Genomic Medicine Alliance

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EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

CHMP Pharmacogenomics Working Party (PGWP)

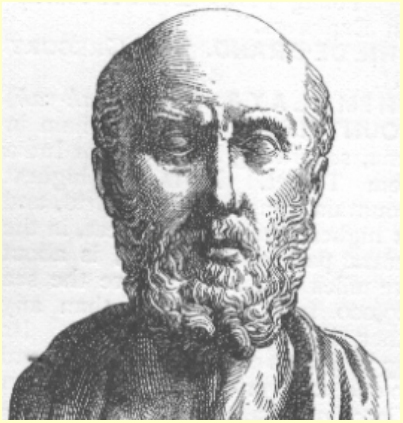
Disclaimer

Declared conflict of interests: None

The opinions expressed in this presentation do not reflect the policies and position of the European Medicines Agency

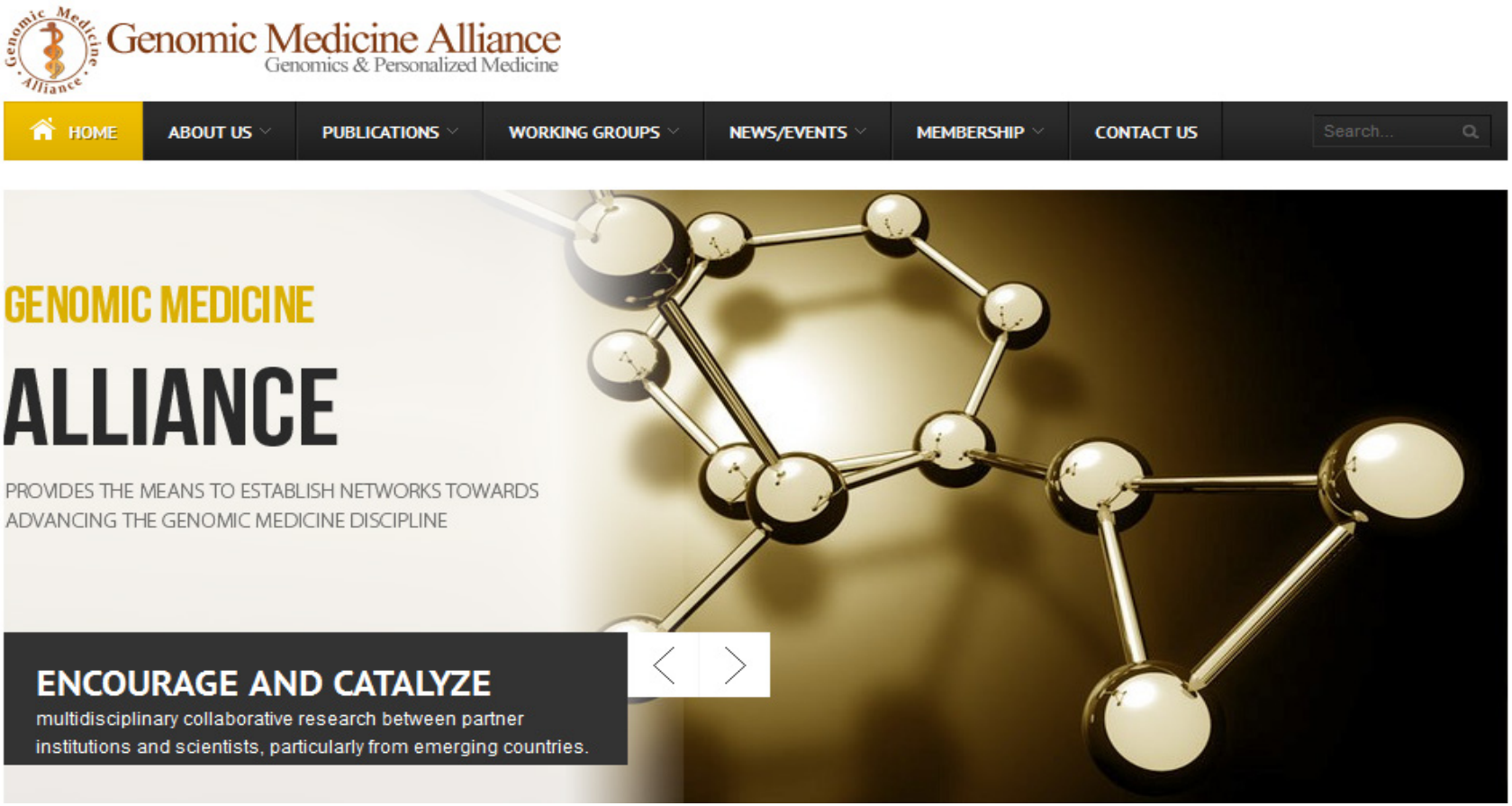
Genomic Medicine Alliance

Genomic Medicine:
