

## The Human Variome Project - Introduction

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The Human Variome Project ([www.humanvariomeproject.org](http://www.humanvariomeproject.org)) was created to focus on the need to collect all variation in all genes causing human disease from all countries, and is dedicated to improving health outcomes by facilitating the unification of data on human genetic variation and its impact on human health. It supports the use of human variation information in clinical & research environments across the world. The Human Variome Project will establish systems to collect and make available information on all genetic variations associated with human disease.

This will enhance genetic health care, therapy and research by making accessible the data which is essential for such care. Naturally much has been done as mutations have been collected for over 50 years, but no routine overall system is available.

These outcomes cannot be achieved or funded by one organisation or one funder, it requires many dedicated persons and funding bodies in all or certainly many countries to spread the load. Therefore pilot collections are now underway and a clear picture of how to proceed is emerging. These systems need to be transferable worldwide so that duplication of system development is avoided.

Therefore we are encouraging community experts in the various diseases to join together to create consortia to initiate and in some cases merge numerous established databases to collect and curate information on their genes of interest.

Recent work by the InSiGHT group, in the inherited HVP/InSiGHT colon cancer study in four mismatch repair genes, is available to assist in this process. Also, work is being carried out by the Australian Node Pilot, to collect information on all genes along with its clinical data from a country. Therefore essential software and pilot work is available and underdevelopment and awareness of the need is rising. Other key pilots are DMuDB in the UK (<http://www.ngrl.org.uk/Manchester/dmudb.html>) and the ETHNOS system (<http://www.goldenhelix.org/>) for collecting in countries.

Disease specific efforts are needed driven by experts to split the work. We hope this meeting will stimulate a vigorous Neurogenetics Database Initiative.