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Golden Helix Institute of Biomedical Research: Interdisciplinary research and educational activities in pharmacogenomics and personalized medicine

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Abstract

The Golden Helix Institute of Biomedical Research is an international non-profit scientific organization with interdisciplinary research and educational activities in the field of genome medicine in Europe, Asia and Latin America. These activities are supervised by an international scientific advisory council, consisting of world leaders in the field of genomics and translational medicine. Research activities include the regional coordination of the Pharmacogenomics for Every Nation Initiative in Europe, in an effort to integrate pharmacogenomics in developing countries, the development of several National/Ethnic Genetic databases and related web services and the critical assessment of the impact of genetics and genomic medicine to society in various countries. Also, educational activities include the organization of the Golden Helix Symposia®, which are high profile scientific research symposia in the field of personalized medicine, and the Golden Helix Pharmacogenomics Days, an international educational activity focused on pharmacogenomics, as part of its international pharmacogenomics education and outreach efforts.

Keywords

Golden Helix Institute; pharmacogenomics; translational medicine; genetic databases; symposia; genetics education; society

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Introduction

The Golden Helix Institute of Biomedical Research [GHI; 101] was established in 2003 as a European-based international non-profit scientific organization with interdisciplinary research and educational activities in the field of pharmacogenomics and personalized medicine. The GHI is named after the house of Francis Crick ("The Golden Helix"; 19/20 Portugal Place, Cambridge, UK) to emphasize the focus of its activities on human genomics and personalized medicine. The Institute's activities focus mainly in Europe and also in Asia, through its Japan representative office and in Latin American countries. As a non-profit organization, the mission of the GHI is the transfer of knowledge and scientific results among local researchers and internationally renowned scientists to the scientific community and the society through participation in collaborative scientific projects, a variety of scientific symposia and educational events, and establishment of public health policies in the field of pharmacogenomics and personalized medicine

The research and educational activities of GHI, outlined in Box 1are supervised by a 14-member International Scientific Advisory Council, consisting of internationally renowned scientists in the field of genomic medicine and supported by local and international research grants, while its mission also relies on the support of corporate, institutional and individual donations and volunteer work.

Research activities

The research activities of GHI are focused on three main areas: the encouragement of a European-wide application of pharmacogenomics, the development of genetic databases and related web applications and the critical appraisal of the impact of genetics to society.

European-wide application of pharmacogenomics

The Golden Helix Institute of Biomedical Research participates in multidisciplinary research to boost application of pharmacogenomics globally through the Pharmacogenetics for Every Nation Initiative (PGENI) [102]. For most common diseases there are a number of treatment modalities for which there is usually large individual variability in both efficacy and/or toxicity. Variation in DNA does help explain some of the differences in a person's response to a medication, but the cost of individual genetic testing will be prohibitive for many countries for the foreseeable future. The PGENI aims to develop innovative strategies for regulatory agencies and ministries to integrate pharmacogenomics into public health decision making (such as selection of a national formulary) without placing an extra burden on sparse healthcare funds. In 2010, the Institute assumed an active role in this project as the European Regional center of the PGENI initiative and is coordinating the recruitment of cases from various European developing countries [1].

In 2011, Czech Republic, Greece, Malta, Poland, Serbia, Slovenia and Turkey have joined this initiative in Europe [2], in early 2012 Croatia, Hungary, Lithuania, Slovakia and Ukraine have agreed to join this initiative (Fig. 1), while more countries have been invited to participate and are expected to join soon.

Golden Helix Genetic Databases

Genetic databases are gradually assuming an increasing importance in all areas of health care. The National and Ethnic Mutation Databases (NEMDBs) aim to record extensive information over the described genetic heterogeneity of an ethnic group or population. Since 2003, the GHI has devoted a significant amount of effort, to develop a number of NEMDBs and related software that record the incidence of genetic diseases and the corresponding

genome variation spectrum, with clinical relevance in various populations and ethnic groups worldwide. Also, the GHI developed ETHNOS, an off-the-shelf bioinformatics suite to facilitate development and curation of National/Ethnic Genetic and Mutation frequency databases [3]. This software has enabled the development of a number of these resources in various populations, such as Greek [4], Cypriot, Iranian [5], Lebanese, Israeli [6], and Egyptian. Also, based on the same concept, FINDbase [103], a worldwide database to record frequencies of causative mutations and pharmacogenetic markers, has been developed [7]. This database has been recently refurbished including many new features and functionalities [8], while in close cooperation with SAGE Publishing Group (London, UK), the first "database-journal" was launched, i.e., a scientific journal that is interconnected with a database [9].

