

WHOLE GENOME ASSEMBLY OF KARELIAN BIRCH AND ANALYSIS OF CHROMOSOME REGIONS ASSOCIATED WITH THE CURLY WOOD PHENOTYPE

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Introduction. Karelian birch, also referred to as curly birch (*Betula pendula* var. *carelica*), is a unique variation of the silver birch native to northern Europe, including Finland, Russia, and parts of Scandinavia [1]. It is highly valued for its distinctive wood grain, which features tight curls, swirls, and waves. Known for its strength and durability, Karelian birch is commonly used in high-end furniture, cabinetry, flooring, and decorative items. The wood's contrasting light and dark patterns enhance its visual appeal, earning it the nicknames "royal tree" and "wooden marble" [2]. The primary challenge in propagating curly birch in plantations is that the initial indications of 'curliness' in the trees emerge when they are between 8 and 15 years old [3]. Until that point, it is essential to manage a large plantation of trees with uncertain yields, as any stands cultivated from Karelian birch seeds will inevitably include a mix of regular (non-curly) birches [4]. Recently, a 54 bp deletion was proposed as a molecular marker for identifying birch trees with the curly wood phenotype (BpCW1) [5]. Unfortunately, the RADseq approach used in that study did not provide comprehensive resolution of this region. Further research is needed to identify candidate genes linked to that trait and to explore structural differences among birches.

Main part. De novo genome assembly was performed using MaSuRCA software which took raw Illumina paired-end reads as an input. The polishing step was conducted with Pilon software. Scaffolding was carried out using RagTag with the Nucmer aligner option, employing the Silver birch genome as a reference [6]. In advance, to check the consistency of alignment, scaffolds were mapped to the reference with Nucmer (MUMmer4) and then visualized with dotPlotly. The quality of the chromosome assembly was evaluated using BUSCO and QUAST. Structural variations between Karelian and Silver birches were detected using SyRI software. Prior to this analysis, whole genome alignment was conducted using the minimap2, allowing for up to 20% sequence divergence (asm20).

Conclusion. To determine the true causal polymorphism whole genome assembly was performed using WGS reads. Additionally, more genetically diverse plant material was utilized to ensure that the identified mutations were not region-specific. The obtained results are consistent: a 54 bp deletion associated with curly phenotype is also present in the current assembly. Furthermore, structural variations between Karelian and Silver birch genomes were detected, resulting in a total of 546223 SNPs, 81862 insertions and 68640 deletions.

References:

1. Vetchinnikova, L. V. & Titov, A. F. "The Karelian birch: A unique biological object". *Biol. Bull. Rev.* 10, 102–114 (2020).
2. Viherä-Aarnio, A. & Hagqvist, R. "Curly birch (*Betula pendula* var. *carelica*), wooden "marble" from finland-soon easily available". In *Proceedings of the International Scientific Conference on Hardwood Processing* (Lahti, Finland, 2017).
3. Ermakov, V. "Mekhanizmy adaptatsii berezy k usloviyam Severa[Mechanisms of birch adaptation to the conditions of the North]". *Izd-vo " Nauka," Leningradskoe otd-nie*, 1986.
4. Vetchinnikova, L. V. & Titov, A. F. "Curly birch: Some secrets remain". *Biol. Bull. Rev.* 13, 162–174 (2023).
5. Gubaev, R., Karzhaev, D., Grigoreva, E. *et al.* "Dissection of figured wood trait in curly birch (*Betula pendula* Roth var. *carelica* (Mercklin) Hämet-Ahti) using high-throughput genotyping". *Sci Rep* 14, 5058 (2024).

6. Salojärvi, J., Smolander, OP., Nieminen, K. *et al.* “Genome sequencing and population genomic analyses provide insights into the adaptive landscape of silver birch”. *Nat Genet* 49, 904–912 (2017).