

**Principal Investigator**

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Applicant Institution

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Application Number / Title

20613 - Assessing allele phasing algorithms

Keywords

Phasing algorithm, GWAS

Application Lay Summary

1a: To identify genetic mutations related to disease, a "genome wide association study" (GWAS) is often conducted. As a technical first step of any such study, it is necessary to assign each heterozygous variant to the paternal or maternal chromosome, a process called "Phasing".

I have developed a new phasing algorithm for large cohorts, and I would like to test this algorithm on the 150,000 genotyped individuals in the UK Biobank.

1b: Health-related, and in the public interest:

The proposed project aims to improve a necessary, technical step in GWAS studies. Improvements would make GWAS studies more efficient, and easier to conduct as cohort sizes grow. This would simplify the discovery of disease-related associations with genetic variants, establishing the health connection. The public interest is served by simplifying the research, potentially speeding up and/or reducing the cost of research.

1c: I would apply the new algorithm to the data, and compare the results with existing algorithms. This is explained in detail here:

<http://biorxiv.org/content/early/2015/12/18/028282>

1d: I am applying for the full data set of genotypes (152,725 individuals).