Mako - High Resolution Immunogenomics From NGS Data

BENTHIC GENOMICS

What NGS Pipelines Miss in the MHC

NGS is ubiquitous in genomics, but the MHC remains difficult to analyze. Most workflows split the job across separate tools for HLA typing, variant calling, and sometimes phasing, even though many of those tools are not designed for the MHC's polymorphism, structural variation, copy-number variability, and paralogous genes.

The result is partial coverage, ambiguous calls, and missing haplotype context in one of the genome's most important immune regions.

What Mako Delivers

Phased MHC Haplotypes

Mako reconstructs haplotypes across the MHC, enabling fine-mapping, association studies, and clearer interpretation of immunogenomics.

High-resolution HLA Typing

Mako delivers 4-field resolution HLA typing with extended coverage across 45 genes in the MHC.

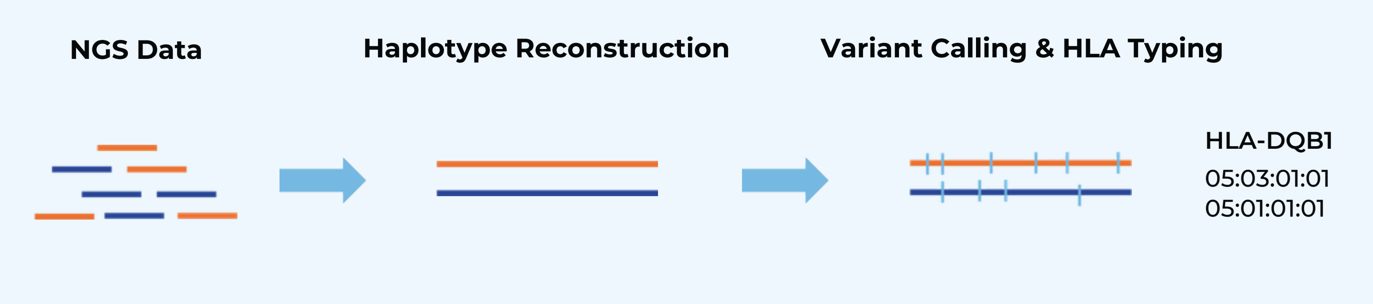
SNPs & SVs across the MHC

Mako goes beyond classical HLA genes to recover dense variant calls across the MHC, including immune genes such as MICA/B and TAP1/2.

What Makes Mako Different

Mako combines capabilities that are typically split across multiple tools in one short-read workflow: phased haplotype reconstruction, high-resolution HLA typing, and comprehensive immune-region variant calling. Instead of treating the MHC as a narrow HLA-calling problem, Mako is built for the most complex regions of the genome, like MHC, and reconstructs haplotypes before calling variants and HLA alleles.

Mako reconstructs long-range haplotypes and calls SNPs and SVs across the entire MHC for an added layer of discovery potential



NGS Data

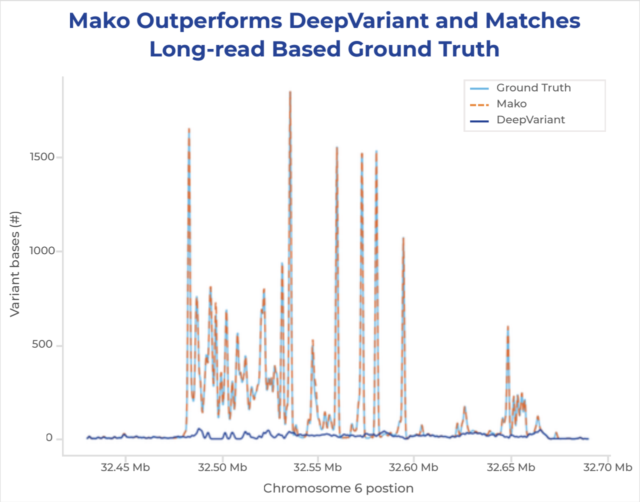
Haplotype Reconstruction

Variant Calling & HLA Typing

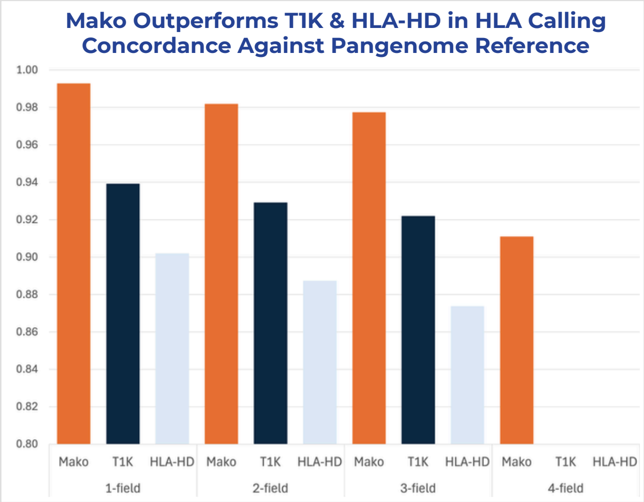
HLA-DQB1
05:03:01:01
05:01:01:01

Performance that Matters

In benchmark comparisons across the MHC, Mako outperformed standard short-read analysis with stronger variant calling performance in complex immune regions and accuracy of HLA allele calling.


Mako Outperforms DeepVariant and Matches Long-read Based Ground Truth

Mako variant calling performance compared to DeepVariant and ground truth from custom long-read sequencing in the MHC delta block


Mako Outperforms T1K & HLA-HD in HLA Calling Concordance Against Pangenome Reference

Mako HLA calling concordance compared to T1K and HLA-HD in 11 medically relevant HLA genes (HLA-A, B, C, DPA1, DPB1, DQA1, DQB1, DRB1, DRB3, DRB4, DRB5)

Broader MHC Completeness

Many tools focus on a narrower set of classical HLA genes. Mako is designed for the extended MHC, where variation in non-classical immune genes, regulatory regions, and structurally complex loci can also influence immune phenotypes and disease associations.

For research teams working in immunology, translational genomics, or population-scale studies, that broader completeness can mean clearer signals and more usable downstream results.

	HLA Typing	Novel Allele Discovery	Number of Genes	Allele Resolution	Variant Calling	Phased Haplotypes
Mako	**Yes**	**Yes**	**45**	**4 - field/8- digit**	**Yes**	**Yes**
TruSight HLA	Yes	Yes	11	4 - field/8- digit	No	No
NGSgo -AmpX	Yes	Yes	12	4 - field/8- digit	No	No
Dragen HLA	Yes	No	41	3-field/6-digit	No	No
OptiType	Yes	No	3	2-field/4-digit	No	No
HLA-HD	Yes	No	29	3-field/6-digit	No	No
T1K	Yes	No	41	3-field/6-digit	No	No
DeepVariant	No	No	-	-	Yes	No

See What's Hiding in Your Data With a Mako Pilot

Evaluate Mako on an initial set of samples and compare results against your current workflow.

**Tailored deployment options for biopharma, biobanks, and genomics service providers available*

Benthic Genomics High-confidence analysis for complex genomic regions
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