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modality analysis software release notes v1.0.0b1 (beta), June 2025

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## modality analysis software v1.0.0b1 (beta)

We are excited to announce the initial beta release of the modality CLI — a robust, scalable command-line toolkit for comprehensive DNA methylation analysis from biomodal 5-base and 6-base genomes.

This release provides researchers with a modular, reproducible workflow for processing, exploring, and interpreting large-scale epigenetic datasets, supporting both discovery and translational research.

### **Overview of Core Functionalities**

### End-to-End Methylation Analysis

Modality enables users to perform quality control, feature extraction, differential methylation analysis, and visualization in a streamlined, modular workflow.

## Flexible Data Handling

Zarr stores are used as the primary data format, supporting efficient, chunked storage and rapid access to large datasets. Utilities are provided for inspecting, renaming, and joining Zarr stores.

## Comprehensive Quality Control

Generate detailed HTML reports with sample correlation heatmaps, PCA plots, and Zarr summaries to assess data quality and experimental design.

### Feature Extraction

Extract methylation statistics (counts, sums, means, regional fractions) for user-defined genomic regions or fixed windows, supporting both hypothesis-driven and exploratory analyses.

## Differential Methylation Analysis

Identify differentially methylated regions (DMRs) between groups, with support for both standard window-based and annotated region analyses. Flexible filtering and overdispersion correction options are available.

## Interactive Visualisation

Create publication-ready volcano plots, summary tables, and customisable genomic track plots that integrate methylation data, DMRs, and gene annotations for biological interpretation.

### **Export and Interoperability**

Export results in standard formats (BED, BedGraph, Bismark, CX-report, Quant) for downstream analysis in other bioinformatics tools.

## Reproducibility & Provenance

All analyses are logged with full provenance metadata, including JSON sidecar files and metadata in output headers, ensuring traceability and compliance with best scientific practices.

### **User-Friendly Design**

The CLI provides clear error messages, detailed logging, and extensive documentation, including

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example workflows and troubleshooting guides, to support both bioinformaticians and bench scientists.

## **CLI Entrypoints**

The following CLI entrypoints are supported in this software version.

## biological-qc

Assess data quality with summary tables, correlation heatmaps, PCA plots, and Zarr summaries.

#### get

Extract methylation statistics for specified genomic regions or windows, supporting both single-base and regional analyses.

### dmr call

Identify differentially methylated regions (DMRs) between groups, with options for region-based or window-based analysis, overdispersion correction, and flexible filtering.

### dmr plot

Visualise DMR results with interactive volcano plots, summary tables, and HTML reports to guide iterative analysis.

#### tracks

Generate customizable genomic track plots, integrating methylation data, DMRs, and gene annotations for biological interpretation.

### export

Export methylation data and analysis results in community-supported formats for use in external tools.

### zarr-utils

Manage Zarr stores: inspect sample IDs, rename samples (with safety checks), generate mapping templates, and join multiple stores for integrated analysis.

### beta testing

This beta version of modality is intended for early access evaluation. We welcome your feedback as we continue to improve modality. Please report any issues or suggestions via the support channels provided in the user documentation or by e-mail to support@biomodal.com

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