

Human Variome Project
Australian Node

GOVERNANCE



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TABLE OF CONTENTS

Introduction	5
The Human Variome Project	5
Introduction	5
International Project Data Flow	5
Overview	6
Overview of Australian Node Project	6
The Human Variome Project Australian Node(HVPA)	7
Management Structure.....	7
Advisory Committee.....	8
Appointment of Committee Members	8
Terms of Reference.....	8
Stakeholders /Users Committee	8
Appointment of Committee Members	8
Access Committee.....	9
Appointment of Committee Members	9
Terms of Reference.....	9

INTRODUCTION

THE HUMAN VARIOME PROJECT

INTRODUCTION

For over a decade, the complete sequence of the human genome has been freely available to every biomedical researcher with access to the internet. For over a decade, the complete sequence of the human genome has been analysed, scrutinised and tested. And for over a decade, we have been told that it is the sequence of the human genome that will provide the answers to curing all genetic illness.

However ground breaking, the Human Genome Project has only provided us with some of the answers to disease. While we now may know the complete sequence of an average individual, there is really no such thing as an average individual. And with many severe and debilitating diseases that can be caused by a single change to this sequence, we are a long way from providing useful benefits for real people using the data from the human genome project alone.

Out of the 20,000 human genes mapped by the Human Genome Project, only 3,000 have any information available on their variations causing disease and this is scattered and incomplete making the information difficult to access. It is widely accepted that in the next few years the number of genes in which disease-causing variations are recognized will increase dramatically. Currently there is no standardized or ongoing way to capture this information and make it of use to clinicians and researchers.

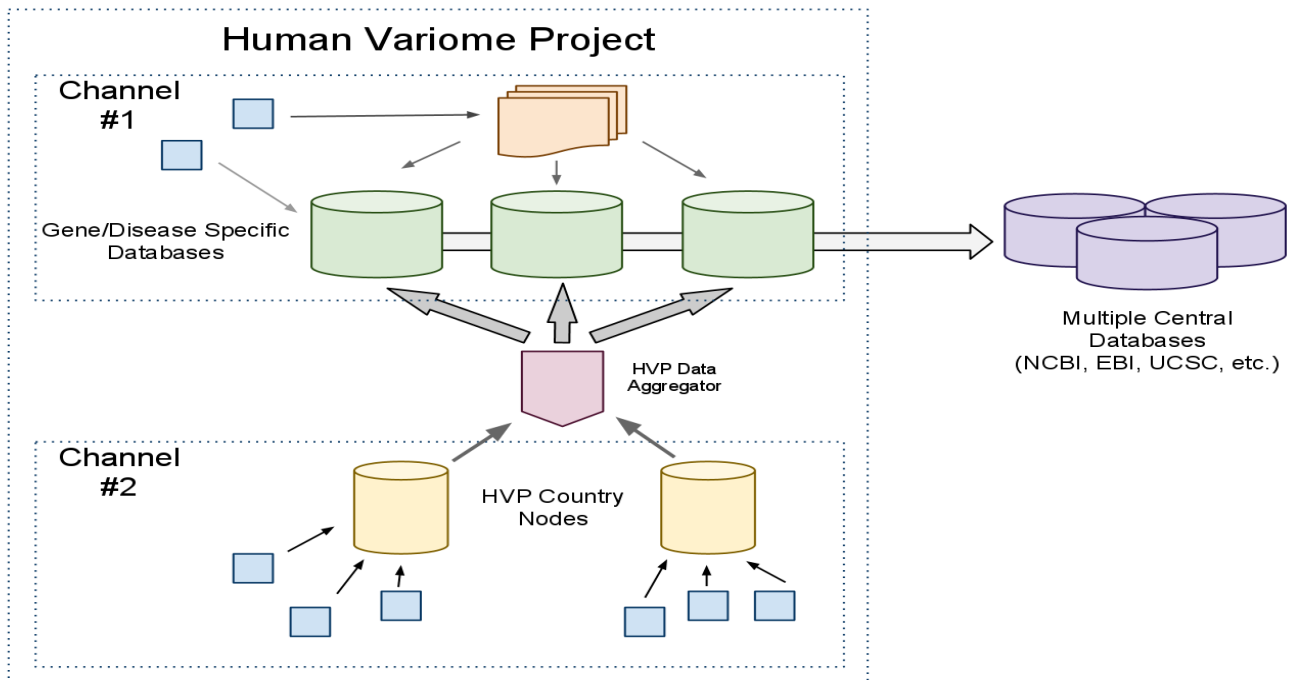
The Human Variome Project, an Australian led initiative, is seeking to provide real answers to the problem of documenting diseases caused by errors in the human genome sequence. The Human Variome Project was initiated at a WHO sponsored meeting in Melbourne in 2006 and aims to establish and maintain standards, systems and infrastructure for the collection and sharing of information of genetic variation effecting human disease.

