

Gene data tool advances prospects for personalized medicine

October 9 2009

A sophisticated computational algorithm, applied to a large set of gene markers, has achieved greater accuracy than conventional methods in assessing individual risk for type 1 diabetes.

A research team led by Hakon Hakonarson, M.D., Ph.D., director of the Center for Applied Genomics at The Children's Hospital of Philadelphia, suggests that their technique, applied to appropriate complex multigenic diseases, improves the prospects for personalizing medicine to an individual's [genetic profile](#). The study appears in the October 9 issue of the online journal [PLoS Genetics](#).

Genome-wide association studies (GWAS), in which automated genotyping tools scan the entire [human genome](#) seeking gene variants that contribute to disease risk, have yet to fulfill their potential in allowing physicians to accurately predict a person's individual risk for a disease, and thus guide prevention and treatment strategies.

The authors say that for many diseases, the majority of contributory genes remain undiscovered, and studies that make selective use of a limited number of selected, validated gene variants yield very limited results. For many of the recent studies, the area under the curve (AUC), a method of measuring the accuracy of risk assessment, amounts to 0.55 to 0.60, little better than chance (0.50), and thus falling short of clinical usefulness.

Hakonarson's team broadened their net, going beyond cherry-picked

susceptibility genes to searching a broader collection of markers, including many that have not yet been confirmed, but which reach a statistical threshold for gene interactions or association with a disease. Although this approach embraces some false positives, its overall statistical power produces robust predictive results.

By applying a "machine-learning" algorithm that finds interactions among data points, say the authors, they were able to identify a large ensemble of genes that interact together. After applying their algorithm to a GWAS dataset for [type 1 diabetes](#), they generated a model and then validated that model in two independent datasets. The model was highly accurate in separating type 1 diabetes cases from control subjects, achieving AUC scores in the mid-80s.

The authors say it is crucial to choose a target disease carefully. Type 1 diabetes is known to be highly heritable, with many risk-conferring genes concentrated in one region—the major histocompatibility complex. For other complex diseases, such as psychiatric disorders, which do not have major-effect genes in concentrated locations, this approach might not be as effective.

Furthermore, the authors' risk assessment model might not be applicable to mass population-level screening, but rather could be most useful in evaluating siblings of affected patients, who already are known to have a higher risk for the specific disease. The authors say that their approach is more effective, and costs less, than human leukocyte antigen (HLA) testing, currently used to assess type 1 diabetes risk in clinical settings.

Source: Children's Hospital of Philadelphia ([news](#) : [web](#))

Citation: Gene data tool advances prospects for personalized medicine (2009, October 9)

retrieved 27 April 2024 from

<https://medicalxpress.com/news/2009-10-gene-tool-advances-prospects-personalized.html>

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