JMP® Genomics

Discover the biological patterns in genomics data. From the two most trusted names in analytic software: SAS and JMP.

What is JMP® Genomics?

JMP Genomics is statistical discovery software from the two most trusted names in analytic software: SAS and JMP. Research organizations use JMP Genomics to uncover meaningful patterns in high-throughput genetics, expression, copy number and proteomics data. Dynamically interactive graphics make it easy to explore data relationships using a comprehensive set of traditional and advanced statistical algorithms.

Why is it important?

Research organizations want to maximize their return on investment of the time, money and resources required to generate high-quality genomic data sets. Specialized statistical analyses can help identify the "nuggets of gold" hidden in long lists of candidate genes or biomarkers. Whether it's used to identify potential drug targets, to explore the biology of a model organism or to develop a predictive disease model, JMP Genomics helps researchers gain a competitive advantage by quickly identifying key genes or proteins.

Who should use it?

JMP Genomics is designed for biologists, biostatisticians, statistical geneticists and students engaged in analyzing the vast stores of data that are common in genomic research. It delivers a comprehensive set of analysis methods in a single desktop software package. Adopting new software across a large organization can be challenging. That's why JMP Genomics offers analytics for many data types in the same package, making it easy for you to move into new areas of genomics as the scope of your research expands.

jmp.com/lifesciences-resources

JMP® Genomics software from SAS provides an all-in-one package for genomic data exploration and analysis. Its unique pedigree integrates the full power of the JMP statistical discovery platform with industry-leading SAS® Analytics tailored for large data sets.

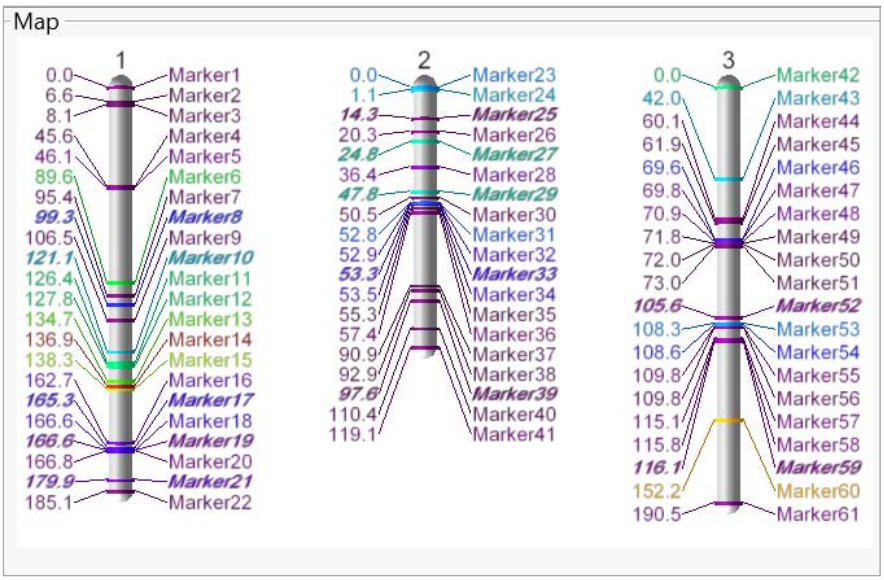
This software solution is used by biologists, biostatisticians and statistical geneticists around the world to analyze continuous intensities, counts or genotypes generated from traditional microarray platforms and summarized from next-generation technologies.

The visual paradigm of JMP Genomics dynamically links statistics with graphics to provide a detailed picture of analysis results. Even students new to genomic analysis quickly begin to discover important trends and outliers in their data, thanks to simplified dialogs and customized workflows that eliminate the need for programming skills or advanced statistical training. That's why a growing number of professors are teaching with JMP Genomics.

> "I think people are starved for software with this level of statistical power and flexibility."
>
> **Erik Sulman, MD, PhD**
> Cancer Researcher, Houston

Just point, click and you're on your way.