An Historical perspective



Hippocrates (4th century B.C.):

«It is more important to know what kind of person suffers from a disease than to know the disease a person suffers»



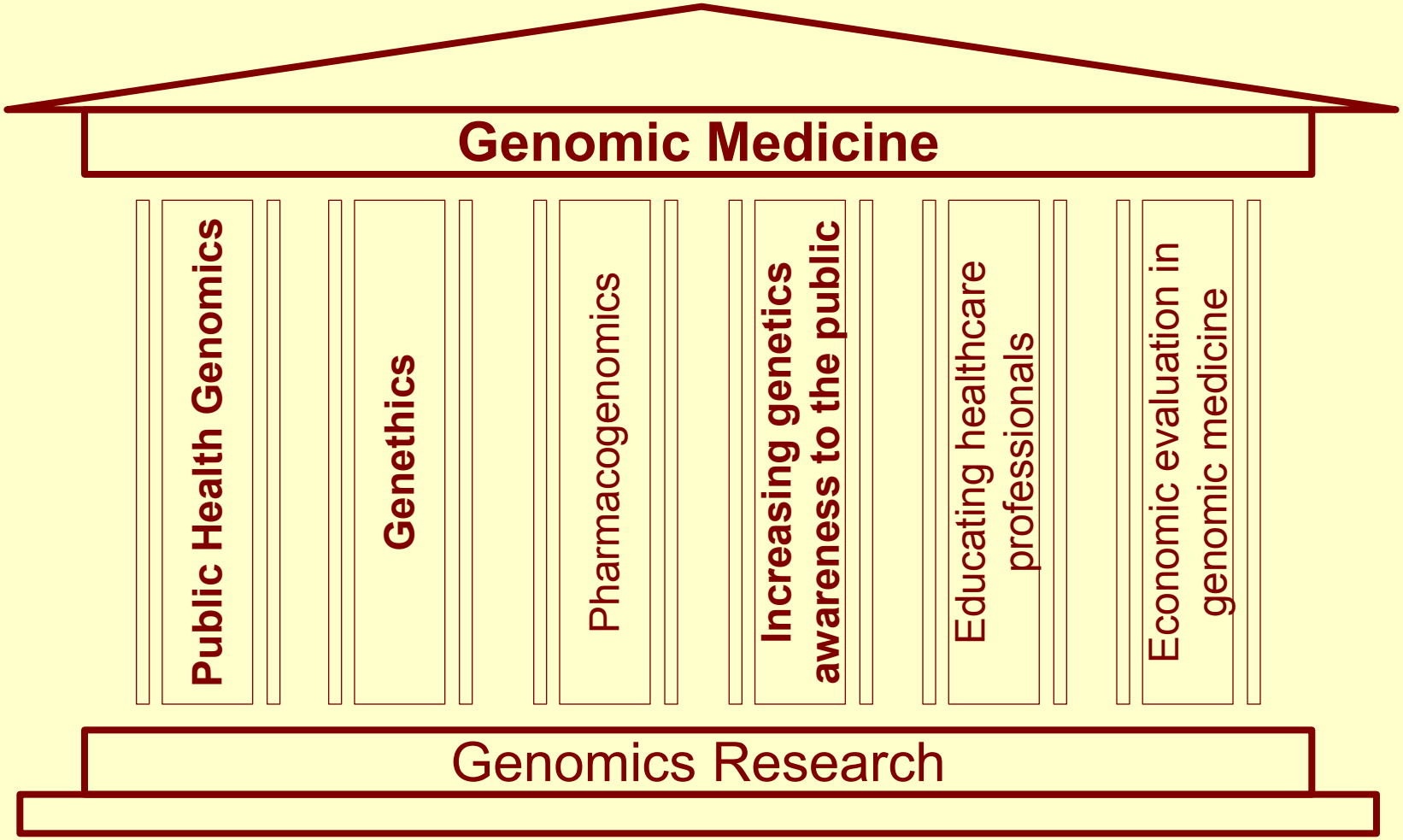
Aims to build/strengthen collaboration ties between academics, researchers, regulators, and the general public interested in all aspects of genomic medicine, focusing in particular on translating results from research into clinical practice.

