Nature Reviews Genetics | AOP, published online 24 July 2012; doi:10.1038/nrg3309

GENOMICS

Personal genome project

This paper describes the Personal Genome Project (PGP), which has established an open-consent format for the collection of combined genomic and phenotypic information, allowing this data to be shared publicly.

The PGP has so far enrolled more than 1,800 participants, who have given consent for their genomic and phenotypic data — including their health records and personal medical histories — to be made publicly available. Here, Ball, Thakuria, Zaranek *et al.* present summary data from a pilot study of 10 individuals and describe a new tool for processing and sharing this information.

Genomic knowledge regarding potentially damaging mutations is one of the requirements to enable personalized medicine. This pilot study revealed several rare variants that have been implicated in serious diseases. The participants did not have these diseases, thus highlighting the challenge of reporting variants in a clinical setting.

To interpret this information, the authors introduce the Genome–Environment–Trait Evidence (GET-Evidence) system: a software tool that processes genomes and prioritizes variants for further interpretation. Because these records are freely editable by any registered user, the authors hope that the database will provide a forum for achieving public-consensus interpretation of genetic variants, and the project encourages others to contribute data and scientific evaluations. In addition to whole-genome sequence data and phenotypic information, cell lines are established for participants, and these are also publicly available so that other researchers can carry out additional characterization and follow-up functional studies.

Because of concerns regarding re-identification and privacy protection, the PGP only enrols participants who fully understand and agree to the hypothetical and unknown risks that are associated with making biological data available in an open-consent format, and in particular the potential risks regarding loss of privacy to themselves and their relatives. In the future, subsets of participants may choose to contribute to disease-specific research and to additional profiling methods, such as allele-specific expression and epigenetic, metabolomic, proteomic or microbiome profiling. The PGP has been approved for up to 100,000 participants and so has the potential to be a widely used resource for moving towards personalized health care.

Bryony Jones

ORIGINAL RESEARCH PAPER M. P. Ball *et al.* A public resource facilitating clinical use of genomes. *Proc. Natl Acad. Sci. USA* 13 July 2012 (doi:10.1073/pnas.1201904109)

Photodisc