JMP Genomics brings the power of SAS to the study of genomics, whether you're screening for significant associations, looking for meaningful patterns from expression studies or assessing copy number differences. As your



Visualize linkage maps created in JMP Genomics or imported from other software.

inquiries expand to new areas, you can explore new data in a familiar environment – without wasting time and money learning multiple software packages and manipulating data sets to move between them.

Beyond its rich library of prebuilt graphics, JMP Genomics includes full access to the extensive analysis and graphical features offered by the JMP platform. You can design experiments that are large yet efficient, and construct a variety of dynamically interactive graphics. Features like the drag-and-drop Graph Builder and interactive Data Filter provide the flexibility for all users to create customized views of their data.

With the JMP Genomics Starter, a customizable home window, new and existing users can quickly access the tools that fit their analytic needs. The JMP Genomics Wizard guides you through the import of sample information and data sets from popular genomics data platforms and text formats. Graphics and follow-up analysis options are organized into tabbed reports, with underlying tables hidden to simplify the presentation of complex analysis results. Easily recall the hidden tables to view details, or close all tables and graphics with a single click.

Expression

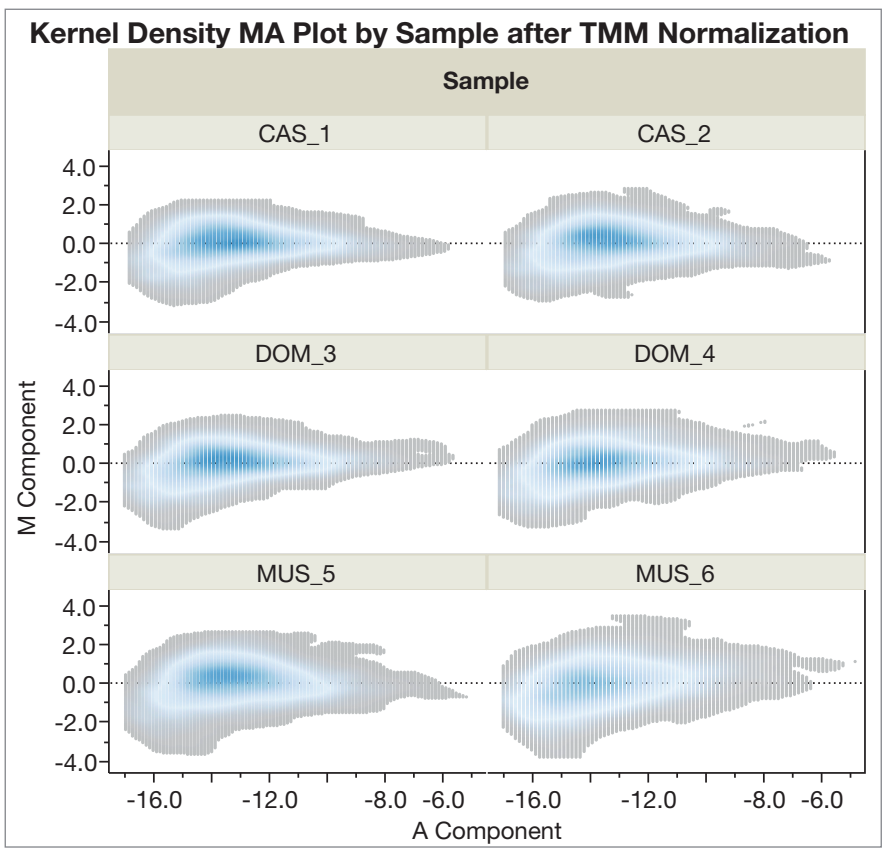
Easy-to-use basic and intermediate workflows simplify analysis of expression intensities and read counts from RNA-seq studies. With filtering, normalization, modeling and pattern discovery methods powered by SAS Analytics, you can process data sets much larger than your available memory would permit. JMP Genomics 6 offers a range of normalization and modeling options for use with microarray expression and raw or transformed count data.

Genetics

JMP Genomics provides exceptional tools for statistical geneticists, from simple case-control association and linkage disequilibrium analyses to complex association tests that support various trait types. Examine associations between SNPs and multiple continuous traits, correct for population structure, and explore SNP-SNP interactions. Point-and-click workflows simplify Q-K mixed model analysis and association analysis of rare and common variants.