www.genomicmedicinealliance.org

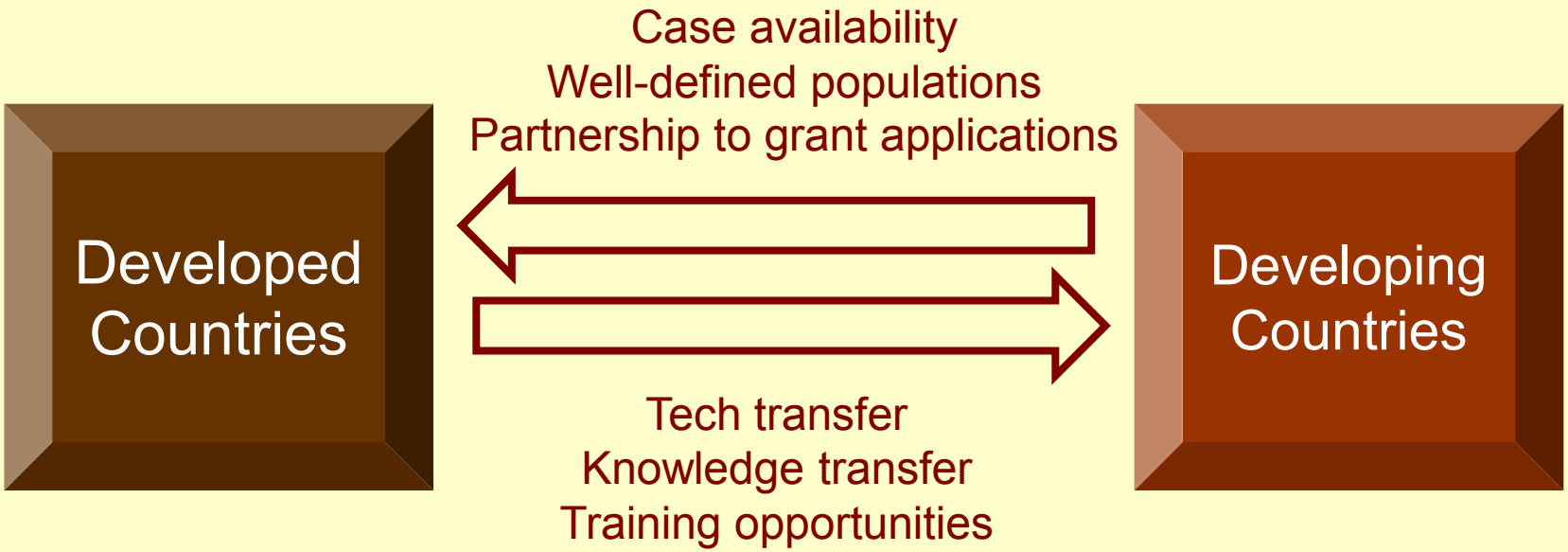
Key features

- The Genomic Medicine Alliance is an international scientific research network.
- At present, research activities are self-financed by participants' own funding sources and partly funded by the Golden Helix Foundation (a London-based UK Charity).
- Scientific coordination is provided by the Scientific Advisory Committee, comprising of 13 internationally renowned scientists in the field of Genomics Research.
- Administrative assistance is provided by the Golden Helix Foundation staff.
- Registration to the Genomic Medicine Alliance is free-of-charge to encourage participation of researchers from developing and low-income countries.

