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Gametic Phasing in a Sample of Distantly Related Individuals Using Segments Shared Identical by Descent (IBD).

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ABSTRACT:

We define "gametic phasing" as the discrimination of the paternal from maternal haplotypes in an individual. Gametic phasing with genome-wide data is straight-forward when one or both parents are included in the sample, but it has not been solved in a sample of unrelated individuals in which genealogical data is unavailable. In the current study, we show how gametic phasing can be accomplished in a sample of conventionally unrelated (i.e., distantly related) individuals with unknown genealogies using segments shared identical by descent (IBD). We show that clustering of IBD sharing among co-relatives of a focal individual into two clusters can allow gametic phasing of the focal individual. By further incorporating SNPs on the mitochondria and Y-chromosome, the paternal and maternal autosomal haplotypes can be distinguished. Such gametic phasing can serve a variety of functions, including estimates of how long assortative mating has occurred in a population, estimates of genetic nurture and vertical transmission without parental data, and parent-of-origin effects.

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