SPECIAL ARTICLE

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Initiating a Human Variome Project Country Node

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ABSTRACT: Genetic diseases are a pressing global health problem that requires comprehensive access to basic clinical and genetic data to counter. The creation of regional and international databases that can be easily accessed by clinicians and diagnostic labs will greatly improve our ability to accurately diagnose and treat patients with genetic disorders. The Human Variome Project is currently working in conjunction with human genetics societies to achieve this by establishing systems to collect every mutation reported by a diagnostic laboratory, clinic, or research laboratory in a country and store these within a national repository, or *HVP Country Node*. Nodes have already been initiated in Australia, Belgium, China, Egypt, Malaysia, and Kuwait. Each is examining how to systematically collect and share genetic, clinical, and biochemical information in a country-specific manner that is sensitive to local ethical and cultural issues. This article gathers cases of genetic data collection within countries and takes recommendations from the global community to develop a procedure for countries wishing to establish their own collection system as part of the Human Variome Project. We hope this may lead to standard practices to facilitate global collection of data and allow efficient use in clinical practice, research and therapy. Hum Mutat 32:501–506, 2011. © 2011 Wiley-Liss, Inc.

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Background

The Human Variome Project is an international initiative committed to reducing the burden of genetic disease on the world's population by collecting and sharing data on all instances of genetic variation effecting human disease. The vision of the Human Variome Project is to be a catalyst for reduction in human disease in the 21st century by facilitating the establishment and maintenance of standards, systems, and infrastructure for the worldwide collection and sharing of genetic information.

Collecting information on all instances of human genetic variation as they are discovered is an ambitious prospect and one that has generated much discussion within the Human Variome Project Consortium. In addition to the obvious technical and organizational challenges of a data collection program of this scale, the nature of genetic variation information introduces a number of ethical, legal, and sociocultural challenges. To address these issues, the Human Variome Project Roadmap (available at http://www.humanvariomeproject.org/) proposes a two tiered method of collection via an integrated network of gene and disease specific databases and country based repositories or *HVP Country Nodes*.

HVP Country Nodes are repositories of genetic variation data that have been generated within each country hosting a Node. These repositories are built, managed, and resourced by individual countries to meet the ethical, legal, and information requirements of those individual countries. The information within each node is collected directly from laboratories and clinics within each country and stored safely and securely in a national repository. This model of collection allows diagnostic lab workers and clinicians access to a core set of information about each instance of genetic variation reported in their own country, while keeping access and storage managed at the local level. Access to such a repository will:

- provide faster and more accurate diagnosis of genetically based illnesses within the country's populations, reducing the cost and suffering of patients;
- help clinicians make more accurate prognoses and develop better treatment plans;
- improve the quality of genetic counseling for families and genetic testing; and
- improve national healthcare planning leading to reduced costs within national healthcare systems.

Ultimately, these repositories will become key sources of data for international gene and disease/specific databases by automatically passing on their data to the appropriate databases.

The HVP Country Node model provides a level of flexibility and modularization that would not be available in a centrally mandated system. As each individual country is responsible for the funding, collection, and storage of the data generated by their own country, they can ensure that it is handled according to their

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