



## Coordinating Office Position Paper

# Assessing Genomic and Genetic Services in a Country: Supporting the Development of HVP Country Nodes

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Human Variome Project International Ltd.  
Level 2, 161 Barry St  
Carlton South VIC 3053  
AUSTRALIA

**Prepared by:**  
Helen Robinson

**Contact:**  
Helen Robinson  
Level 2, 161 Barry Street  
Carlton South VIC 3053  
AUSTRALIA  
+61 (0)3 9035 5133  
rhm@unimelb.edu.au

**Authorisation:**

This Position Paper has been prepared by Helen Robinson 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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*Heather Howard*

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## I Purpose - what are we trying to do here?

The aim is to come up with an agreed definition of what is a well-functioning country node and how it connects with other related services both nationally and internationally. Developing such a definition provides a 'standard' of performance and can assist in identifying the key areas to help develop and strengthen the performance of individual nodes. As part of an international network, individual country nodes contribute to open and transparent data sharing. If a definition of a well-functioning country node can be developed that almost all Human Variome Project Consortium members can agree on, it can also help achieve several other related objectives:

1. Promote discussion among members on what is a 'well-functioning' country node - bring into the open areas of agreement and disagreement
2. Devise a common language and agreed terms to describe the functions of a country node
3. Bring the membership together on an agreed and acceptable standard
4. Help to set standards for education, ethical practice, dealing with issues of consent
5. Promote knowledge sharing across membership to support the development and growth of new nodes in a more focussed way

## II Diversity not standardisation

We all know that nodes operate in different settings, environments and so on - one size does not fit all; diversity is good, in fact, it is a major means of improving performance because it helps us look critically and constructively at other ways of working and reflect on our own operations. But at the same time, if all nodes are to be networked to share data effectively, then should probably be able to achieve similar outcomes in support of effective and easy global collaboration. By agreeing on the outcomes - what a well functioning country node CAN REALLY DO - we can leave the process to be determined at the country level in accordance with local needs.

## III How would you start to define a well-functioning country node?

The attached 5 X 5 matrix makes a first attempt at setting out a matrix for assessing the genomic and genetic services in a country and the role of the country node in this - as a basis for discussion.

### *Five Rows*

The rows are the various elements or aspects that go to make up a fully functional Country Node. These five align with the main activities of HVP. They are:

- A Data collecting and sharing
- B Having the necessary resources to deliver cost-effective and equitable services - human, technical, financial
- C Ethical, legal and social issues - having the necessary regulatory frameworks in place
- D Planning for service delivery in context of public health is linked
- E Degree of connectedness with others in the community - health and non-health, national and international

### *5 Columns*

The columns represent stages in development and functioning of Country Nodes; they seek to define or describe benchmarks or stages in at various points in the progressive development of country nodes. This progressive development is a continuum from the early stages of awareness that establishing a country node would be a "good idea" to have a fully functioning, sustainable and competent Country Node. After defining each of these "end points", it is then a matter of determining points in the middle - in this case 3 points, making a total of 5 points or stages in all:

1. STAGE 1 - getting ready, getting started or being a informal group with similar objectives
2. STAGE 2 - moving to a more formalised way of working - we know we need to do something, and we are in the process of determining/agreeing our plan; key players are being identified
3. STAGE 3 - some idea of what is needed exists and those formally involved are doing what they can, given the resources available; there are some signs that progress is being made and that these successes will help align future efforts; key players are coming 'on board'
4. STAGE 4 - what needs to be done to formally defined and agreed; there is a plan and demonstrable progress is being made; key players are aligned around an increasingly consistent set of activities
5. STAGE 5 - fully functional and improving in quality, and efficiency; helping others to improve

### *Indicators*

The boxes in the 5 X 5 matrix are filled with indicators. These indicators are suggestions of what you might observe if you were watching a country node functioning over a few months- the aim is to set some levels of performance, some ways of measuring performance, and so to stimulate discussion and action on improving performance. It would be possible to 'tick off' how closely a particular node was matched the ideal and then to diagnose some priority areas of activity for improving performance .

### **IV Would others agree with us and rate our Country Node in the same we that we see it?**

This depends on:

- how well we can define what a well functioning nodes does; and
- on how much agreement there is on the quality and validity of these measures; and
- how that information might be used.