1. Paving the path from Genomics Research to Genomic Medicine



2. Bridging developed with developing and low-resourced countries




Cagliari, IT


Rotterdam, NL


Patras, GR

Scientific Advisory Committee

- Fahd Al-Mula (Safat, KW)
- Angela Brand (Maastricht, NL)
- David Cooper (Cardiff, UK)
- Vita Dolzan (Ljubljana, SI)
- Paolo Fortina (Philadelphia, PA, USA)
- Federico Innocenti (Chapel Hill, NC, USA)
- Michael Lee (Yokohama, JP)
- Milan Macek Jr (Prague, CZ)
- George P. Patrinos (Patras, GR)
- Barbara Prainsack (London, UK)
- Alessio Squassina (Cagliari, IT)
- Effy Vayena (Zurich, CH)
- Athanassios Vozikis (Piraeus, GR)

Working groups

- Genome Informatics
- Pharmacogenomics
- Cancer Genomics
- Public Health Genomics
- Ethics in Genomics
- Economic evaluation in Genomic Medicine

GMA - Membership basis

Scientific Advisory Committee

Senior National

Representatives and Working

Group Leaders

Regular Members

GMA - Membership basis

Europe: 92 members from 31 countries

Middle East: 35 members from 6 countries

Africa: 12 members from 4 countries

Australasia: 24 members from 6 countries

The Americas: 16 members from 6 countries

<u>TOTAL</u>: 179 members from 53 countries

*** Updated: December 2013**

Pharmacogenomics

- Determination of the incidence of pharmacogenomics biomarkers in 26 European populations to optimise medication treatment modalities and to minimize adverse reactions.
- Provide the proof of principle of the use of whole-genome sequencing for pharmacogenomic testing.
- Establish a detailed correlation among genomic biomarkers and adverse drug reactions in European and Southeast Asian populations.

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The EuroPGx project: Overview

- Collection of 45-90 (1st tier) or 450-900 (2nd tier) DNA samples primarily from developing nations and also selected developed countries in Europe [independently and in collaboration with the PGENI initiative (The Golden Helix Foundation is the European Regional PGENI centre).
- Genotyping for 1,936 pharmacogenomically-relevant variants in 231 ADMET-related pharmacogenes.
- Develop off-the-shelf solutions for pharmacogenomic testing in participating developing countries.
- Assist in prioritize medication selection in participating developing countries (in collaboration with the PGENI initiative).

The GoldenHelix FOUNDATION

PGENI

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The EuroPGx project

McLeod et al., in preparation, 2014; Dalabira et al., in preparation (2014)

