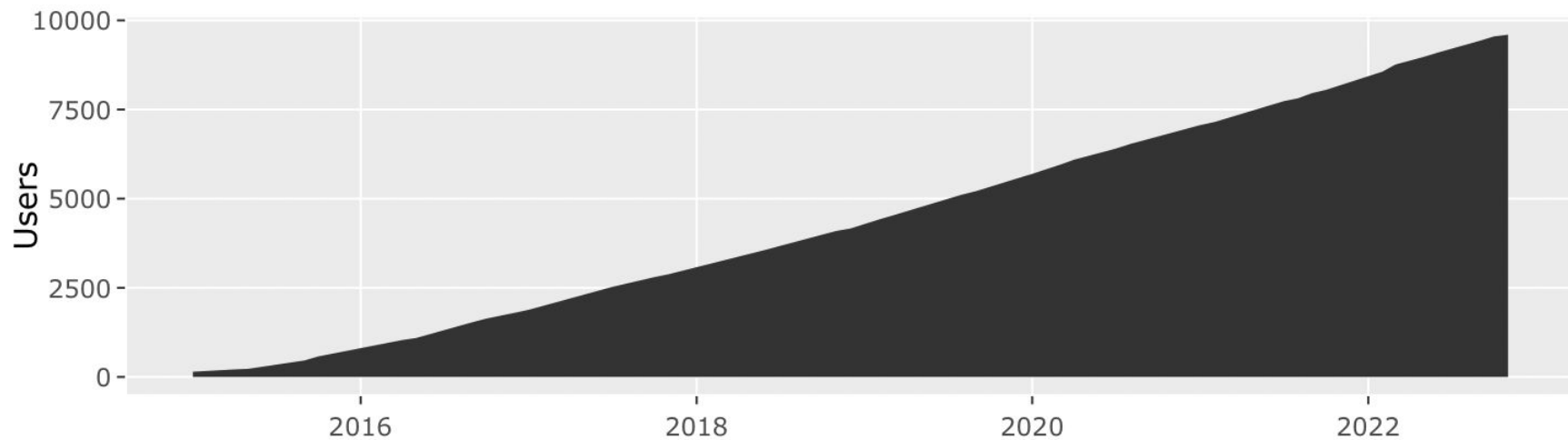
Put a "Yes" in the chat

Or

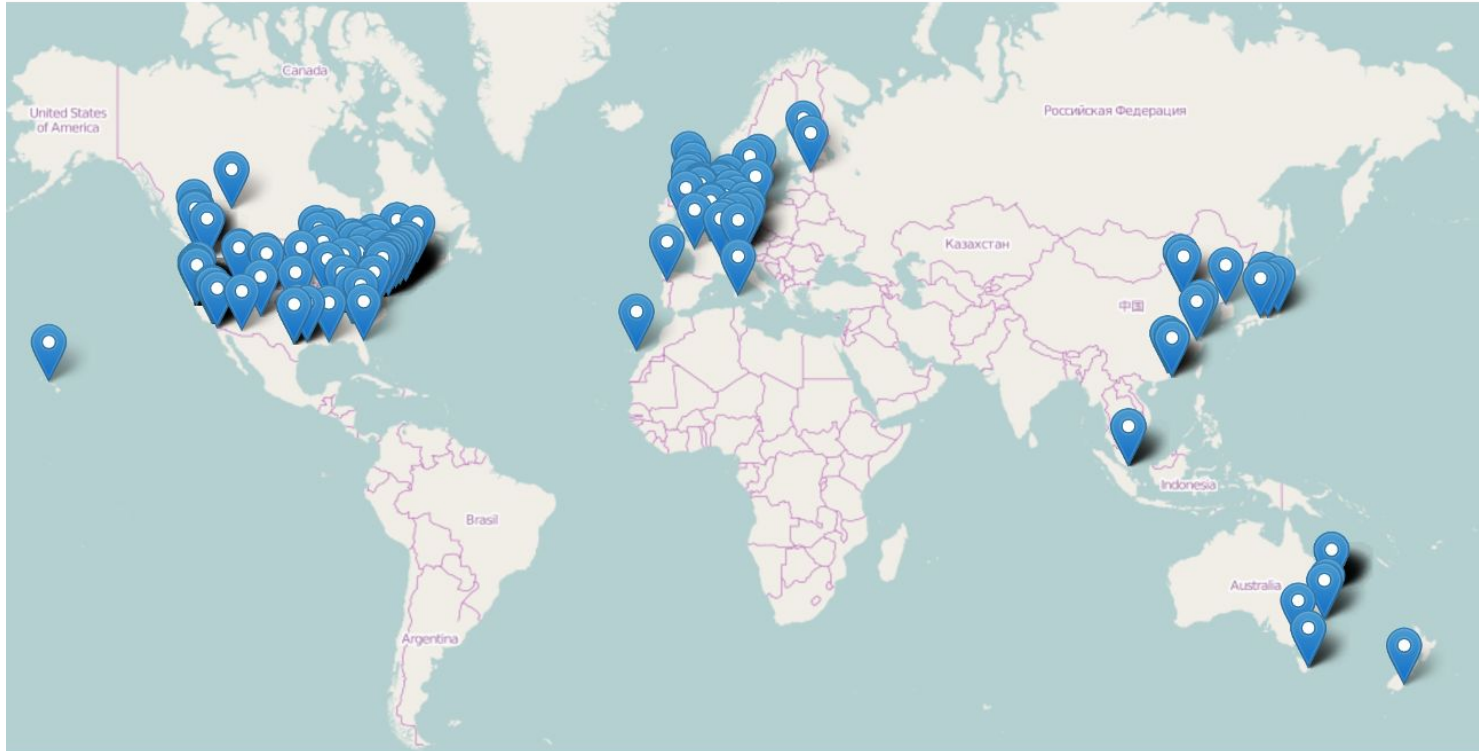
Thumbs up



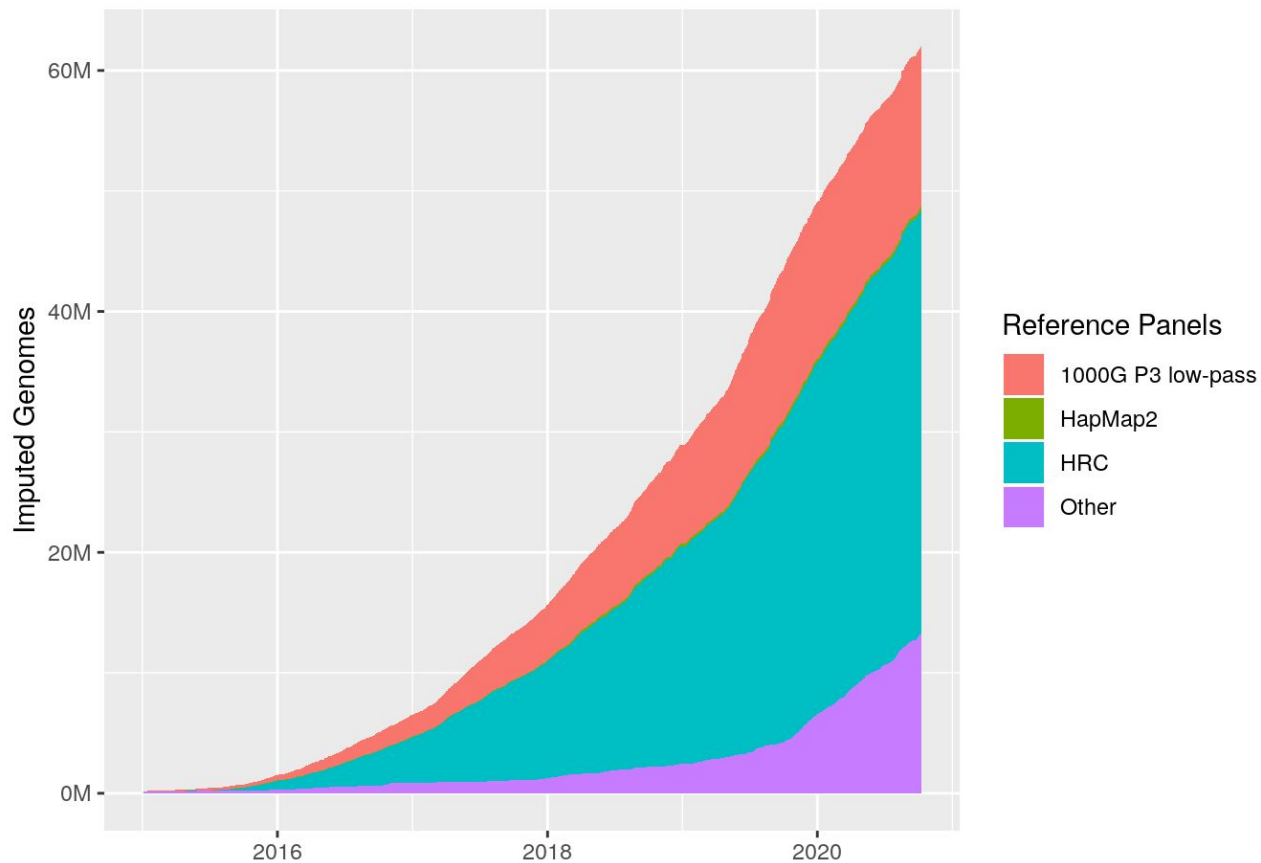
>9,500 users



Used by researcher world-wide



>90M imputed genomes



What's new?

New panels:

- 1000G Phase 3 deep WGS (hg38)
- 1000G Phase low coverage (hg38)
- HLA Imputation Panel
- Genome Asia version 2

New tools

- Beagle phasing
- PGS calculation
- Rsq browser

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>