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IN BRIEF

PLANT GENOMICS

Associative transcriptomics

The authors developed a method for carrying out genome-wide association studies in polyploid plants that is applicable to species with no genomic resources. Their rationale is to make use of the SNP and gene expression variation data available in RNA sequencing (RNA-seq) data sets. These data are mapped onto a genomic architecture assembled by SNP linkage mapping and using data from related species, and the variants are then correlated with traits. They applied this method to a panel of 84 *Brassica napus* accessions and identified quantitative trait loci for glucosolinate content of seeds.

ORIGINAL RESEARCH PAPER Harper, A. L. *et al.*, Associative transcriptomics of traits in the polyploid crop species *Brassica napus*. *Nature Biotech.* 22 Jul 2012 (doi:10.1038/nbt.2302)

GENOMIC TECHNOLOGY

Recombination maps get personal

Wang *et al.* developed a microfluidic method for single-cell, whole-genome amplification. This enabled the generation of detailed maps of recombination and mutation in sperm, thus revealing the changes that occur in individual meioses. The recombination rate was consistent with estimates from human population studies, although the sites of recombination hotspots differed. Furthermore, the authors quantified the rates of aneuploidy and *de novo* point mutations; such measurements of genomic instability are confounded by negative selection in population studies.

ORIGINAL RESEARCH PAPER Wang, J. *et al.* Genome-wide single-cell analysis of recombination activity and *de novo* mutation rates in human sperm. *Cell* **150**, 402–412 (2012)

COMPLEX TRAITS

Extreme phenotypes assist association studies

Infection with *Pseudomonas aeruginosa* is a common complication in patients with cystic fibrosis. Emond *et al.* carried out exome sequencing of 91 patients who represented either end of the phenotypic spectrum of infection severity. They found variants in dynactin 4 (*DCTN4*) that correlated with earlier and more severe infection, and these were validated by targeted resequencing of an independent cohort. This demonstrates the proof of principle that exome sequencing and an extreme phenotype design can identify complex trait genes.

ORIGINAL RESEARCH PAPER Emond, M. J. *et al.* Exome sequencing of extreme phenotypes identifies *DCTN4* as a modifier of chronic *Pseudomonas aeruginosa* infection in cystic fibrosis. *Nature Genet.* **44**, 886–889 (2012)

GENOMIC INSTABILITY

Meta-analysis of aneuploid cell gene expression

How aneuploidy leads to aberrant cellular and organismal phenotypes remains poorly understood, although a transcriptional signature that is characteristic of a stress response has been seen in aneuploid yeast and mouse cells. In this study, the authors analysed a large collection of gene expression data from aneuploid cells from diverse organisms. This revealed that transcriptional changes associated with stress and decreased proliferation occur across species and are largely independent of the identity of the aneuploid chromosome. Thus, eukaryotic cells seem to have a generic response to a chromosome-scale gene dosage change.

ORIGINAL RESEARCH PAPER Shelzter, J. M. *et al.* Transcriptional consequences of aneuploidy. *Proc. Natl Acad. Sci. USA* **109**, 12644–12649 (2012)