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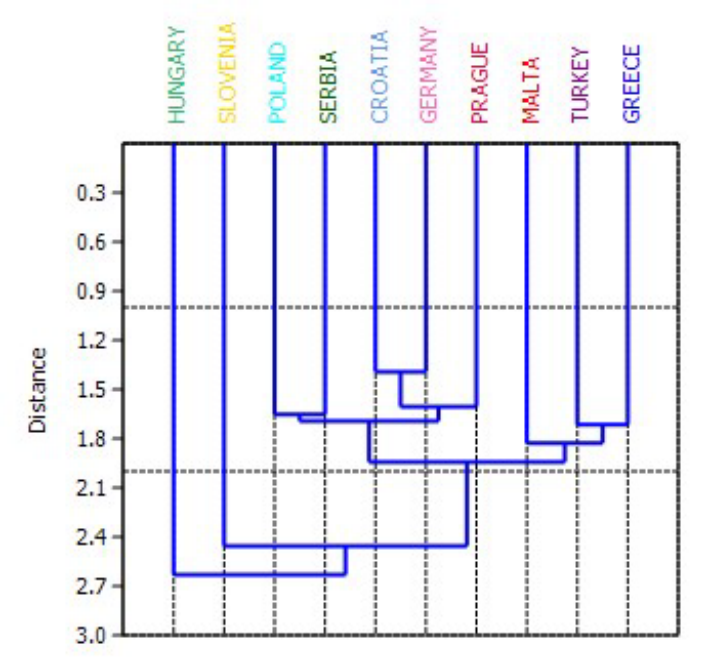
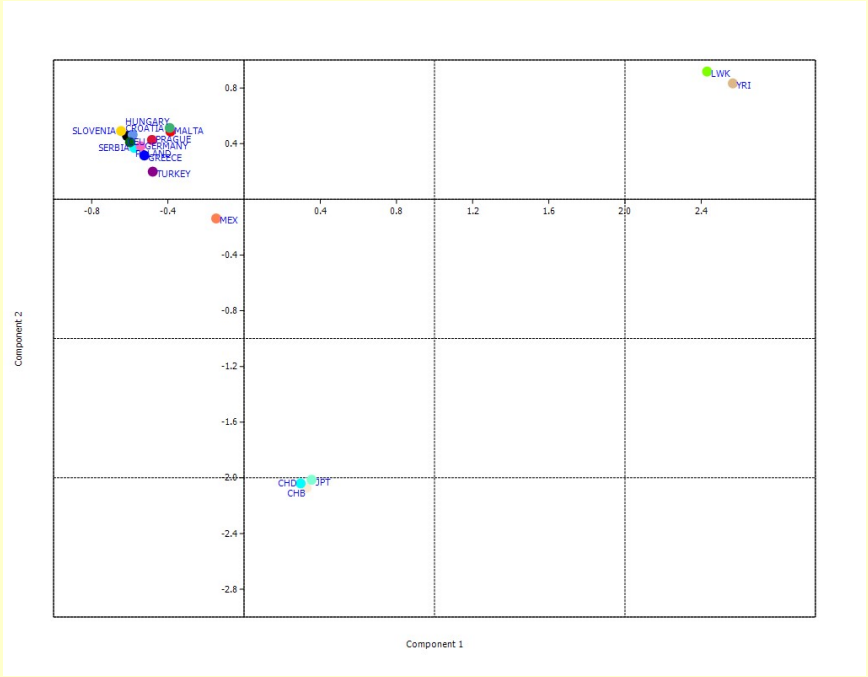
The EuroPGx project





- European populations display significant differences in >130 pharmacogenomic biomarkers each.
- Replication of these findings in larger population samples to establish common grounds for pharmacogenomic testing in developing countries.

McLeod et al., in preparation, 2014; Dalabira et al., in preparation (2014)

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The NextGenPGx project

Pilot: 69 whole genomes (CG69 collection)

Follow-up: 413 whole genomes (adult Caucasians)

- ✓**All genomes were whole-genome sequenced (110x).**
- ✓**Analysis of all variants (known and novel in ADMET-related genes; Inclusion of variants with the highest quality score only).**
- ✓***In silico* analysis of novel variants.**
- ✓**Independent whole-genome sequence analysis of a 7-member Greek family in the ADMET-related genes.**

