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# **IN BRIEF**

### **GENOME EVOLUTION**

#### To have or have not — recurrent intron loss in plants

A comprehensive analysis of five sequenced grass genomes provides new insights into the patterns of recurrent intron loss throughout evolution and its underlying molecular mechanisms. Wang *et al.* identified 93 recurrent intron losses in 883 regions where genomes varied with regards to intron presence or absence. Recurrent intron loss was nonrandom and affected only a small number of introns that were repeatedly lost in multiple lineages. The study additionally highlights a possible role for DNA methylation in the process of intron loss. **ORIGINAL RESEARCH PAPER** Wang, H., Devos, K. M. & Bennetzen, J. L. Recurrent loss of specific introns during angiosperm evolution. *PLoS Genet.* **10**, e1004843 (2014)

## CHROMATIN

#### High-resolution 3D mapping of loops

US researchers have mapped the spatial organization of the human genome at an unprecedentedly high resolution of 1 kb using *in situ* Hi-C, an updated Hi-C methodology that enables DNA proximity ligation in intact nuclei. This approach yielded maps comprising 5 Tb of sequence data, with more than 15 billion distinct contacts recorded, which is an order of magnitude greater than all previously published Hi-C data sets combined. The maps reveal conserved contact domains and the existence of ~10,000 chromatin loops, which often demarcate these contact domains. A great majority of loops were found to be anchored at a pair of convergent CTCF binding sites.

ORIGINAL RESEARCH PAPER Rao, S. S. P. *et al.* A three-dimensional map of the human genome at kilobase resolution reveals principles of chromatin looping. *Cell* <u>http://dx.doi.org/10.1016/j.cell.2014.11.021</u> (2014)

## TECHNOLOGY

#### Nanopore sequencing for clinical diagnostics

The MinION nanopore sequencer, a device the size of a large USB memory stick, has been successfully used with Illumina HiSeq data to identify a previously uncharacterized insertion site of a chromosomal antibiotic resistance island (which is flanked by repetitive sequence and likely to have been horizontally transferred) in a strain of *Salmonella enterica* subsp. *enterica* serovar Typhi. Nanopore technology can produce long sequence reads that are necessary for characterizing such complex repetitive sequence regions, and this proof-of-principle study demonstrates the potential of this technology for tracking the spread of bacterial pathogens and, ultimately, its future applicability in the clinic.

ORIGINAL RESEARCH PAPER Ashton, P. M. et al. MinION nanopore sequencing identifies the position and structure of a bacterial antibiotic resistance island. *Nature Biotech*. <u>http://dx.doi.org/</u>10.1038/nbt.3103 (2014)

# **EVOLUTION**

#### Gene age begets constraint

Gene expression constraints increase over time, which may be the result of the integration of genes into gene networks and the functional cellular environment. *Cis* expression quantitative trait loci (eQTLs) of younger genes have a higher effect size, are located nearer to transcription start sites and usually influence genes in more tissues and populations. In addition, the heritability of gene expression increases over time, which indicates that the influence of environment on gene expression levels decreases with gene age.

ORIGINAL RESEARCH PAPER Popadin, K. Y. et al. Gene age predicts the strength of purifying selection acting on gene expression variation in humans. Am. J. Hum. Gen. 95, 660–674 (2014)