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Personalized Genetics: A Responsible Approach

To the Editor: A recent paper¹ in The Journal aptly described the challenges inherent in using genomic profiles to predict risk for common diseases and to develop personalized risk-prevention advice. Companies or other organizations that take a responsible approach to these challenges can potentially offer new opportunities for disease prevention, early detection, and treatment.

The authors took a sample of the loci covered by the tests of seven companies in the field and have shown that most of these loci do not pass simple quality criteria. The challenge with the authors' analysis is that it analyzes the pool of loci used by all seven companies, instead of breaking the analysis down by company or organization; hence, the approach does not distinguish between organizations that take a rigorous and responsible approach to the evaluation of risk and organizations that base the risk assessment on unreliable scientific information.

We share the authors' concerns about companies that report genetic risk based on a single association study or on studies with methodological weaknesses. However, we strongly believe that customers can benefit from a personalized report of those genetic associations found in genome-wide-association studies that were replicated in multiple populations with sound epidemiological, statistical, and laboratory practices. Many examples of reliable, replicated associations have been reported, including between transcription factor TCF7L2 (MIM 602228) and diabetes and between NOD2 (MIM 605956) and Crohn's disease.²

Taking a responsible approach means that companies utilize only high-quality association studies to bring customers accurate genetic risk predictions, as well as effective strategies for reducing risk for those genetic conditions to which they are predisposed. In addition to using rigorous and transparent scientific standards for inclusion in the testing panel, a responsible approach is to provide customers and their doctors with resources such as genetic counselors, physician expertise, and epidemiologists. By taking a responsible approach, the personalized genomics community can work together with individuals and their medical providers to enable people to live longer, healthier lives.

Dietrich Stephan, 1,2 David Agus, 1,3 Michael Nierenberg, 1 and Elana Silver^{1,*}

¹Navigenics, 1 Lagoon Drive, Suite 450, Redwood Shores, CA 94065, USA; ²Translational Genomics Research Institute, 445 N. Fifth Street, Phoenix, AZ 85004, USA; ³Spielberg Family Center for Applied Proteomics, Cedars-Sinai Medical Center, 8631 W. Third Street, Suite 215E, Los Angeles, CA 90048, USA

*Correspondence: esilver@navigenics.com

Web Resources

The URL for data presented herein is as follows:

Online Mendelian Inheritance in Man (OMIM), http:// www.ncbi.nlm.nih.gov/Omim/

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