

# Supplemental Materials

## Estimating the size of long tandem repeat expansions from short reads with ScatTR

Rashid Al-Abri<sup>3,2</sup> and Gamze Gürsoy<sup>1,2,3\*</sup>

<sup>1</sup>Department of Biomedical Informatics, Columbia University, New York, USA

<sup>2</sup>New York Genome Center, New York, USA

<sup>3</sup>Department of Computer Science, Columbia University, New York, USA

\*Corresponding author: Gamze Gürsoy, gamze.gursoy@columbia.edu

## List of Supplemental Materials

- **Supplemental Figures**

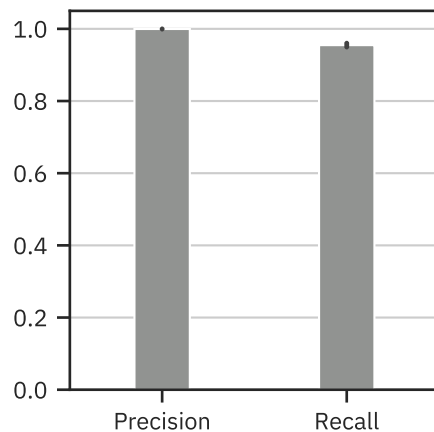
- **Supplemental Figure S1.** Precision and recall of IRR extraction from 540 simulated WGS samples.
- **Supplemental Figure S2.** Absolute error of ScatTR across varying sequencing error rates in simulated WGS samples. Error bars represent 95% confidence intervals across 30 TR loci.
- **Supplemental Figure S3.** Comparison of the estimated sum of alleles for the *C9orf72* repeat expansion by ScatTR, ExpansionHunter, GangSTR, and STRling. The analysis was performed on short-read WGS data from 15 ALS patients with confirmed expansions.

- **Supplemental Tables**

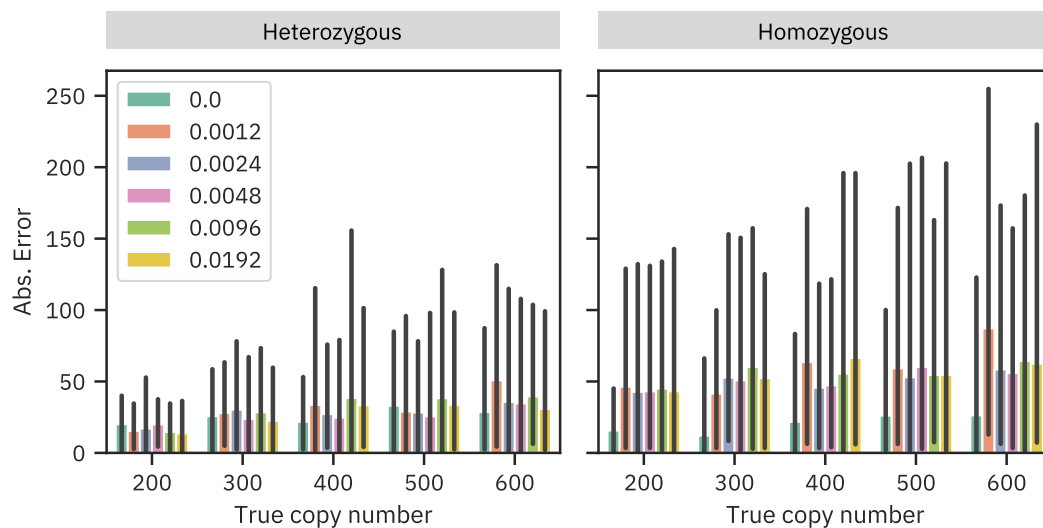
- **Supplemental Table S1.** Regions and motifs of tandem repeat loci used for benchmarking RMSE of ScatTR and state-of-the-art methods

- **Supplemental Table S2.** Regions and motifs of tandem repeat loci with long motifs (21-50 bp) used for benchmarking RMSE of ScatTR and state-of-the-art methods
- **Supplemental Table S3.** Regions and motifs of tandem repeat loci used for benchmarking effect of mutations and sequencing error on ScatTR’s performance
- **Supplemental Table S4.** Sample IDs from the ALS cohort used in this study

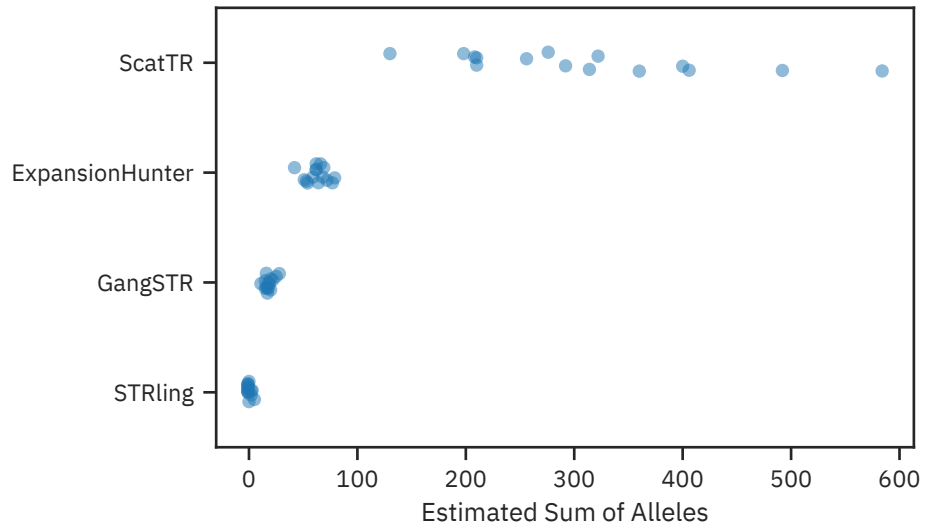
## Supplemental Figures



**Supplemental Figure S1.** Precision and recall of IRR extraction from 540 simulated WGS samples.



**Supplemental Figure S2.** Absolute error of ScatTR across varying sequencing error rates in simulated WGS samples. Error bars represent 95% confidence intervals across 30 TR loci.



**Supplemental Figure S3.** Comparison of the estimated sum of alleles for the *C9orf72* repeat expansion by ScatTR, ExpansionHunter, GangSTR, and STRling. The analysis was performed on short-read WGS data from 15 ALS patients with confirmed expansions.