Linkage maps and breeding analysis

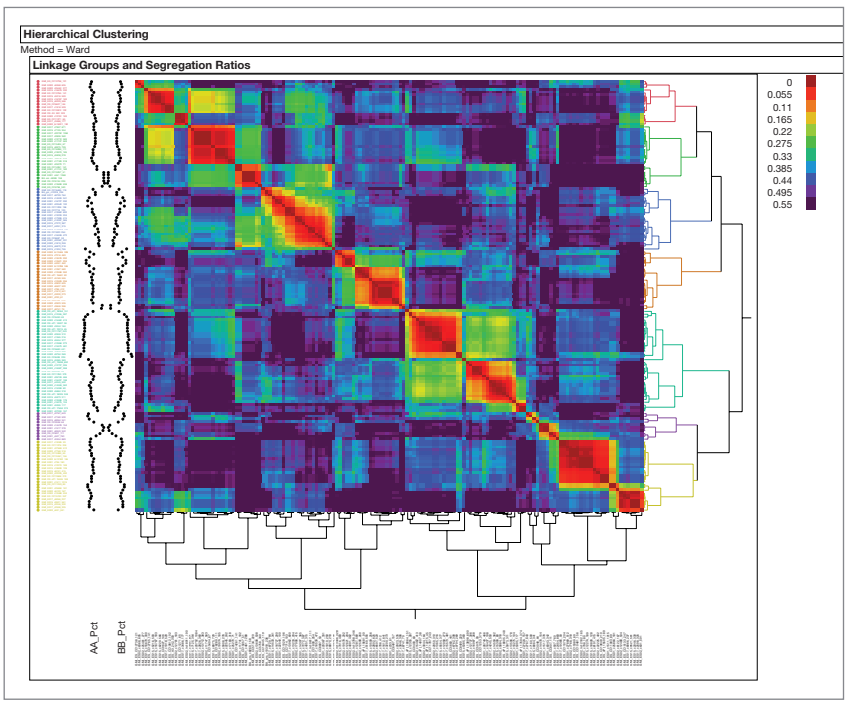
JMP Genomics features a suite of processes for the construction, optimization and visualization of marker linkage maps used to improve agronomic crops or guide animal breeding efforts. Identify linkage groups automatically or interactively, with options to designate consensus groups and framework



Scale count data across samples using TMM normalization, compare TMM factors between samples, and view kernel density plots of normalized data.

“ We take the graphical features for granted. But to be able to visualize that separation [in high-dimensional data] is so wonderful. Important differences just pop right out.”

Faye Schilkey
National Center for Genome Resources



JMP Genomics offers automated and interactive options for linkage group identification and marker ordering.

markers during order optimization. Visualize and filter marker maps using simple interactive graphics or create high quality multichromosome views. You can also summarize phenotype information and explore genotype-environment interaction analysis in multienvironment trials and perform QTL single marker or IM/CIM analysis with permutation option.

Copy number

Easily explore copy number differences between groups or within individuals. Assess data quality at the sample level to identify potential outliers and filter features based on statistical criteria. Additionally, you can adjust copy number or LOH data using paired or grouped reference samples. Use circular binary segmentation (CBS) to identify breakpoints or ANOVA-based methods to find significant differences across groups or relative to a reference group. Interactive graphics help pinpoint shared regions of variation.

Predictive modeling

JMP Genomics provides eight predictive modeling methods, with integrated predictor filtering, predictor lock-in and cross-validation. The software can be used to identify the most significant biomarkers from data sets containing multiple data types – SNP, expression, copy number. Replication and iteration strategies implemented in the software seek to reduce bias, with honest cross-validation approaches that can simultaneously assess the relative performance of hundreds of different models.