INTERNATIONAL PROJECT DATA FLOW

The Human Variome Project Consortium envisions a world where a newly discovered genetic variant in a patient in Australia will lead to the diagnosis, prognosis, treatment and cure of patients in Germany, Peru and Tonga, and vice-versa. Known genetic based illnesses will be catalogued, integrated and curated. Achieving this vision will require an unprecedented scale of cooperation among the healthcare systems of the world's countries, but the benefits, in concrete terms of decreased morbidity and mortality, increased diagnosis and treatment rates, lower costs of healthcare delivery, higher quality of life, make the effort required seem trivial.

The global nature of the Human Variome Project, combined with the sensitive nature of genetic information, presents a unique set of challenges for our goal of universal collection. A centrally mandated, "one-size-fits-all" approach is not feasible, as it would be subject to the ethical and legal requirements of all participating nations, as well as needing to be sensitive to various cultural differences relating to genetic health. For this reason, the Human Variome Project has developed a two channel data collection strategy. One channel collects and collates data on a gene or disease specific basis from all sources including the scientific literature. The second channel collects data on a country specific basis; National Nodes. This strategy is designed to ensure complete collection of all genetic variation data and its accurate and timely expert curation.

OVERVIEW



OVERVIEW OF AUSTRALIAN NODE PROJECT

The Human Variome Project Australian Node was established by consensus following a meeting of a consortium of Australian clinicians and pathologists in November of 2008. The pilot phase was funded as an Australian Federal Government National eResearch Architecture Taskforce (NeAT) projects supported by the NCRIS Platforms for Collaboration. The pilot phase established and tested software systems for the collection of molecular information from Australian laboratories together and established an ethical framework and a management structure.

THE HUMAN VARIOME PROJECT AUSTRALIAN NODE(HVPA)

The Human Variome Project Australian Node is a project conducted in collaboration with but fully independent of the International Human Variome Project (HVPI). The aim of HVPA is to work within Australia to aid the efforts of clinicians, diagnostic and clinical laboratories, researchers and counsellors dealing with Genetic illness. This will be achieved foremost by collecting and sharing Australian information on genetic variation and its impact on human health. This information will be stored within a centralised electronic data repository(s) readily accessible by members of the Australian medical and research community. This data will then be shared with global gene/disease specific databases where the information will inform the global community be instrumental in the decisions regarding pathogenicity of changes and diagnosis.

It is intended that the final repository for all data reside with global centralised databases which will allow access by researchers and clinicians to large cohorts of genetic data taken from around the globe, contribute to quality assurance within genetic testing laboratories and facilitate the standardisation of decisions regarding the clinical significance of mutations and patient therapy.

The World Health Organization (WHO), European Commission (EC), United Nations Educational, Scientific and Cultural Organization (UNESCO) and the Organisation for Economic Co-operation and Development (OECD) have all recognised the need for such systems to assist researchers and clinicians to reliably diagnose, treat and inform patients about all genetic disorders.

The Human Variome Project is in official partnership of UNESCO and collaborates closely with the Basic Sciences Division.

