

TRANSFORMING COMPLEX DATA INTO CLINICAL ANSWERS

THE GENE BEHIND THE DISEASE



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Case/Control and cohort studies are key tools for finding associations between specific variants or genes, and phenotypes or particular diseases.

Traditionally, analyzing such data requires a high number of samples, significant storage capacity and computational resources, data streaming, analysis parallelization, and hundreds to thousands of multi-core processors. Until now, the need for deep knowledge of advanced statistical modeling, extensive computational resources, together will the associated costs have made analysis on a routine basis nearly impossible.

Genoox has now developed cutting-edge case/control and cohort analysis capabilities designed to simplify these complex issues. Utilizing a powerful statistical engine that examines correlations between different groups allows its users to easily compare between multiple patient groups, case vs. control, enabling complex drug response analysis.

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The Analysis Process

Teva performed Whole Exome Analysis for hundreds of patients and uploaded the data to the Genoox platform. All sample data was uploaded to the cloud and interrogated using a multi-faceted variant calling process that employs proprietary and sensitive statistical models. This allowed for extremely high variant detection sensitivity, providing an exceptional level of confidence.

During the analysis process, Teva Pharmaceuticals included both variants in genes known to be related to the studied phenotype and more broadly, of variants in genes within similar pathways which contribute to the mechanism of pain related disorders. The analysis was performed using the Genoox interpretation engine, crawling through hundreds of sources both structured and unstructured to reveal different layers and probable medical relations.

The Genoox Advantage

The Genoox platform makes use of a sensitive statistical model providing superior bioinformatics by utilizing proprietary data structure which allows its users to detect variants with better accuracy. This is in part due to the use of historical data. The advanced algorithms allow for extremely high variant detection sensitivity, affording an exceptional level of confidence with variant calls as well as a significant reduction in sequencing costs.

Genoox offers multi-sample analyses at different levels by supporting advanced statistical models , from a standard chi-square test to advanced tests including Linear Regression Logistic regression and Linear Mixed Models (LMM), Multivariate analysis (e.g. at the gene level) was performed using different statistical tests e.g. burden tests, CAST and SKAT.

The platform relies on a robust computational infrastructure which easily scales to tens of thousands of samples, relieving the user of the need to deal with the significant costs and other resources required when usually performing these types of analyses.