Finally, the GHI supports the development of the Greek node of the Human Variome Project [HVP; 104] and hosts the Node at the Golden Helix Server. The HVP Hellenic node has been formally incorporated in the Human Variome Project in 2010. At present, the Hellenic Node is supervised by a board of 11 geneticists originating from various academic institutions in Greece and involved in human genetics research, most of which sit at the Board of the Hellenic Society of Human Genetics, and has been built around the Hellenic NEMDB [3]. These activities have not only promoted the collection of genomic variation in the Hellenic population but have also encouraged new studies to document the genetic heterogeneity of the commonest genetic disorders in various parts of the country. These studies confirmed that the β-thalassemia mutation spectra indeed differ in different parts of Greece, compared to the frequencies calculated for the entire country [10, 11], further outlining the need to perform such investigations to better orientate molecular genetic testing services in various parts of the country. Also, board members of the HVP Hellenic node participate in nationwide studies to critically ascertain the general public's awareness and healthcare professionals' opinion on genetic and pharmacogenomic testing services in Greece [12–14].

Web services in genomic medicine

To facilitate seamless access and retrieval of genomic data, the Golden Helix Institute of Biomedical Research has employed a Service Oriented Architecture (SOA) for its genomics databases [3, 10, 11]. Services have been designed to support both schematic recognition of data available as well as data retrieval using interoperability features. Extensible Markup Language (XML) web services have been implemented and deployed to serve key searching functionality at the back-end services. The same technology has been used to deliver Open Services which can be utilized after request by third parties in order to access the available Institute's data. The services are hosted at a data center infrastructure to ensure availability and quality of service features.

Genetics and Society

In the post-genomic era, despite the rapid incorporation of genetic testing services into the mainstream clinical practice, in many European countries there is very little knowledge as to how either the general public or medical practitioners perceive genetic and pharmacogenomic testing services.

The GHI has encouraged and financially supported nationwide studies in Greece in order to better understand how the general public, medical practitioners and public and private genetic laboratories perceive genetic and pharmacogenomic testing services. These surveys could be readily replicated in other countries in Europe with the ultimate aim of improving the public understanding of genetic and pharmacogenomic testing services, and facilitating the incorporation of genomic medicine into everyday clinical practice [12–14].

Educational activities

Apart from the above mentioned research activities, the GHI organizes high profile international research symposia and educational activities in the field of pharmacogenomics and personalized medicine.

The Golden Helix Symposia®

The Golden Helix Symposia [105] are high profile international research meetings. The topic of these symposia revolves around the fields of genomic and personalized medicine. In particular, these symposia series have the following features: (a) Symposia topics are selected by GHI International Scientific Advisory Council, so that new and often ground-breaking science is presented at every meeting, (b) Symposia range in length from 2 to 3 days, and average attendance is 200 participants, mainly staff scientists, post-doctoral fellows and students from academia, working in the field of personalized research, and also scientists from pharmaceutical and biotechnology industries, government agencies and private and research foundations from various countries worldwide, (c) Conference venues are usually prestigious locations in major cities or summer retreats in the Southern Mediterranean or Eastern regions. Previous Golden Helix Symposia have focused on molecular cytogenetics [2008; 15, 16], pharmacogenomics [2009; 17] and translational medicine [2010, 2012; 18]. In several cases, the GHI provided fellowships to under-graduate and post-graduate students from low-medium income countries to defray registration, and often accommodation and travel, costs to participate in the Golden Helix Symposia®.

Golden Helix Pharmacogenomics Days

The Golden Helix Institute of Biomedical Research also organizes the Pharmacogenomics days, an international educational event that is organized in major cities with large academic hospitals. The aim of this event is to provide timely updates on the field of pharmacogenomics and personalized medicine to the local biomedical scientists and healthcare providers, to inform them on the application of pharmacogenomics in modern medical practice, and to bring together faculty members from universities and research institutes from the local scientific arena working in the field of pharmacogenomics in order to initiate collaborative projects in this field to the benefit of society. Previous Golden Helix Pharmacogenomics Days have been organized in Athens (Greece; 2009), Thessaloniki (Greece; 2010), Alexandroupolis (Greece; 2011), Cagliari (Italy; 2011) and Msida (Malta; 2011), while in 2012 two more Golden Helix Pharmacogenomics Days are scheduled and will be hosted in Belgrade (Serbia) and Patras (Greece; Fig. 1).