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Complete genomics

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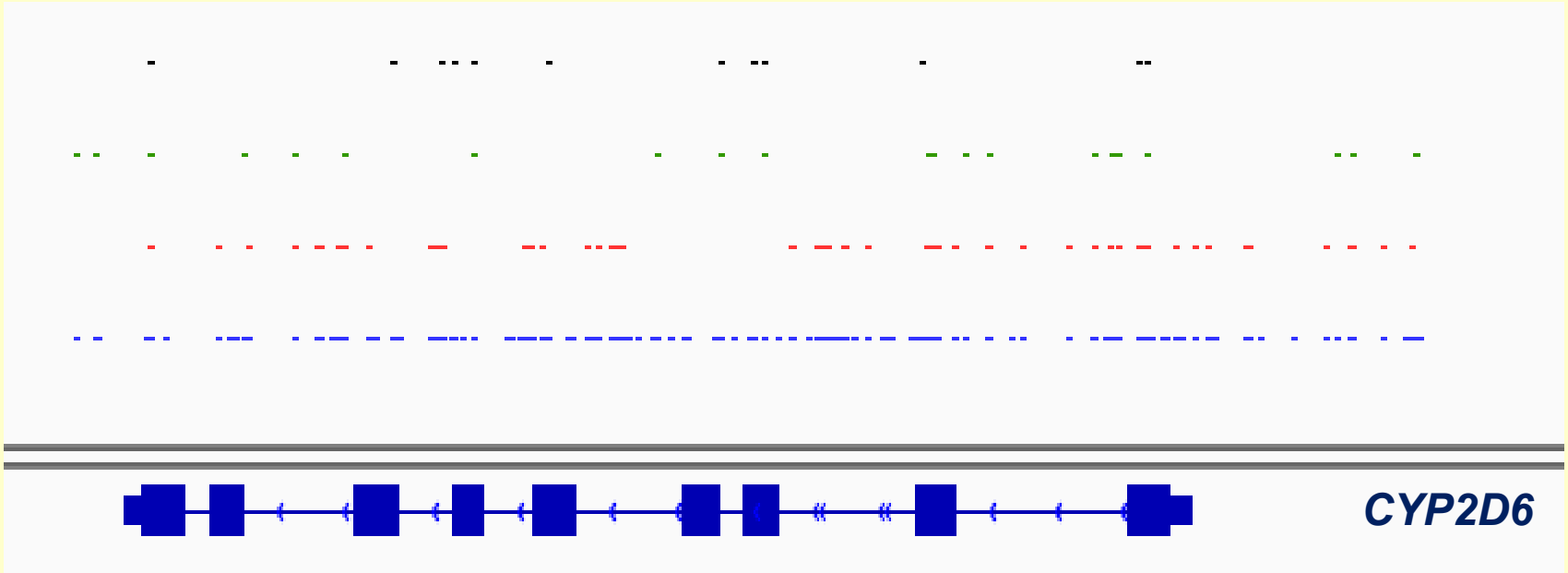
Mizzi et al., Pharmacogenomics (accepted, 2014)

The NextGenPGx project



- 18,058 variants in each individual
- 16,485 novel potentially functional variants (961 variants with freq >1%)
- 4,480 novel potentially functional variants in the exome.

Mizzi et al., Pharmacogenomics (accepted, 2014)

Genome Informatics

- Develop 3 new National/Ethnic Genetic databases (Kuwaiti, Moroccan, Bahraini) to document the incidence of genetic disorders in these countries.
- Migrate 5 existing National/Ethnic Genetic databases (Lebanese, Serbian, Cypriot, Greek, Egyptian) using the upgraded ETHNOS software.
- Developing an electronic Molecular Diagnostic Assistant, for translating Pharmacogenomics results into a meaningful format for clinicians.
- Establishment of a whole-genome National data repository to provide allele frequency data in a aggregate level.

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eMoDia

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Genome Informatics

❖ **Closely collaborate with major European research ini-tiatives, such as RD-Connect, RD-Neuromics, etc**

- Harmonize and develop common standards for databases and patient registries for rare diseases.
- Develop clinical bioinformatics tools, including data mining and knowledge discovery tools for analysis and integration of molecular and clinical data to discover new disease genes, pathways and therapeutic targets.
- Endorsing scientific and educational meetings.

