

Genome Wide Association Study Graphical Interfacing for KBase System

Concept

Our app will use the genome information to conduct a genome-wide association study or GWAS which is an observational study of the genetic variants in an individual in comparison to a population to determine what traits are associated with a genetic variant.

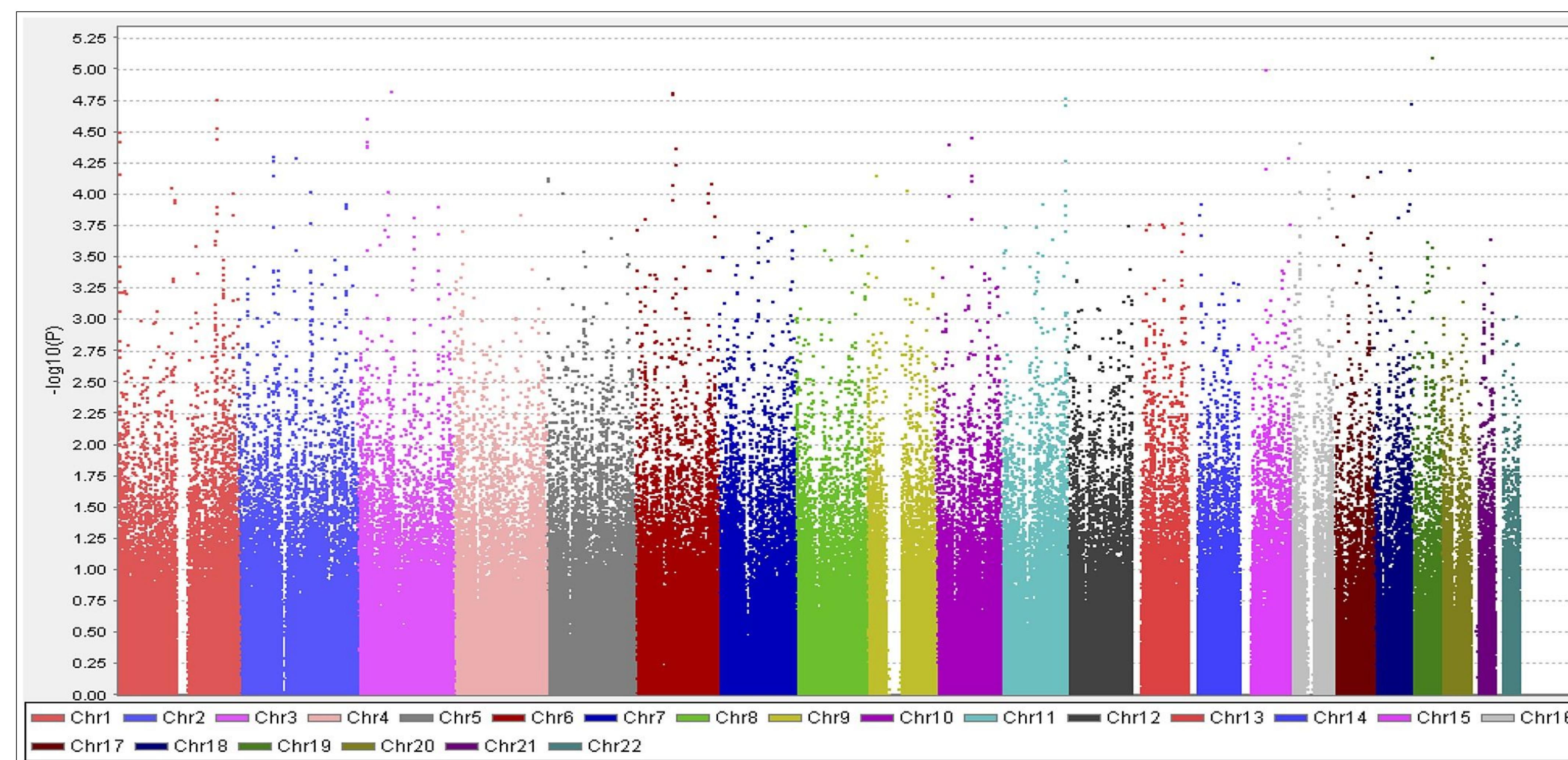
We are creating a graphical interface for data input and updates from the running processes. Lastly, we will use graphics to display the results in relevant charts (i.e. Manhattan plots) and tables.

Elevator Statement

For researchers who need easy to access tools for genome analysis, the GWAS KBase App is a tool that provides a graphical interface to a cloud-based bioinformatic infrastructure. Unlike other GWAS apps or doing the computations by hand, our app uses cloud computing to offer substantial time savings, gives users access to research data, and collects previously computed results for access in a knowledge base.

What is a GWAS?

- A GWAS compares the frequencies of genetic variation in individuals with a given trait and in control individuals from the same population.
- What is genetic variation? Genetic variation refers to the differences in genetic material both within and among populations. One type of genetic variation is the single nucleotide polymorphism (SNP), which is a variation at a single position in a DNA sequence among individuals.
- In a GWAS, researchers examine the relationship between every DNA position and a particular trait (such as diabetes) using the signal from each position as an indicator for the DNA sequence that surrounds it. A strong "association" between a DNA position and a particular trait marks the general location of the associated genetic alteration, even if the associated SNP itself is not directly responsible for the trait.



Manhattan plot for the GWAS of diabetic retinopathy in stage 1. P values of the Fisher's exact test under the allele model for SNPs in 446 patients were shown.

Awata, T., Yamashita, H., Kurihara, S., Morita-Ohkubo, T., Miyashita, Y., Katayama, S., Mori, K., Yoneya, S., Kohda, M., Okazaki, Y., Maruyama, T., Shimada, A., Yasuda, K., Nishida, N., Tokunaga, K., ... Koike, A. (2014). A genome-wide association study for diabetic retinopathy in a Japanese population: potential association with a long intergenic non-coding RNA. *PLoS one*, 9(11), e111715. doi:10.1371/journal.pone.0111715

Sponsor

Our sponsors for this project are the US Department of Energy, Oak Ridge National Laboratory, and the University of Tennessee, Knoxville. They are seeking a graphical interface for sharing hypotheses in order to expand current knowledge of genome associations and provide visual support to large sets of data. This will allow researchers using the system to identify other research along the same vein of the topic they are exploring and further connect previously unexplored avenues for further research.

Our Project

- Integration into the KBase system
 - Open source
 - Uses cloud computing
- Makes your research reproducible, accessible and reusable.

