TOOLBOX

Massive atlas documents genome's regulatory regions

BY **JESSICA WRIGHT**

7 MAY 2014

Researchers have sequenced hundreds of human and mouse genomes to generate an atlas of 180,000 regions, called promoters, that regulate the expression of adjacent genes, they reported 27 March in *Nature*[1].

In a companion study, they identified another 43,000 regions, called enhancers, that influence the expression of distant genes[2].

The new resource may clarify whether genetic variants linked to a particular disorder, such as autism, disrupt an enhancer or promoter region.

The members of a large consortium, called FANTOM5 (for functional annotation of the mammalian genome), sequenced all the genetic transcripts, or RNAs, in 975 human and 399 mouse samples. These samples include cells particular to certain organs, tissues and cancer cell lines.

The researchers linked the expression of these regulatory elements to networks of genes. They identified at least one such region for more than 95 percent of the genes in the human genome, and cataloged 43,011 enhancers.

They also found one group of enhancers that is specific to the fetal brain, suggesting that these enhancers play a role in development. One of these is located near **MEF2C**, a gene linked to autism and developmental delay. About 40 percent of enhancers probably influence the nearest gene, the study found.

The researchers also looked at results from genome-wide association studies, which link common variants to the likelihood of having a certain disease or disorder. Genetic variants associated with a disease are more likely to land in enhancer regions than in other non-protein-coding regions of the genome, the study found.

The new findings may help researchers understand the function of genetic variants and link regulatory regions to patterns of gene expression.

References:

1: FANTOM consortium and the RIKEN PMI and CLST *Nature* **507**, 462-470 (2014) **PubMed**

2: Andersson R. *et al. Nature* **507**, 455-461 (2014) **PubMed**