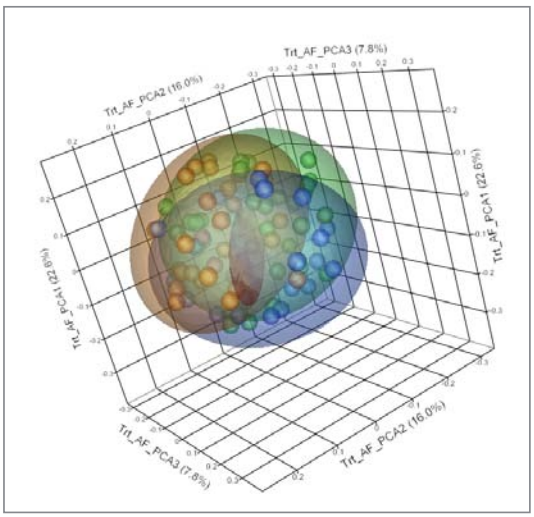
Next-generation sequencing

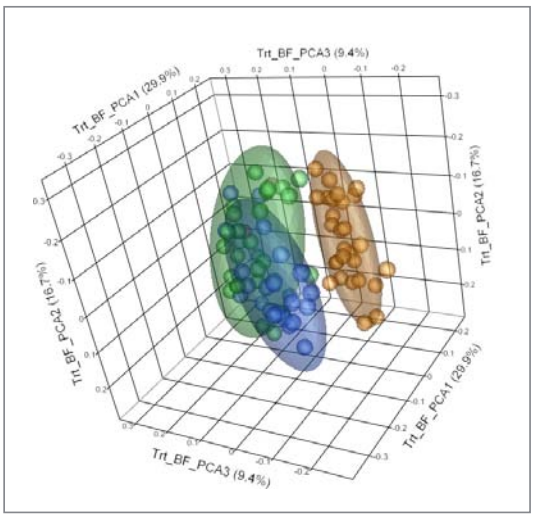
JMP Genomics provides sophisticated downstream statistical analysis capabilities for summary data from state-of-the-art sequence analysis pipelines. Import counts from text formats or summarize counts from SAM, BAM and Eland input files to take advantage of normalization and generalized linear modeling methods. Basic workflows streamline the steps in the analysis process.

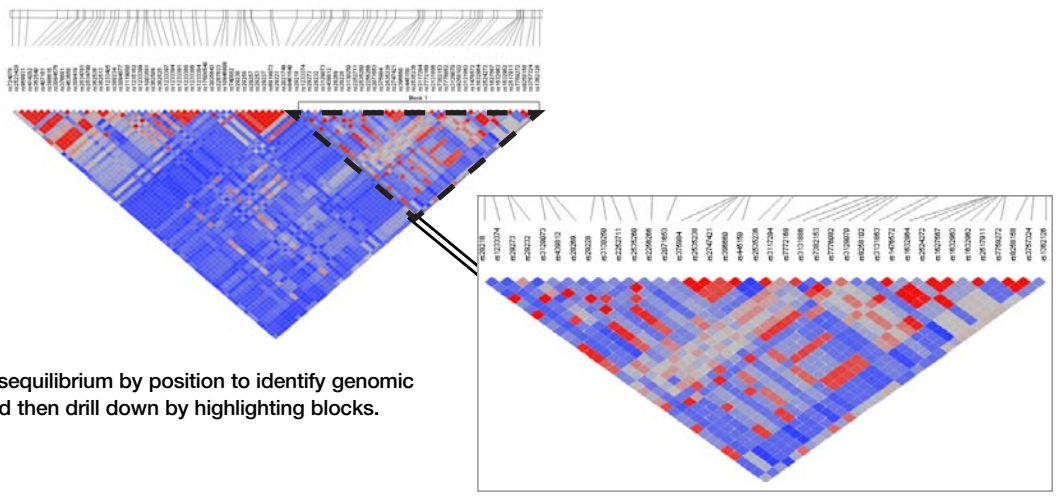
You can import genotypes directly from a variety of text formats or VCF files, or elect to call variants from BAM files using a reference genome. Perform association analysis for rare and common SNP variants or identify regions identical by state (IBS) between related or unrelated individuals.

