Access for All: A Personalised Approach

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Editors
Angela Brand, Maastricht
Kapaettu Satyamoorthy, Manipal
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- Policy Statements

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of two complete Ara h 2 isoforms cDNA. Int Arch Allergy Immunol 2003;131:14–18.
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Call for Research Collaboration to Understand the Genetic Architecture of Founder Populations Throughout the World to Impact Health and Disease

Announcing the DRIFT Consortium:

**Discovery Research Investigating Founder Population Traits**

The Regeneron Genetics Center (RGC), located in Tarrytown, New York, is a large-scale, fully integrated genomics program that builds on Regeneron Pharmaceuticals, Inc. well-established expertise in genetics and related technologies. The objective of the RGC is to use DNA sequencing to gain new insights into disease mechanisms, identify novel therapeutic targets and ultimately speed drug discovery and development to help patients in need. We work collaboratively with academic colleagues worldwide and use a number of study designs and experimental approaches. One approach is to identify and study individuals with rare loss of function mutations and their associated phenotypes. Founder populations offer a unique opportunity to identify these informative alleles since they may be enriched in the population through drift and also may be more often found in their homozygous state through endogamy.

We have founded the DRIFT Consortium with the goals of (i) cataloging population-specific allelic architecture; (ii) understanding the biological and functional consequences of specific mutations identified; and (iii) sharing and establishing best practice approaches to relieve disease burden in these populations.

DRIFT is planning two tiers of collaboration models (see details below). For both models, we intend to broadly share data and results with the research community. If exciting new results are generated from a Tier 1 or Tier 2 collaboration, there will be potential for the design and funding of follow-up "genotype-first call-back" studies for additional collaborative research to delve more deeply into biological mechanisms and pathways.

**Tier 1**

**GOAL:** To canvas the allelic architecture of the population by exome sequencing and GWAS chips from relatively unrelated individuals

- Collaborator will provide the RGC de-identified DNA samples (300-400)
- RGC will provide high-depth exome sequence and GWAS chip data back to collaborator free of charge
- No exchange of phenotype information is necessary for Tier 1
- If the joint sequence data is used for any genotype-phenotype analyses, we ask that results be shared with Regeneron; collaborator is free to publish results
- A short form material transfer agreement will be used to govern the collaboration

**Contacts:**

- **Alan R. Shuldiner, M.D.**  
  *Vice President & Co-Head, RGC*  
  E: Alan.Shuldiner@regeneron.com  
  T: +1-914-847-1081

- **George P. Patrinos, Ph.D.**  
  *Associate Professor, University of Patras & Member, Scientific Advisory Committee, Genomic Medicine Alliance*  
  E: gpatrinos@upatras.gr  
  T: +30-2810-962-339

**Tier 2**

**GOAL:** To establish a partnership/ collaborative effort focused on novel gene discovery for phenotypes of mutual interest

- An academic collaboration model where Collaborator and the RGC will jointly develop the research plan
- A larger number of DNA samples (100s – 10,000s) will be provided by Collaborator; the exact number will be determined jointly
- RGC will provide all exome sequence data to Collaborator free of charge
- As part of the collaboration, deidentified phenotype data will be shared
- Data analyses of the combined sequence and phenotype data set will be performed collaboratively and each party is free to use the data set for its internal research
- Collaborators will be encouraged to publish results and each party is free to use published results for any and all purposes
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All submitted abstracts will be published in a Supplement issue of Public Health Genomics, the official journal of the Genomic Medicine Alliance indexed in PubMed (2014 Impact Factor: 2.208)

Registration and abstract submission deadline: July 2, 2016

The Genomic Medicine Alliance is a global research network aiming to create collaboration ties between academics, researchers, regulators, and the general public interested in all aspects of genomics and personalized medicine. The Alliance provides the means to establish networks and to encourage collaborative work towards advancing the Genomic Medicine discipline, focusing in particular on translating results from academic research into clinical practice.

Public Health Genomics is the official journal of the Genomic Medicine Alliance, aiming to provide an ideal publication forum to members of the Genomic Medicine Alliance.

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Key Challenges and Solutions in Personalised and Precision Medicine

Getting Personal: The Future of Medicine and Clinical Trials

Editors
Denis Horgan
Mark Lawler
Kapaettu Satyamoorthy
Angela Brand

Personalised and precision medicine (PPM) has the potential to revolutionise not only the treatment but also the prevention of diseases. Advances such as the successful development of targeted therapies like imatinib mesylate for chronic myeloid leukaemia and Herceptin/trastuzumab for erbB-2-positive breast cancer have changed clinical practice for these cancer indications. However, for every breakthrough therapy, there is a succession of ‘one-size-fits-all’ approaches that deliver minimal or no benefit despite the billions of euros/dollars and the number of years invested in their development.

