

**Digital RNA allelotyping reveals tissue-specific and allele-specific gene expression in human.**

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**Public Summary:**

**Scientific Abstract:**

We developed a digital RNA allelotyping method for quantitatively interrogating allele-specific gene expression. This method involves ultra-deep sequencing of padlock-captured single-nucleotide polymorphisms (SNPs) from the transcriptome. We characterized four cell lines established from two human subjects in the Personal Genome Project. Approximately 11-22% of the heterozygous mRNA-associated SNPs showed allele-specific expression in each cell line and 4.3-8.5% were tissue-specific, suggesting the presence of tissue-specific cis regulation. When we applied allelotyping to two pairs of sibling human embryonic stem cell lines, the sibling lines were more similar in allele-specific expression than were the genetically unrelated lines. We found that the variation of allelic ratios in gene expression among different cell lines was primarily explained by genetic variations, much more so than by specific tissue types or growth conditions. Comparison of expressed SNPs on the sense and antisense transcripts suggested that allelic ratios are primarily determined by cis-regulatory mechanisms on the sense transcripts.

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