

# GWAS 2.0 using BC|SNPmax

## Entering the era of Genome-Wide Association Study 2.0

When using reference data from the HapMap or 1000 Genomes Project<sup>1</sup>, it is possible to accurately impute<sup>2</sup> very large number of new SNPs for each subject. These new SNPs may be better surrogates than SNPs in the chip, or even causal variants, giving superior results in association analysis. Some experts even recommend re-analyzing all the performed genome-wide association studies with this new method.

### Benefits of imputing

In addition to the use of imputing to improve association results, imputation tools can be used when combining data from different genotyping platforms and chips. This dramatically increases the power of meta-analyses. Imputation can also be applied to candidate gene studies with dense fine mapping data.

### Challenges in data management and analysis workflows

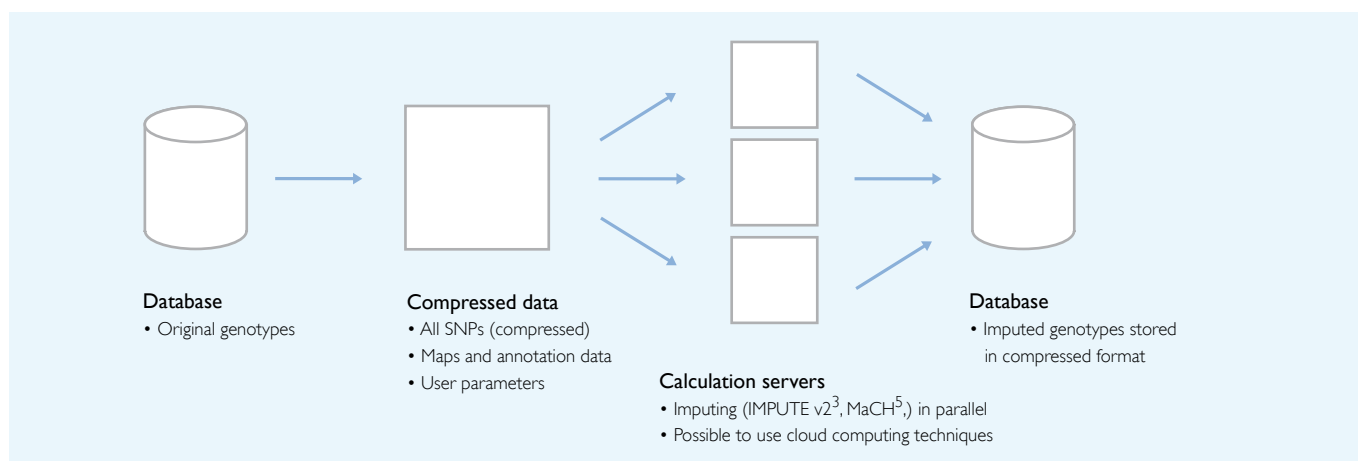
Entering the era of GWAS 2.0 presents new challenges to many research groups:

- Analysis and management of these large datasets and imputation itself requires a considerable amount of computing power and storage
- Management of required reference and annotation files is often challenging
- Data format conversions and solving possible strand problems is often complex

### GWAS 2.0 workflow using BC|SNPmax

The new generation of our BC|SNPmax and BC|GENE products offer full support for performing GWAS 2.0, ie for genome-wide association studies using imputation methods, including the following features:

- Required reference and annotation data provided as preloaded datasets in the database in the correct format, ready for use
- Support for IMPUTE v2<sup>3</sup>, SNPtest<sup>4</sup>, MaCH<sup>5</sup>, MaCH2QTL<sup>6</sup>, PLINK<sup>7</sup>, Eigenstrat<sup>8</sup> and ProbABEL<sup>9</sup> with automated data conversions and graphical user interfaces
- Support for high-performance, parallel data analysis using available calculation resources
- Amazon EC2 cloud computing technology supported, making it possible to rent calculation capacity on demand
- Training, GWAS 2.0 step-by-step manual and full professional support provided to our customers



Parallel imputing using the BC|SNPmax solution

## About BC|SNPmax Research Database Solution

BC|SNPmax is a database solution for the management and analysis of large amounts of SNP genotyping data, clinical phenotype data and CNV data. Additionally, SNP quality information, marker maps, pedigrees and analysis results can be stored in the database. The BC|SNPmax system offers graphical user interfaces for analysis programs and automated data conversion tools, creating a flexible workflow for high-performance, parallel data analysis.

The database can be set up to provide secure remote access, creating an ideal research environment for collaborative networks and multi-center research projects.

## Additional information

For more information, please visit [www.bcplatforms.com](http://www.bcplatforms.com) or contact us by email at [info@bcplatforms.com](mailto:info@bcplatforms.com)

BC Platforms is an IBM® Advanced Business Partner™, an Affymetrix GeneChip-compatible™ company and belongs to the Illumina® Connect™ partnership program.

### References:

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