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Genomic Medicine VI
Executive Summary

The Sixth Genomic Medicine Centers Meeting: *Global Leaders in Genomic Medicine* was held on January 8-9, 2014, at the National Academy of Sciences Building in Washington, DC. This meeting focused on identifying areas of active translational and implementation research, potential common strategies, and opportunities for collaborative efforts in genomic medicine implementation internationally.

Active Translation and Implementation Projects (selected examples)

- Using specialized databases to access privacy-protected electronic health records (EHRs), allowing for novel clinical research studies (UK, Estonia, Kuwait, others)
- Using whole genome sequencing to map 100,000 patients' genomes to target variants for rare diseases, cancer, and pathogens; generating technology "bake-off" competitions and sharing information and methods (UK)
- Creating national framework for clinical exome sequencing (Belgium)
- Pilot testing *TGFB1* testing for risk detection and disease diagnosis in stromal corneal dystrophies (Singapore)
- Developing population-specific genotyping array from sequencing 5K individuals and applying it 500K adults with linked EHR data (Estonia)
- Examining population-specific traits and developing population-specific reference genomes (Korea)
- Pilot testing a pharmacogenomics (PGx) card to prevent Stevens-Johnson Syndrome in susceptible individuals and assess cost-effectiveness (Thailand)
  Reporting family history data of first degree relatives into EHRs without revealing index cases' identity (Israel)

Potential Common Strategies and Collaborations

- Develop an evidence base and identify variants important for clinical implementation internationally, perhaps building on efforts such as ClinVar and ClinGen
- Expand NIH's Inter-Society Coordinating Committee on Practitioner Education in Genomics to involve international societies
- Establish international next generation sequencing (NGS) diagnostic sequencing guidelines and standards rather than multiple individual nations' guidelines
- Engage with other global initiatives for data sharing such as GA4GH and the International Rare Disease Research Consortium (IRDiRC) to synergize efforts and avoid wasteful duplication
- Capitalize on ability of small or developing countries to be nimble and creative in developing new genomic medicine programs; ensure international collaborations encourage rather than impede this
- Consider potential for international pilot projects to join established NIH networks such as eMERGE and IGNITE as affiliate members

- Develop international collaborative pilot programs for the European Commission's 2014 solicitation for personalized medicine pilot programs
- Map ongoing pilot projects to determine best candidates for international pilot demonstration projects; could be hosted by EuroBioForum Observatory on Personalized Medicine (http://www.eurobioforum.eu/2028/observatory/) or IGNITE consortium
- Augment international cooperation through meta-data sharing and harmonization of both policy and regulation, helping foster agreement on what constitutes evidence that a variant as clinically actionable
- Circulate GMVI presentations to augment public awareness of genomic medicine developments
- Create laboratory reference samples with ample representation of diverse ancestries
- Develop or participate in a global Exome Variant Server, especially if linked to simple non-identifying phenotypes
- Foster both "push" from financially-empowered policymakers and grassroots "pull" from clinicians and patients to move implementation forward

Break-Out Groups' Major Ideas for Genomic Medicine Action

- Bioinformatics/IT: Identify key elements to be stored in EHRs; create a global resource for actionable variants
- Education: Define professional genomics and other healthcare providers' workforce needs; develop novel educational tools for sharing amongst meeting members
- Evidence: Identify ongoing evidence generation projects, develop systems to both capture and share evidence for the utility of genomic medicine implementation
- Pharmacogenomics: Consider a signature initiative for global eradication of Stevens-Johnson Syndrome through the use of Pharmacogenomics ID cards; encourage addition of pharmacogenetic components to induced pluripotential stem cell initiatives to promote basic mechanistic science
- Policy: Pursue engaging stakeholders, data sharing, and regulatory oversight with other ongoing efforts; improve capacity and encourage inclusion of economic assessments and cost/benefit analyses of genomic medicine

Immediate Next Steps

The five break-out groups will comprise the initial working groups (WGs) for this effort, along with a sixth on Communications. Participants will nominate members and potential chairs for each of these groups along with an over-arching steering committee. Slide presentations and videos will be posted on the meeting website. Teri and Geoff will draft and distribute a meeting summary and executive summary for comment, and a draft white paper with the presenters and break-out group leaders as co-authors.

Summary of Action Items

1) Teri Manolio (USA) and Paul Lasko (Canada) will discuss the possibility of expansion of NHGRI's Inter-Society Coordinating Committee (ISCC) to involve international agencies.
2) GMVI attendees will reach out to their connections with Global Alliance for Genomic Health to promote GMVI efforts.
3) Wasun Chantratita (Thailand) will share publications on SJS which list potential predisposing factors within specific gene regions with GMVI attendees.
4) Warwick Anderson (Australia) will share a link to a framework for translating "-omics based" discoveries into clinical care.
5) GMVI attendees should identify important groups or individuals who were unable to attend this meeting but may be interested in Genomic Medicine to participate in future meetings through an email to Geoff Ginsburg. The Communications WG will play a key role in reaching out to such individuals.
6) Potentially, the GMVI break-out groups may write reports for a special issue of *Pers Med* in collaboration with George Patrinos and the Genomic Alliance.
7) NHGRI will post the video recording of this presentation on its website and distribute a meeting summary and an executive summary. Attendees who presented and breakout leaders will be authors on a white paper of this meeting. BoG leaders who decide to write journal articles of their BoG's meetings are encouraged to move forward with this process.
8) Any attendee who would like to volunteer for WG leadership or membership will email Geoff and Teri. The Steering Group and WGs will be convened once members have been identified.
9) The full committee will consider a follow-up meeting.
10) GMVI attendees will contact Paul Lasko (Canada) if interested in attending an upcoming Canadian meeting on genomic medicine in April.
11) The full committee decided that in order to establish more formal relationships with European organizations that that address personalized medicine, group members should send relevant information to Geoff and Teri.
12) Attendees interested in receiving Eric Green's (USA) monthly newsletter *The Genomics Landscape* which monitors genomic medicine events and milestones across the NIH will contact Eric Green (egreen@nhgri.nih.gov).