

TOOLS IN BRIEF

GENOMICS

The United Kingdom's 10K genome project

To learn how rare genetic variations affect human phenotypes, one needs linked genotypic and phenotypic data at large scale. The United Kingdom's 10K (UK10K) project provides such data. Researchers carried out sequencing of the genomes and exomes of close to 10,000 individuals, at low and high read depth, respectively, together with extensive phenotypic characterization. Participants ranged from healthy individuals to those with rare diseases, severe obesity and neurodevelopmental disorders. The UK10K consortium characterized more than 24 million new variants and assessed their association with diseases. Although the project found few rare mutations with large effects on phenotype, the data will inform the design of future association studies and provide an accurate imputation reference panel. The project provides a valuable resource of genetic and phenotypic data at the individual level. The UK10K Consortium. *Nature* **526**, 82–90 (2015).

IMAGING

Optical clearing with a gentle touch

Fluorescence imaging of biological tissues is challenging owing to high absorption and strong scattering of light. One strategy for enhanced imaging is to render tissues transparent by optical clearing. However, clearing reagents can damage tissues, obscuring morphological features and reducing the brightness of genetically encoded fluorophores contained in the tissue. To overcome these limitations, Hama *et al.* have introduced a clearing technology called ScaleS that uses sorbitol, a mild tissue-permeant sugar alcohol for gentle clearing that inflicts minimal damage to tissues. They found that ScaleS allowed good preservation of fluorescence signals as well as morphology even at the level of electron microscopy, and they were able to use it to study amyloid plaque formation associated with Alzheimer's disease in mouse and human brains.

Hama, H. *et al. Nat. Neurosci.* **18**, 1518–1529 (2015).

GENOMICS

De novo genome assembly with nanopores

Nanopore sequencing holds the potential to deliver long, single-molecule reads that should be ideally suited for *de novo* genome assembly. But the current accuracy of this technology—only ~85%—makes this application challenging. To correct many of the errors in nanopore reads, Szalay and Golovchenko developed PoreSeq, an algorithm that increases accuracy to up to 99%, as shown for a bacteriophage genome. PoreSeq models uncertainties in the measurement of changes in ion current as a DNA molecule is ratcheted through the pore and compares multiple reads of the same region to compute the most likely sequence for each read. The authors used these error-corrected reads to assemble the genomes of *Escherichia coli* and the λ -phage. They also were able to call single-nucleotide variants with 99.1% accuracy at 16 \times coverage, an order of magnitude lower than previous results.

Szalay, T. & Golovchenko, J.A. *Nat. Biotechnol.* **33**, 1087–1091 (2015).

PLANT BIOLOGY

Lighting up root architecture

A plant's root architecture determines its access to water and nutrients in the soil. However, most roots are studied in the light on artificial media, and much remains unknown about how soil-embedded roots grow and change in response to environmental cues. Rellán-Álvarez *et al.* have developed a new imaging platform called Growth and Luminescence Observatory for Roots (GLO-Roots). In GLO-Roots, *Arabidopsis* plants expressing one or more luciferases are grown and imaged in soil in thin chambers in the dark. Prior to imaging, luciferin is added to the plants during watering to generate a bioluminescent signal that is recorded with a custom imaging system. The team also developed algorithms for analyzing root data that can be implemented in ImageJ. GLO-Roots enabled the authors to track root growth under different conditions and make quantitative comparisons of root growth in different *Arabidopsis* strains.

Rellán-Álvarez, R. *et al. eLife* doi:10.7554/eLife.07597 (2015).