BpPIN1 的可遗传表观修饰与裂叶桦的叶形相关联

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摘 要：【目的】欧洲白桦（*Betula pendula*）的变种裂叶桦（*B. pendula* ‘Dalecarlica’）叶缘呈掌状开裂，因此具有很高的观赏价值。本研究旨在鉴定裂叶形成的遗传基因及作用机理。【方法】我们进行了混池分离群体分析（BSA-Seq）、基于分子标记的精细定位和 SNP 等位性检测，以鉴定控制裂叶桦叶形的基因。随后，展开了实时荧光定量 PCR，染色质免疫共沉淀测序，全基因组重亚硫酸盐测序，小 RNA 测序，McrBC-PCR 等工作，探究目的基因的表观遗传学变异。【结果】裂叶桦叶形由 *BpPIN1* 基因控制，该基因编码一个作为生长素外排载体的 PIN-FORMED 蛋白。在裂叶桦中，*BpPIN1* 的表达水平升高，但不存在基因序列的变异，且位于 *BpPIN1* 启动子区的 SNP 变异不能与裂叶性状相关联。多组学测序联合分析表明，欧洲白桦 *BpPIN1* 启动子上存在 RdDM 甲基化位点，维持着高度 CHH 型甲基化，在裂叶桦中有两个 CHH 型甲基化位点的甲基化水平显著下降，且伴随着小 RNA 的减少。进一步的 McrBC-PCR 检测证实了 *BpPIN1* 启动子区域的低甲基化与裂叶性状紧密关联。【结论】*BpPIN1* 启动子区域的 DNA 甲基化程度与裂叶桦的叶形有关，降低的甲基化水平导致裂叶桦 *BpPIN1* 表水平的上升，促进了叶脉的生长，从而形成裂片。本研究结果揭示了 *BpPIN1* 调控裂叶桦叶片形状的表观遗传机制，有助于观赏性状的分子育种。

关键词：裂叶桦；DNA 甲基化；叶形；PIN-FORMED (PIN)家族基因；混池分离群体分析

Heritable epigenetic modification of *BpPIN1* is associated with leaf shapes in Betula pendula

Abstract: The new variety *Betula pendula* ‘Dalecarlica’ selected from Betula pendula, shows high ornamental values owing to its lobed leaf shape. In this study, to identify the genetic components of leaf shape formation, we performed bulked-segregant analysis (BSA) and molecular marker-based fine mapping to identify causal gene responsible for lobed leaves in B. pendula ‘Dalecarlica’. The most significant variations associated with leaf shape were identified within the gene BpPIN1 encoding a member of PIN-FORMED family, responsible for the auxin efflux carrier. We further confirmed the hypomethylation at the promoter region promoting the expression level of BpPIN1, which cause stronger and longer veins and lobed leaf shape in B. pendula ‘Dalecarlica’.

These results indicated that DNA methylation at the BpPIN1 promoter region is associated with leaf shapes in Betula pendula. Our findings revealed an epigenetic mechanism of BpPIN1 in the regulation of leaf shape in birch, which could help in molecular breeding of ornamental traits.

Key words: *Betula pendula* ‘Dalecarlica’; DNA methylation; Leaf shape; PIN-FORMED (PIN); BSA-seq