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Using system genetics
to explore the complexity
of leaf shape variation
in *Populus tremula*

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Abstract

Leaves are essential for sustaining humanity as they function as the energy and oxygen-producing organ of plants. Intensive research on physiological processes has contributed immensely to our understanding of the function of leaves. However, comparatively little is known about how leaf size and shape is determined. The aim of my PhD was to assay leaf shape variation among individuals of *Populus tremula* (European aspen) sampled across the distribution range of Sweden to characterize the genetic architecture underlying variation, including elucidating contributing molecular mechanisms.

In this PhD I employed an integrated systems genetics and systems biology approach to identify genetic components of variation and to assign biological function to these. We integrated population-wide data on leaf shape, gene expression and genome variation from a collection of *P. tremula* genotypes and used this to perform genome-wide association studies. We then integrated these results with a systems biology transcriptomics study of leaf development to provide developmental and biological context. We demonstrate that our developmental gene expression series captured known homologs of functionally characterized *Arabidopsis thaliana* genes and biological processes of importance during leaf development. In addition to these known genes of high importance, we also identified many novel candidate genes. Our systems genetics approach identified numerous genes with a potential role in leaf development that was supported by the developmental time series. From our association studies and population analyses we have shown that there are no large-effect loci contributing to variation in leaf shape and that highly ranked loci associated with leaf shape are primarily located in the regulatory regions of genes. Furthermore, we identified loci controlling variation in gene expression and sets of genes with significant differential expression between groups of genotypes with highly contrasting leaf shapes. We show that genes with significant associations influencing expression among genotypes are enriched in the periphery of the corresponding gene co-expression network and that they experience relaxed selective constraint. Taken together, these results suggest that leaf shape is a highly complex trait controlled by a large number of loci, each contributing only a small effect, that those loci likely act via modulation of gene expression and that they do not show signals of adaptive selection. In addition, we adapted and optimized the method of spatial transcriptomics for use in plant species. This method provides a transcriptome-wide *in situ*, spatially-resolved assay of transcript expression at high spatial resolution.

Keywords

Populus, Arabidopsis, systems biology, systems genetics, spatial transcriptomics (ST), single nucleotide polymorphism (SNP), Genome wide associations study (GWAS), expression GWAS (eGWAS)

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