There are various challenges now to facilitate an accelerated ‘concept-to-clinic’ culture, but in a way that translates preliminary discovery ‘hits’ into robust but affordable clinical wins for patients. In this special issue of Public Health Genomics, several stakeholders outline the key challenges and the potential solutions that will help embed PPM in evolving innovative clinical trial frameworks. These have been brought together by the Brussels-based European Alliance for Personalised Medicine, a multi-stakeholder organisation that includes patient groups, researchers, front-line healthcare professionals, industry representatives and more, and is in regular discussions with the European Council, Commission, Parliament and Member State governments as it aims to see PPM integrated into healthcare systems across the EU.

Editorial

• Getting Personal: Accelerating Personalised and Precision Medicine Integration into Clinical Cancer Research and Care in Clinical Trials: Horgan, D.; Lawler, M.; Brand, A.

• Personalised and Precision Medicine in Cancer Clinical Trials: Panacea for Progress or Pandora’s Box?: Lawler, M.; Sullivan, R.

• The Challenges of Precision Oncology Drug Development and Implementation: Hollingsworth, S.J.; Blankin, A.V.


• Decision-Making Process Related to Participation in Phase I Clinical Trials: A Nonsystematic Review of the Existing Evidence: Gorini, A.; Mazzocco, K.; Pravettoni, G.

• Virtual Clinical Trials, an Essential Step in Increasing the Effectiveness of the Drug Development Process: Lehrach, H.

• Integrating Personalized Medicine in the Canadian Environment: Efforts Facilitating Oncology Clinical Research: Syme, R.; Carleton, B.; Leyens, L.; Riches, E.

• Charting the Course for a New Cancer Clinical Research Culture: Teijpar, S.; Salgado, R. on behalf of the Working Groups ‘Biomarker’ and ‘Clinical Trial Framework’ of the European Alliance for Personalised Medicine (EAPM)


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Public Health Genomics

Public Health Genomics is the leading international journal focusing on the timely translation of genome-based knowledge and technologies into public health, health policies, and healthcare as a whole. This peer-reviewed journal is a bimonthly forum featuring original papers, reviews, short communications, and policy statements. It is supplemented by topic-specific issues providing a comprehensive, holistic and ‘all-inclusive’ picture of the chosen subject. Multidisciplinary in scope, it combines theoretical and empirical work from a range of disciplines, notably public health, molecular and medical sciences, the humanities and social sciences. In so doing, it also takes into account rapid scientific advances from fields such as systems biology, microbiomics, epigenomics or information and communication technologies as well as the high potential of ‘big data’ for public health.

What was until very recently no more than a vision for a new era of public health, in which advances in the ‘-omic’ sciences would be integrated into strategies aiming at benefiting population health, has now become a response to the very pressing need for the development of effective personalized healthcare which is complementary to health protection and health promotion. The aim of Public Health Genomics is to facilitate a broad dialogue between academia, the private sector and government bodies.

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Selected contributions

• Ask Not What Personalized Medicine Can Do for You – Ask What You Can Do for Personalized Medicine: Budin-Ljøsne, I.; Harris, J.R. (Oslo)
• Racial Differences in Attitudes toward Personalized Medicine: Diaz, V.A.; Mainous III, A.G.; Gavin, J.K. (Charleston, S.C.); Wilson, D. (Orangeburg, S.C.)
• The General Public’s Understanding and Perception of Direct-to-Consumer Genetic Test Results: Leighton, J.W. (Glenaside, Pa./New Brunswick, N.J.); Valverde, K. (Glenaside, Pa.); Bernhardt, B.A. (Philadelphia, Pa.)
• Public and Biobank Participant Attitudes toward Genetic Research Participation and Data Sharing: Lemke, A.A.; Wolf, W.A.; Hebert-Beirne, J.; Smith, M.E. (Chicago, Ill.)
• Genetic Variants Associated with Complex Human Diseases Show Wide Variation across Multiple Populations: Adeyemo A.; Rotimi, C. (Bethesda, Md.)
Access for All:
A Personalised Approach

There is a recognised need to define barriers and develop putative solutions to the issue of patient access to personalised medicine across the EU. To meet this need, the Brussels-based European Alliance for Personalised Medicine (EAPM) assembled a multi-stakeholder panel to address the topics.

Certain key challenges form the basis for this issue of Public Health Genomics. The authors outline them and present potential solutions to help embed personalised medicine into national health care systems.

Part of the access problem concerns the EU’s directive on patients’ rights to cross-border care. The effectiveness of the directive depends on collaboration at the European level. Unfortunately that sort of collaboration is in very short supply – a deficiency that threatens to undermine what is a well-intentioned initiative. Meanwhile, challenging fiscal times and an ageing population of 500 million potential patients across the EU’s 28 Member States, are making health care in Europe more expensive than ever. People are living longer and will, in most cases, be treated for not just one but several ailments during their lifetime. As such, a publication such as this is a valuable resource for EU/national politicians, health care planners, patients, medical professionals, and researchers.