Docket #: S15-361

Identifying individuals from a collection of genomes using allele presence queries

Researchers in Carlos Bustamante's lab at Stanford have developed a method for detecting whether an individual is present in a mixture of genomes. It uses only queries about the presence of alleles to make this decision using genome-wide SNP data. The technology could be extremely useful in forensics when allele frequencies in the mixture cannot be determined, or when individuals from different ethnicities may be present in the mixture.

Currently, all existing methods to detect whether an individual is present in a DNA mixture require knowledge of allele frequencies. But this method detects individuals in a mixture without requiring allele frequencies. The invention allows robust identification of individuals from DNA mixtures (using SNP array data) for many populations in a frequency-independent way.

Applications

- In forensics, could be used to identify individuals (suspects/perpetrators) from a mixture of genomes.

Advantages

- Easy to use, only requires counting number of responses to allele presence queries.
- Does not require information about frequency of alleles.
- The thresholds required to make a confident detection are similar for many different populations.

- Can identify individuals even from a mixture of different populations.
- Can use external frequency information to improve the power of the method.

Publications

- Shringarpure, SS; Bustamante, CD, "Privacy Risks from Genomic Data-Sharing Beacons," The American Journal of Human Genetics, 2015 Nov 5;97(5):631-46. doi: 10.1016/j.ajhg.2015.09.010. Epub 2015 Oct 29.

Patents

- Published Application: WO2017062599
- Published Application: 20180285593
- Issued: 10,747,899 (USA)

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