Take advantage of the rich information provided by sequencing experiments to screen for significant correlations between different data types. View counts or statistical analysis results in the JMP Genomics Browser, and overlay histogram and heat plot tracks with individual- or group-level summaries to complement known SNP and gene tracks.

See batch effects in your data and remove them prior to statistical analysis. JMP Genomics offers several different options for batch normalization. At left, samples collected in different batches group closely together, outweighing treatment effects. At right, the same samples are shown after batch effect removal.



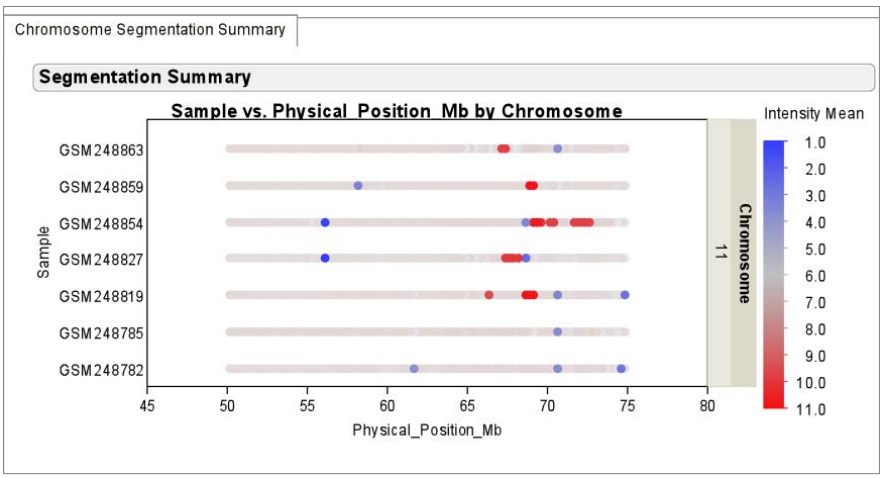




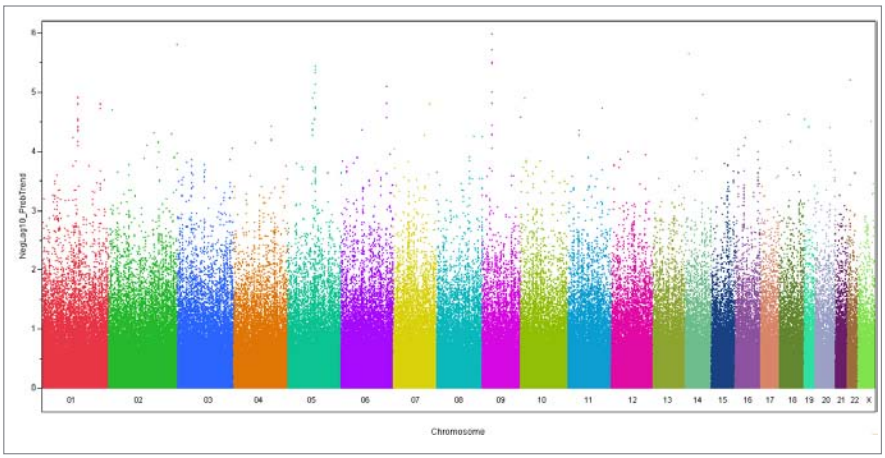
Examine patterns of linkage disequilibrium by position to identify genomic regions of greatest interest, and then drill down by highlighting blocks.

"When you're going from looking at 10 genes to looking at thousands of genes, making biological sense of the results isn't easy – it's impossible to do if you don't have the tools that help you easily visualize and explore the annotation of the results. JMP is great for that."