For example, it could be used in:

1. Accreditation processes
2. Peer review of performance
3. Award giving, grant giving, rating process

## SUGGESTED 5 x 5 MATRIX FOR ASSESSING GENOMIC AND GENETIC SERVICES IN A COUNTRY

	<b>GETTING READY =not really on the agenda yet</b>	<b>DETERMINING THE FRAMEWORK = we know we need to do something, but not sure what</b>	<b>PUTTING PIECES IN PLACE=we have some idea of what is needed and are doing what we can now</b>	<b>MOVING FORWARD = we know what needs to be done and are getting on with achieving it</b>	<b>FULLY FUNCTIONAL = and improving</b>
<b>Data collection and sharing</b>	Data not being collected or shared	Framework for sharing data has been developed but not fully implemented	Some laboratories are sharing data	All molecular and clinical data is pooled and linked to a functional electronic health record  Some data is shared freely within country and internationally	All molecular and clinical data is pooled and linked to a functional electronic health record  Data is shared freely nationally and internationally
<b>Availability of key resources and services:</b>	Nothing really organized Services happen in an ad hoc manner Absence of any overall strategic plan Supply and demand for services are not well understood Personal contacts make things happen Funding is ad hoc and not based on present or future needs	Limited services only available in big cities, nothing in rural areas  Costs are not subsidised – user pays Few appropriate education course available  Hard to fill key positions – brain drain  Samples sent out of country for analysis	Service provision is uneven, not planned systematically  Lot of vacant positions, unskilled people doing work; work load high  Some education courses are planned, but students need to study in other countries  Routine work is done in country, more complex work sent outside	Barriers to access to are known and strategies are in place to handle them  Education and training of all groups required to full service provision is understood and plans are place Longer term plans are being discussed  Plan for providing services in place	Fully sustainable; all available jobs are filled promptly by well qualified competent people  Public and private funds are available as required; health insurance covers core services Research is funded Level of support commensurate with BOD Services accessible to poor, urban/rural on equitable basis
<ul style="list-style-type: none"> <li>● <b>Human resources, education and training of key professionals and supporting roles</b></li> <li>● <b>Financial resources – public and private</b></li> <li>● <b>Laboratory, technical support</b></li> <li>● <b>Computing data analysis and reporting support</b></li> </ul>	Aware of these needs but nothing is in place to deal with these issues in a progressive manner	Dealt with on an ad hoc basis  Some are expressing the need for frameworks and	Handled as needs arise Some limited education and training done for some groups	All issues fully integrated into services provided Information and support available	All frame works up and running and evaluated regularly  Feedback is used to
<b>Ethical, social, legal issues:</b>	<ul style="list-style-type: none"> <li>● <b>key regulatory frameworks are in place and well used</b></li> <li>● <b>patient consent</b></li> </ul>				

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<ul style="list-style-type: none"> <li>• <b>conflict of interest</b></li> </ul>	Issues are a low priority in government Issues are not well understood in community	structures for handling these issues more strategically	Aware that a longer term strategy is needed	Appeals, privacy handled well Legal and regulatory framework works in place	improve responsiveness and keep up-to-date
<p><b>Degree of integration genetic/genomics services in health care system:</b></p> <ul style="list-style-type: none"> <li>• Burden of disease is known, monitored</li> <li>• Level of understanding of genetic medicine vs genomic in health sector</li> </ul>	No/little basic research is done No link between the available resources and the needs in the national health care system	Research community is small Not part of national health plan	MOH engaged Funds go to basic research in the area, to core health services	Genetic and genomic services, requirements are part of national health plan Disease registries in place and becoming better known	Co-morbidities etc Genetics and genomics services seen as way of reducing health costs, prevention
<p><b>Level of engagement in debate/public awareness issues related to human genomics:</b></p> <ul style="list-style-type: none"> <li>• Politicians</li> <li>• Public</li> <li>• public health officials</li> <li>• consumers/patients and carers</li> </ul>	Non-existent or low priority Level of awareness is limited to a few professionals, patients and those who are directly involved in services No links to national Human Genetics Society	Poor political understanding of need MOH has some understanding Civil society advocacy weak Weak links to national Human Genetics Society	Support and understanding growing in health/medical community Patient groups are becoming aligned/on-board	Civil society organized, patient groups and consumer groups well organized Genomics is seen as a public health issue Good links to national Human Genetics and Genetics Society	At cutting edge, leading the discussions in media, scientific journals, consumers, professional groups Political commitment is high Effective leadership role in national, regional and international Genetics Societies