Other conferences

The Institute has been involved in the co-organization of scientific conferences in the field of genome medicine with other scientific organizations. In 2010 (May 10-14), the Institute organized in conjunction with the US Food and Drug Administration the 1st Latin American Pharmacogenomics Conference in San Juan, Puerto Rico. Also, in 2011, the GHI co-organized a conference related to the impact of Genetics to Society in Athens, Greece (4-6 November 2011) to enhance awareness of the general public over various issues pertaining to genetics and their application to modern medical practice. This meeting was encouraged by the Howard Hughes Medical Institute. Also, the Institute has been supporting the organization of international scientific events, such as the Human Genome Variation Society International Symposium on Mutations in the Genome in Paphos, Cyprus (2009) and the World Pharmacogenomics Summit in Boston, MA, USA (2011).

Future perspectives

Almost a decade after the launch of the GHI activities, the initial deliverables of this international effort already look promising. The future goals of the GHI include the expansion of the PGENI activities in Europe by inviting more developing countries to join this truly pan-European effort. Also, from the educational point of view, the GHI aims to establish the Golden Helix Pharmacogenomics Days as one of the main educational activities for PGENI. Educational and training activities for healthcare professionals on pharmacogenomics will significantly contribute to the adoption of pharmacogenomics particularly in developing countries and, reciprocally, to the spread of pharmacogenomics applications to the benefit of the general public in these countries.

At the same time, the GHI plans to expand the genetics and society research and outreach program to critically ascertain the views of the general public and physicians on genetics in order to facilitate smooth integration of genome medicine into the mainstream medical practice. The envisaged outreach program ranges from production of simplified comics for high school students, to public conferences to inform the general public on the benefits and pitfalls of genetics and public surveys related to genome medicine. Also, the long-term goal of GHI is to be more actively involved, in close partnership with academic institutions in educational programs on genome medicine to enable proper education of the future healthcare professionals in this discipline. Our previous efforts identified this as one of the major hurdles that hold back this field [12, 13].

Finally, the GHI, in close collaboration with faculty members from academic institutions in the region, plans to provide, on a competitive basis, internship opportunities to a limited number of talented post-graduate students, particularly from low-medium income countries, to perform part of their training in a state-of-the-art laboratory abroad and a number of long-term fellowships for top-class graduate students and post-doctoral scientists wishing to pursue an academic career in a center of excellence abroad.

Overall, the GHI, through the commitment and enthusiasm of its scientific advisory council, will steadily continue to serve the field of genomic medicine aiming to contribute to the incorporation of genomics into modern medical practice.

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Web sites

101. Golden Helix Institute of Biomedical Research: http://www.goldenhelix.org.
102. Pharmacogenomics for Every Nation Initiative: http://www.pgeni.org.
103. FINDbase database: http://www.findbase.org.
104. Human Variome Project: http://www.humanvariomeproject.org.
105. Golden Helix Symposia: http://www.goldenhelixsymposia.org.

Executive Summary

- The Golden Helix Institute of Biomedical Research is an international non-profit scientific organization active in Europe, Asia and Latin America.
- The Institute has interdisciplinary research and educational activities in the field of pharmacogenomics and translational medicine, which are supervised by an international scientific advisory council, consisting of world leaders in the field of genomics and translational medicine.
- The Institute is the regional coordination center of the Pharmacogenomics for Every Nation Initiative in Europe, in an effort to integrate pharmacogenomics in developing and other countries.
- Also, the Institute has been involved in the development of several National/Ethnic Genetic databases and related web services to document the genetic heterogeneity in different populations worldwide.
- As part of these efforts, research projects include the critical assessment of the impact of genetics and genome medicine to society in various developing countries.
- Educational activities include the organization of the Golden Helix Symposia®, which are high profile scientific research symposia in the field of personalized medicine, and the Golden Helix Pharmacogenomics Days, an international educational activity focused on pharmacogenomics, as part of the Institute's pharmacogenomics education and outreach efforts.
- Future plans include the expansion of the PGENI activities in Europe and of genomics education and society outreach through various fora, such as the Golden Helix Pharmacogenomics Days and other educational activities, in order to facilitate smooth integration of genome medicine into mainstream medical practice.

Box 1. Outline of the GHI research and educational activities

a. **A. Research activities**
   - European Regional Center of the PGENI project
   - Development of the Golden Helix Genetic databases
   - Web services in genomic medicine
   - Critical appraisal of the impact of genetics on society

b. **B. Educational and outreach activities**
   - Organization of the Golden Helix Symposia®
   - Organization of the Golden Helix Pharmacogenomics Days
   - Co-organization of other scientific conferences in the field of pharmacogenomics and personalized medicine

Figure 1.
Depiction of the GHI main research and educational activities in Europe. The PGENI logo indicates the developing countries that participate in the PGENI project in Europe (the European PGENI coordinating office in Athens is indicated with the PGENI logo in a rectangle) and the Golden Helix Institute's logo indicates the cities that hosted or will host, in 2012, a Golden Helix Symposium® and/or a Golden Helix Pharmacogenomics Day.