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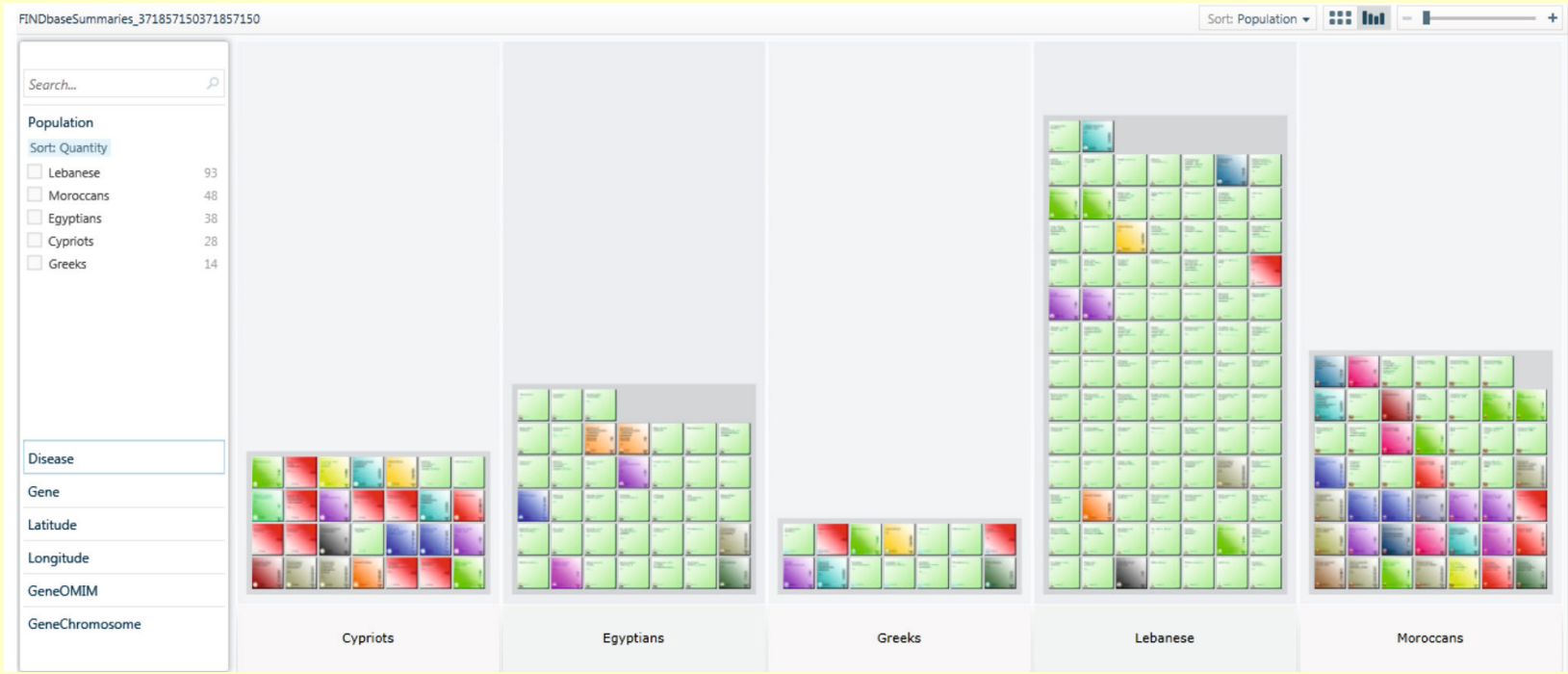
Genome Informatics



D1020–D1026 *Nucleic Acids Research, 2014, Vol. 42, Database issue* *Published online 14 November 2013*
doi:10.1093/nar/gkt1125

Developments in FINDbase worldwide database for clinically relevant genomic variation allele frequencies

Papadopoulos et al., *Nucleic Acids Res*, 42:D1020-6, 2014

Public Health Genomics

- Undertake nationwide studies to better understand the level of general public awareness and healthcare professionals (physicians, pharmacists, nutritionists, *etc*) genetics education (Mai et al., *Pers Med* 2014).
- Engage into a stakeholder analysis to determine the measures to be undertaken to expedite genomic-based medical decision making process (Mitropoulou et al., in prep).
- Encourage and facilitate the co-organization of educational events over pharmacogenomics (Golden Helix Pharmacogenomic Days) in various European countries (currently 13 events in 8 countries).

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Genethics

- Surveying the landscape of DTC and OTC genetic testing in various developing countries (Mai et al., *Pers Med* , 2014).
- Working together with National Genetic Societies and Ethics Committees to establish guidelines for ELSI pertaining to genetic testing.


