Coordinating Office Position Paper

Mechanism for the establishment of new gene/disease specific databases under the *Country Node and Cooperation Agreement* with China

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Authorisation:
This Position Paper has been prepared by Timothy D. Smith and represents the official position of the Human Variome Project Coordinating Office only. It does not represent an official position of the Human Variome Project, its Consortium, Advisory Councils or International Scientific Advisory Committee.

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Prof. Richard Cotton
Scientific Director

I Background

Human Variome Project International Ltd. (HVPI) recently entered into a partnership agreement with the National Institute of Gene Science and Technology Industrialisation Development (China)—representing the Chinese Government. Under part of this agreement, China has agreed:

- to provide the funding, resources and personnel necessary to establish the 5,000 to 8,000 gene/disease specific databases (where none currently exist or are dormant) within ten years; and
- to provide the funding, resources and personnel necessary to contribute to the maintenance of international databases as is agreed with HVPI.

In return, HVPI has agreed:

- to facilitate and provide all necessary guidelines, protocols and support to warrant China to establish and maintain between 5,000 and 8,000 gene/disease specific databases (where none currently exist or are dormant) within ten years.

In particular, HVPI has agreed:

- to assist China in the choice of genes and/or diseases for which databases are to be established; and
- to provide access to the global networks which will enable this process to be effected.

In order to fulfill these obligations, a process must be established by the International Scientific Advisory Committee (ISAC) that will enable the International Coordinating Office (ICO) to engage with our Chinese colleagues and assist them in choosing the 5,000–8,000 databases to be established or and supported.

This position paper outlines the ICO's position on what such a process might look like.

II Database Selection Sub-Committee

A dedicated sub-committee of the ISAC should be convened to assist with the formulation and management of the entire process. Membership of the sub-committee can include non-ISAC members, but the majority of members should consist of ISAC members. It is expected that this committee will endure past the end of the terms of current ISAC members. As such, the terms of reference for the committee must take this eventuality into account.

The Database Selection Sub-Committee should have the following functions:

- conduct research to determine priority areas for database construction
- develop the parameters and specifications to which new databases should be constructed—see §III
- confirm with Chinese colleagues the level of support that can be offered to existing databases
- develop a process for soliciting and assessing requests for support from existing databases
- managing the support request process in conjunction with the ICO
- proposing a list of existing databases to support and new databases to create to the ISAC

The ICO will make staff available to assist the sub-committee with its functions, specifically research and secretarial functions.

III Parameters and Specifications

Possibly the most important function of the Database Selection Sub-Committee will be to determine the position of HVPI on what comprises a "databases" under this agreement. The agreement only specifies that 5,000 to 8,000 databases will be created or supported—no mention is made of what those databases will look like. Parameters and specifications for these new databases must be agreed with our Chinese colleagues before construction can commence to ensure that the new databases are useful and meet the needs of the Human Variome Project now and into the future.

Obviously, all databases associated with the Human Variome Project must agree to abide by the relevant Standards and Guidelines developed by the Project[1].However, the structure of the Human Variome Project places the responsibility of developing standards and guidelines for the creation and operation of gene/disease specific databases upon the Gene/Disease Specific Database Advisory Council and this Advisory Council has not yet been convened. Hence, beyond specifying that the new databases must abide by the relevant Standards and Guidelines once they are developed, the Database Selection Sub-Committee must stipulate some further parameters and specifications in the intervening period.

Without trying to perform the work of the Sub-Committee, the ICO believes that the following should be included in such specifications:

- new databases should only be created for genes/diseases that do not already have an active, curated database
- existing databases that require significant restructuring or are struggling because of lack of funding, leadership or direction should be supported
- where an inactive database is to be recreated, the new and old databases should be merged and every effort made to involve the existing curator(s)
- every new database must have at least one curator signed on who is an expert in that gene/disease
- each new database must contain a public declaration that the data it contains is available free of charge and without restriction
- each new database must be supported for a period of at least 10 years

IV Next Steps

This position paper will be forwarded to the ISAC Chair for his information. It will then be up to the ISAC to move forward with this process, or a variant thereof. The upcoming ISAC meeting in Canada in October would be an ideal place to convene the sub-committee.

The ICO will be available to the ISAC Chair or his nominee to assist in the drafting of the terms of reference for the Sub-Committee.

[1] see *Project Roadmap 2010–2012* for a definition of Standards and Guidelines