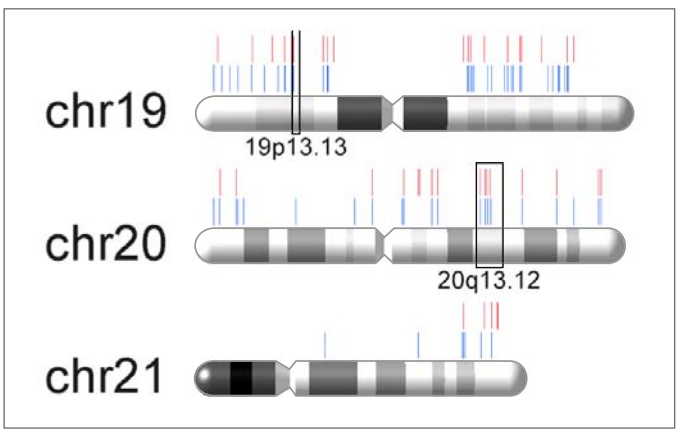
Tom Juenger, PhD
University of Texas, Austin



Segmentation summary plots can be filtered interactively to identify shared regions of copy number loss or gain.



View p-values from statistical tests individually by chromosome, or create custom, multichromosome views.



Create custom genome color themes and overlay statistical results, then zoom and drill down to visualize gene and SNP tracks.

Key Features in JMP® Genomics 6.0

JMP Genomics imports data from a variety of formats, including:

- Read counts, intensities or genotypes in single or multiple text files.
- Aligned reads in SAM or BAM format and variants in VCF files.
- Complete Genomics pipeline and *testvariant* output files.
- CLC Bio SNP and indel summary files.
- Illumina BeadStudio or GenomeStudio expression, SNP, copy number and other data types.
- A variety of Affymetrix CEL and CHP files, as well as BAR, LOHCHP, CNCHP, CNAT, and Cytogenetics CEL and CHP files.
- GenePix, QuantArray, one-color and two-color Agilent files.
- Excel and comma-separated files, including data formats from multiple NimbleGen platforms.

Flexible workflows for new and experienced users:

- JMP Genomics Wizard guides import of new data sets.
- Basic workflows for expression, exon, RNA-seq, copy number and linkage map construction.
- Intermediate options for expression QC and analysis.
- Q-K and rare variant association workflows.
- Workflow Builder for creation of custom workflows.

Integrate statistics into next-gen sequencing workflows to:

- Normalize and analyze sequence counts generated at the exon, transcript, or gene level by various pipelines.
- Assess RNA-seq data using point and click workflows.
- Test for association between traits and rare or common variants.
- Perform cross-correlation analysis to relate sequence counts to other numeric genomic measures.

Assess genome-wide variant data sets to:

- Examine individual and marker missing data patterns.
- Summarize marker properties including allele and genotype frequencies, HWE, heterozygosity and diversity.
- Filter data sets by marker or sample attributes.
- Explore associations with one or more binary or quantitative traits.
- Calculate interactions and adjust for covariates.
- Test for associations using imputed SNP data.
- Calculate and visualize linkage disequilibrium and LD blocks.
- Visualize and adjust for population structure using PCA or MDS.
- Perform GWAS meta-analysis using p-values or effects.

Expand analysis options for marker data to incorporate:

- Testing SNP variants grouped within a locus or pathway.
- Rare variant association tests with permutation options.
- Genetic distance matrices for individuals or populations.
- Calculation of IBD, IBS and allele-sharing relationship matrices.
- Compression of K matrices to save computational time.
- Association test corrections for relatedness and structure.
- Identification of genomic regions shared identical by state.
- Estimation of haplotypes and haplotype-trait association.
- Selection of haplotype tagSNPs or LD tagSNPs.
- Reconciliation of strand differences between studies.

Improve crop and livestock breeding strategies by:

- Visualizing categorical and continuous phenotype distributions among individuals, genotypes or lines.
- Assessing genotypes in multienvironment trials.
- Identifying linkage groups from experimental cross data.
- Ordering markers within linkage groups using advanced optimization algorithms.
- Visualizing linkage maps created in JMP Genomics and other mapping software packages.
- Performing single-marker, interval and composite-interval QTL mapping with permutation option.

Assess the quality of large expression data sets to:

- Identify data quality issues and remove outliers.
- Pinpoint factors that explain variance in your data.
- Visualize distributions, PCA plots and sample clusters.
- Normalize across samples.
- Remove batch effects due to technical variables.
- Adjust count distributions with TMM and KDMM.
- Perform loess, quantile, RMA, GCRMA, and ANOVA normalization or standardize to a variety of statistics.