Patrinos *et al. Human Genomics* 2013, **7**:17
http://www.humgenomics.com/content/7/1/17

Human Genomics

OPINION ARTICLE Open Access

Genetic tests obtainable through pharmacies: the good, the bad, and the ugly

Patrinos et al., *Hum Genomics* 7:17, 2013

Economic Evaluation

• Cost-effectiveness and cost-utility analyses of various genetic-based medical treatments by reducing the incidence of adverse drug reactions, and reciprocally healthcare expenditure at the national level.
Current projects focus on anticoagulation treatment of warfarin (Croatia), acenocoumarol (Serbia, Greece) and clopidogrel (Serbia) and nicotine addiction treatment (Greece).

• Endorsement of the production of the textbook “Economic Evaluation in Genomic Medicine” by Elsevier/Academic Press in early 2015.

Genomic Medicine Alliance

Co-organization of educational activities

Pharmacogenomics and Genomic Medicine: Bridging Research and the Clinic

September 11-15, 2014

Aegina island, Athens, Greece

Genomic Medicine Alliance

http://summerschools.goldenhelix.org

Special issue in *Personalized Medicine* - "Working towards personalization of Medicine: Genomics in 2014"

Future Medicine

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- 2013 Catalogue
- Library Recommendation
- Future Science Group

Write for *Personalized Medicine's* upcoming special issue

Personalized Medicine will publish a special issue in September 2014 entitled **"Working towards personalization in medicine: Genomics in 2014"** guest edited by **George Patrinos** (Universityof Patras, Greece) and **Barbara Prainsack** (King's College London, UK). The contents of this special issue will include review, perspective and original articles reporting timely updates over the various disciplines related to genomic medicine. Compilation of this special issue is encouraged by the **Genomic Medicine Alliance** (www.genomicmedicinealliance.org).

Personalized Medicine translates recent genomic and proteomic advances into the clinical context, and addresses the impact of pharmacogenomics in modern medicine, providing an integrated forum for all players involved in the healthcare value chain. Coverage includes news and views, current awareness regarding new biomarkers, concise commentaries and analysis, reports from the conference circuit and full review articles. *Personalized Medicine* is indexed by Scopus, EMBASE/ExcerptaMedica, Chemical Abstracts, ISI Journal CitationReports®, Science Citation Index Expanded (SciSearch®) and Current Contents®/Clinical Medicine, with an **Impact Factor** of **1.51** (2012). The aims and scope, editorial board and published issues to date are all available on our website.

Contributions to this special issue should address at least two of the following questions:

- What practices and technologies in your field have been enabling factors of more personalized medicine and healthcare in recent years?
- What would be your vision, in very concrete terms, for personalized medicine and healthcare from the perspective of your field?
- Where are the main obstacles to reaching this vision, from today's perspective?
- What are the health-economic implications of practices and technologies of personalization that are in use today in your field?
- What are the social justice implications of practices and technologies of personalization that are in use today in your field?
- What are the perspectives of patients on these issues?
- How can initiatives such as the Genomic Medicine Alliance help to bring us closer to a vision of personalized medicine that leads to better, more patient-centered, and ultimately also more cost-effective healthcare?

We are delighted to invite you to submit an article for publication in this special issue (subject to peer-review). If you are interested in contributing to the issue or would like further information please contact the Commissioning Editor Hannah Wilson at hannah.wilson@futuremedicine.com

Genomic Medicine Alliance

http://www.futuremedicine.com/page/PMESFI

Future plans

- Expand the membership basis, particularly with members from developing countries in the Middle East, Asia, Latin America and Africa.
- Partner with other multinational groups (e.g. European Alliance for Pers Med) to pursue common goals.
- Expand the educational and outreaching activities in Europe, Middle East and Southeast Asia.
- Establish, in collaboration with the Golden Helix Foundation, short- and long-term research fellowships for graduate students and early-stage researchers from developing countries to pursue research in Centers of Excellence in developed countries.
- Affiliate with an Elsevier open-access genomics journal as the official journal of the Genomic Medicine Alliance.

Thank you

www.genomicmedicinealliance.org