RESEARCH HIGHLIGHTS

IN BRIEF

COMPLEX TRAITS

Genome-wide association mapping in rice

This study advances research aiming to understand the genetic basis of agriculturally important traits in Asian rice, *Oryza sativa*. The authors present a genome-wide association study (GWAS) including 413 rice varieties from 82 countries and provide a public resource of all of the genotypes and phenotypes and of the seed stocks. They found substantial genetic heterogeneity among varieties and suggest strategies for performing GWASs in species with extensive population structure, such as rice.

ORIGINAL RESEARCH PAPER Zhao, K. et al. Genome-wide association mapping reveals a rich genetic architecture of complex traits in Oryza sativa. Nature Commun. 2, 467 (2011)

CHROMATIN

An evolutionary perspective on nucleosome positioning

Through a comparison of genome-wide nucleosome occupancy maps from 13 species of fungi, these authors provide insight into the relationship between genome evolution and chromatin structure. Although they found substantial conservation across species in levels of nucleosome depletion at many sites, they also found important differences among species in the roles played by sequence or *trans*-acting factors in determining nucleosome positioning. This work suggests evolutionary plasticity in regulation of chromatin structure.

ORIGINAL RESEARCH PAPER Tsankav, A. et al. Evolutionary divergence of intrinsic and trans-regulated nucleosome positioning sequences reveals plastic rules for chromatin organization. Genome Res. 13 Sep 2011 (doi:10.1101/gr.122267.111)

HUMAN GENOMICS

Relationships between *cis*-regulatory and coding variants

Analysis of data from the human <u>1000 Genomes Project</u> has revealed that common *cis*-regulatory variants can modify the distribution of rare coding variants. When a regulatory single-nucleotide variant (SNV) increases expression of a gene, there tend to be lower numbers of coding variants in that gene than would be predicted from the action of purifying selection. Interestingly, this pattern was not seen for disease-associated regulatory variants. This work has implications for understanding the function of genetic variants and how the spectrum of variation has arisen.

ORIGINAL RESEARCH PAPER Lappalainen, T. et al. Epistatic selection between coding and regulatory variation in human evolution and disease. Am. J. Hum. Genet. 89, 459–463 (2011)

DNA SEQUENCING

Exome sequencing technologies compared

In this paper, three commercial whole-exome sequencing platforms — from Agilent, Nimblegen and Illumina — are systematically compared based on their ability to detect variants in the same human sample. The balance between exome coverage and target-enrichment efficiency was influenced by the different design features (such as bait density) and parameter settings of each platform. Interestingly, exome sequencing detects variants that are missed by wholegenome sequencing, suggesting that deep-targeted sequencing affords greater sensitivity than even genome coverage. **ORIGINAL RESEARCH PAPER** Clark, M. J. *et al.* Performance comparison of exome DNA sequencing technologies. *Nature Biotech.* 25 Sep 2011 (doi:10.1038/nbt.1975)