



GENOMENON

For Immediate Release

Genomenon Delivers Evidence-Based Gene Panel Designs

New Rational Panel Design Service Delivers Candidate Genes and Variants Culled from Medical Literature

Ann Arbor, Mich - March 21, 2017 - Genomenon announces the launch of Mastermind Panel Design Service, a novel approach to finding genes and variants associated with a particular disease in the medical literature. Genomenon uses its Mastermind literature-driven knowledge-base to deliver a candidate gene list for any disease supported by medical literature citations and customized around customers' specific requirements.

"Mastermind Panel Design was born from customer demand" said Mike Klein, CEO of Genomenon. "Labs were using Mastermind to find all the disease-associated genes and were looking to further accelerate their panel design process. By mining the Mastermind database backend, we have been able to deliver a turnkey gene panel in just a matter of days."

Clinicians and researchers face a daunting challenge when developing gene panels for any particular disease. Often the panels are designed by committee and require laborious searches of scientific literature to support the genes, exons and hotspots to include on a panel. As a result, each clinical reference lab screens different gene sets for the same disease.

Looking across 55 clinical labs testing for cardiomyopathy, a total of 250 unique genes are targeted. However, of these 250 genes only 1 gene is included in every panel (*ACTC1*). Furthermore, only 60% of the panels included the top 20 most common genes and only 33% of the labs included the top 50 genes on their panels.

Genomenon provides a more scientific, reproducible method to deliver the primary evidence required to build a gene panel to screen for a specific disease. Mastermind Panel Design not only drastically cuts the time it takes to create a new panel, it provides literature citations for each and every candidate variant, gene and exon selected to go on the panel. Reference citations are important for clinical labs to provide a rationale for the diagnostic tests they are asking insurers to reimburse.

Genomenon is able to deliver this break-through approach to panel design by mining Mastermind's comprehensive database of disease-gene-variant relationships extracted from primary medical literature. Genomenon scientists provide a comprehensive list of candidate genes and variants (coding and non-coding, single nucleotide changes and small indels, amplifications/deletions and fusion events) tagged with a list of publications that associate the disease to the biomarker. The final panel can be curated from the candidate genes, exons and variants based on the associated literature.

About Genomenon:

Genomenon has eliminated the manual search process for gene and variant curation with a scientific literature database and visualization software of prioritized evidence for use in clinical decision-making. We have mined the full text of millions of genomic-related scientific articles to create a comprehensive genomic knowledge

base that enables pathologists and geneticists to quickly and accurately curate disease-causing variants from genomic-sequencing datasets. With the comprehensive data set and deep insight into all of the genomic literature, we're able to provide fascinating results – from accelerated gene and variant curation to evidence-based panel design – by scouring the literature to find genes and variants linked to specific diseases within the literature. For more information, visit www.genomenon.com or email sales@genomenon.com.

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