Personalized genomic medicine faces many hurdles

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When the human genome project was completed in 2003, some expected it to herald a new age of personalized genomic medicine, but the resulting single "reference" sequence has significant shortcomings for these applications and does not account for the actual variability in the human population, as reported in a study published July 11 in the open access journal *PLoS ONE*.

Using genomic data from a large number of individuals, the authors of the study, led by Todd Smith of PerkinElmer in Seattle, Washington, show that current genomic research resources and bioinformatics methods are inadequate for the level of genomic variation among individuals in the population, and that much work will be required before personalized genomic medicine can reach its full potential.

"Resources such as microarrays and bioinformatics programs, as well as guiding assumptions used in genetic studies need to be revised," Dr. Smith explains. "For example, regions of linkage disequilibrium and runs of homozygosity, used to tag and predict disease alleles, are much shorter than previously estimated and we found that many GWAS studies contain potentially complicating unprobed variants."

More information: Rosenfeld JA, Mason CE, Smith TM (2012) Limitations of the Human Reference Genome for Personalized Genomics. *PLoS ONE* 7(7): e40294. doi:10.1371/journal.pone.0040294