Benefits of reporting data to a central Australian database: *(from minutes of Melbourne Meeting, Initiating the Australian Node, November 24, 2008, Desirée du Sart)*

- Allows continuous updating of information and testing assays.
- Provides quality assurance of in-house data.
- Allows greater assurance of a gene's pathogenicity if observed across a wide range of genetic backgrounds.
- Can give consistency of interpretation of genetic variation between different branches of the same family.
- Allows new areas of research to flourish, encourages collaboration and translation

MANAGEMENT STRUCTURE

The Human Variome Project Australian Node exists as a section within the Pathology Department of the University of Melbourne. As such, its management is overseen broadly by the Department of Pathology for the University of Melbourne. The Director/CEO of HVPA will report on a regular basis to the Head of the Department of Pathology, University of Melbourne. Day to day operational matters are the responsibility of the Director/CEO and the management team

HVPA aims to meet world best practice in Governance, policy development, strategic planning global management and budgeting and fund raising. Two committees have been established to assist these committees will also ensure the interests of all stakeholders and users are represented.

- an Advisory Committee to advise the Director/CEO of HVPA and a Stakeholders and users Committee to communicate the views of the users of the Australian Node service; and

- such sub-committees that are necessary from time to time to manage the day-to-day operation of the Human Variome Project Australian Node.

ADVISORY COMMITTEE

The Advisory Committee will advise on Policy, Strategic Planning and Budgeting of the HVP. The committee will meet quarterly (face to face once a year, otherwise by teleconference) to review reports on all aspects of the Australian Node including collection, storage and dissemination procedures. The committee in conjunction with the Director /CEO will give an update on progress on a regular basis to the Human Variome Project Australian Node Consortium membership via a quarterly bulletin and yearly seminars.

The Advisory Committee will also maintain a close interaction with the international community through their appointed representative on the Human Variome Project International Confederation of Countries Advisory Council.

There should be a balance of skills experience and independence on the committee appropriate to the expected responsibilities and function

APPOINTMENT OF COMMITTEE MEMBERS

The Advisory Committee will consist of

- A maximum of 8 members, a majority of which are independent of the stakeholder/users
- Include 3 members which are representatives of the stakeholder /users committee
- The Chairperson be the Director/CEO of the HVP

TERMS OF REFERENCE

- The Advisory Committee will advise on the establishment and maintenance of all the administrative, ethical and practical components required to implement and maintain the Human Variome Project Australian Node.
- The Advisory Committee is responsible for advising how the Human Variome Project Australian Node should implement the Standards and Guidelines developed by the International Human Variome Project.
- The Advisory Committee may create subcommittees to oversee certain functions the Australian Node, such as data access, variant interpretation and incentives for submission.

STAKEHOLDERS /USERS COMMITTEE

The Users Committee is a representative body of users of the Australian Node Repository that exists to advise Project staff on how to improve the Australian Node services (i.e. the Repository, collection systems, etc.)

APPOINTMENT OF COMMITTEE MEMBERS

The Project Director may appoint users of the Australian Node Repository to the Users Committee at his or her discretion. The Advisory Committee on the advice of the project director shall endeavour to ensure that all user types are equally represented.

The membership should where possible consist of 1 representative from each of the six states and two mainland territories of Australia. Members will be elected by the Human Variome Project Australian Node Consortium. The Committee shall elect from amongst themselves a chairperson.

- The Stakeholders /Users Committee shall determine:
- the genes to be included in the Australian Node database;
- the minimal and complete dataset definitions for each gene; and

- any validation constraints applicable to specific fields.
- The Users Committee shall determine:
- the data access levels supported by the Repository;
- the user types that make up each access level; and
- the level of access required for each field defined within the Repository.
- The Users Committee may be asked by the Project Director to render an opinion on any aspect of the Repository.

ACCESS COMMITTEE

The Access Committee is a sub-committee of the Advisory Committee set up to review all requests for access to data held by the Human Variome Project Australian Node.

APPOINTMENT OF COMMITTEE MEMBERS

The Chair of the Advisory Committee shall appoint three members of the Advisory Committee to the Access Committee. The Project Director shall be the fourth member. The Access Committee shall elect a chairperson from amongst themselves.

TERMS OF REFERENCE

- The Access Committee shall review all requests for access to data held by the Human Variome Project Australian Node.
- The Access Committee shall, in performing its review, consider:
- relevance/importance of stated need for access;
- completeness/accuracy of submitted information confirming applicants identity;
- completeness/accuracy of submitted information confirming applicants stated need; and
- any applicable ethical considerations.
- The Access Committee may delegate all or some of its review functions to Project staff.
- The Access Committee shall review all current access rights holders every two years.
- The Access committee may revoke access rights at any time.