Apply trusted statistical modeling methods to:

- Discover significant differences using ANOVA and generalized linear models.
- Apply a variety of multiple test adjustments.
- Adjust for covariates and random effects.
- Screen for allele-specific expression.
- Analyze censored survival data.
- Generate expression profiles by sample or group with dynamic selection and filtering.
- Perform hierarchical and K-means clustering.

Use advanced predictive modeling analysis tools to allow:

- Identification of biomarkers from large wide data sets.
- Selection of predictors from multiple data types.
- Customized predictor filtering during model construction.
- Lock in of key class or continuous predictors.
- Performance comparison across eight different methods.
- Cross-validation with many hold-out and iteration options.
- Learning Curve analysis to assess sample size impact.

Assess copy number data sets to:

- Examine data quality with PCA and distribution analysis.
- Adjust copy number measures using paired or grouped controls.
- View segments detected by circular binary segmentation (CBS).
- Visualize shared patterns of copy number loss or gain.
- Find genomic areas that display statistically significant differences between groups, or individuals and a control group.

(continued)

Key Features in JMP® Genomics 6.0, cont.

Use JMP Genomics annotation tools to:

- Merge functional information with statistical results.
- Upload results to Ingenuity Pathways Analysis to seek points of interaction between SNP, gene and protein lists and color pathways.
- Perform enrichment analysis using functional information from Ingenuity Pathways Analysis.
- Merge pathway information from mSigDB, KEGG or other sources to perform enrichment analysis or gene set scoring.
- Visualize sets of co-regulated genes in KEGG pathways.
- Download annotation and library files from Affymetrix NetAffx.
- Compare list membership for up to five groups and display overlaps with Venn diagrams using common gene identifiers.

Create genome-level views that allow you to:

- Visualize chromosomes with customizable color themes.
- Compare multiple experiments to find regions of shared significance.
- Overlay gene, SNP, bar chart, and color map tracks on results.
- Use a variety of continuous measures for p-value summarization.
- Drill down on interesting regions to plot p-values and view gene, SNP, bar chart and color map tracks.

The JMP software platform provides:

- Graph Builder for visual exploration of data patterns.
- Point-and-click creation of a variety of custom graphics.
- Easy copy-and-paste into Word and PowerPoint.
- Built-in scripts for capturing and sharing findings.
- Add-in capabilities for external analytics (e.g., R, SAS).

Interactive graphics generated automatically during analysis:

- Are organized into dynamic reports linked to underlying data.
- Offer point-and-click selection and easy subset creation.
- Can be queried dynamically using the JMP Data Filter.
- Can be converted to static reports.

JMP® Academic Suite

SAS is proud to partner with colleges and universities to hone the statistical thinking skills that help prepare students for an increasingly competitive and knowledge-based global economy. Academic discounts on departmental and campuswide licenses of JMP® software support the use of technology in classrooms, labs, teaching and research settings. If your institution already licenses the JMP Academic Suite, you can add JMP Genomics or its sister product, JMP Clinical, at a further discount.

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Complimentary webcasts

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Operating systems guidelines

JMP Genomics is supported on most 32- and 64-bit business versions of Windows XP, Vista and Windows 7 desktop and server operating systems.

JMP® Genomics becomes a part of everyday studies

JMP Genomics has been used for years in academic research labs. Now, this dynamic and interactive software from SAS is found increasingly in campus classrooms as well.

Susan Singer, the Laurence McKinley Gould Professor of Natural Sciences at Carleton College in Northfield, MN, attributes this development to a “huge paradigm shift” in the study of genomics. Students are now being challenged to “take advantage of the scale of data that’s available,” she explains.

Genome-scale studies require a capacity for seeing the big picture, then drilling down for details. Combining interactive JMP graphics and robust SAS® Analytics on the desktop, JMP Genomics enables students to do just that.

Read more: jmp.com/jmpgoncampus

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JMP is a software solution from SAS. To learn more about SAS, visit: **sas.com**

For JMP sales in the US and Canada, call 877 594 6567 or go to **jmp.com**