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TECHNOLOGY

A step closer to personal genomics?

Advances in sequencing technologies are primarily driven by the promise of medical genomics, which requires efficient and cost-effective sequencing. Stephen Quake and colleagues now show that single-molecule sequencing holds the promise to fulfil these requirements.

First described in 2003, single-molecule sequencing does not require cloning or amplification. In the new study, the authors used the first commercially available single-molecule sequencer, which is made by Helicos Biosciences. Although the method had been described previously (it relies on sequencing by synthesis and direct parallel visualization of DNA molecules), they applied it for the first time to a large and complex genome — that of a human.

Several human genomes have already been sequenced, but these efforts have either used traditional Sanger sequencing or one of the short-read next-generation approaches. The increased efficiency of the single-molecule method allowed the authors to generate the sequence from four data collection runs using a single instrument. The results are of high enough quality for the reliable detection of SNPs and copy-number variations.

This study sets the scene for using single-molecule sequencing to study human variation at an individual genome level. As the authors point out, the fact that this can be achieved with a single commercial instrument 'democratizes' genomic research and brings us a step closer to personal genomics.

Magdalena Skipper, Senior Editor, Nature

ORIGINAL RESEARCH PAPER Pushkarev, D., Neff, N. F. & Quake, S. R. Single-molecule sequencing of an individual human genome. Nature Biotech. 10 Aug 2009 (doi:10.1038/nbt.1561) FURTHER READING Frazer, K. A., Murray, S. S., Schork, N. J. & Topol, E. J. Human genetic variation and its contribution to complex traits. Nature Rev. Genet. 10, 241–251 (2009)