The Genotype-Tissue Expression (GTEx) Project

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Understanding the functional consequences of genetic variation, and how it impacts complex human disease and quantitative traits, remains a critical challenge for biomedicine. Over the last decade we've witnessed a dramatic increase in our understanding of the role of genetic variation in common disease, with thousands of genetic variants, influencing human diseases and other traits, now catalogued. The majority of these variants are non-coding and presumed to exert their effects via regulation of gene expression and epigenetic processes. Given that pathogenic processes are often tissue-specific, characterizing the regulatory architecture of the human genome from a range of disease-relevant tissues is essential to understand both basic biology and interpreting GWAS association data. The Genotype-Tissue Expression (GTEx) Project was created to support this effort by establishing a public resource database to enable studies of the relationship between genetic variation and gene expression in multiple human tissues. The project has an end goal of analyzing ~20,000 tissues from over 900 donors. Data will be presented from analyses conducted on the pilot and latest datasets (from ~400 donors with RNA sequence data from >7,000 tissues), highlighting the transcriptional landscape across human tissues, and how those data can be integrated with genetic association data to unravel patterns of genetic variation and gene regulation across diverse human cell types, and uncover tissue-specific processes of complex disease.