PublisherInfo		
PublisherName	:	BioMed Central
PublisherLocation	:	London
PublisherImprintName	:	BioMed Central

Rapid SNP scanning

ArticleInfo		
ArticleID	:	3954
ArticleDOI	:	10.1186/gb-spotlight-20010112-03
ArticleCitationID	:	spotlight-20010112-03
ArticleSequenceNumber	:	25
ArticleCategory	:	Research news
ArticleFirstPage	:	1
ArticleLastPage	:	2
ArticleHistory	:	RegistrationDate : 2001–01–12 OnlineDate : 2001–01–12
ArticleCopyright	:	BioMed Central Ltd2001
ArticleGrants	:	
ArticleContext	:	130592211

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Single nucleotide polymorphisms (SNPs) are pouring into public databases, but methods for analyzing large numbers of SNPs in population studies lag behind. In the January 16 Proceedings of the National Academy of Sciences, Buetow *et al*. report that mass spectrometry (MS) and pooling of DNA samples can be combined to yield a rapid SNP genotyping method (*Proc Natl Acad Sci USA* 2001, **98:**581-584). Buetow *et al*. define candidate SNPs as those sequences that show variation in multiple sequencing runs performed at Washington University (St Louis). DNA from 94 individuals is pooled, and candidate variable regions PCR-amplified. Low nanoliter aliquots are then transferred onto individual 200 µm elements of a 96-element silicon chip. This is the substrate for matrix-assisted laser desorption/ionization time-of-flight (MALDI-TOF) MS. Less than four weeks of work confirms the polymorphism of, and yields accurate allele frequencies for, 3,646 SNPs. This is not yet enough for genome-wide studies of linkage disequilibrium, but suggests that an affordable system for carrying out such studies is not far off.

References

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