Day 1 Summary – International Projects

- Database for accessing anonymized EMR (UK, Estonia)
- Bake-off (comparison) projects—way to share and build upon (UK)
- Genotyping centers (UK, Belgium, France) vs deployed within existing framework (Singapore)
- Critical importance of depositing data (Genomic Medicine Alliance, GA4GH)
- NGS guidelines for diagnostic sequencing—US, UK, Netherlands?
- Reach out to GA4GH and IRDiRC
Day 1 Summary – International Projects

- Population-specific traits: potential for broader insights, “experiments of nature” (Korea)
- Population specific reference genome (Korea)—intermediate strategy leading to “population graph” representation
- Pharmacogenomics card for SJS/TEN drugs (Thailand)
- Challenges of mixed ancestry populations and potential stratification (Israel)
- Pushing FHx data of first degree relatives into their EMR without revealing index cases (Israel)
- Importance of negative studies in changing practice (Israel)
Day 1 Summary – Panel Discussion

- Europ Comm recent solicitation open on piloting roll-out of personalized medicine (I Norstedt)
- Enhance data sharing through meta-data sharing (V Dzau)
- Harmonize policy and regulation (V Dzau)
- Agree on what we will consider as evidence that variant is actionable (A Chakravarti, A Kolbe)
- Need case studies (A Chakravarti)
- Need map of ongoing pilot projects – EC-funded observatory on personalized medicine projects (I Norstedt)
Day 1 Summary - Panel Discussion

- How to make GM6 ppts aware of and engaged in ongoing efforts (P Tan)

- Reference samples for genotyping – “Genome in Bottle” effort in US

- Some kind of global Exome Variant Server just showing variation across $10^6$ human genomes

- Focus on best implementation pilot(s) using implementation science methods

- Move beyond talking (M Ulfendahl, A Kolbe)

- Generate demand from policy makers (pull) and involve them from beginning, along with push from clinicians and grassroots efforts from patients
Day 1 Summary – International Projects

- NHMRC Framework translation of omics into care – link to be sent (W Anderson)
- Pers Electronic Health Record can accept genomic data – talk with IT group! (W Anderson)
- To realize Genomic Medicine, existing programs will be integrated to implement a Multi-component Project for public benefit (Japan)
  - With both disease-oriented and population-based approaches
  - Supported with large-scale genomic research data including database of genomic variation
  - With clinical research strengthened by use of highly-specialized hospital functions
Products – Working Groups

- IT/Bioinformatics
- Education/workforce
- Pharmacogenomics
- Evidence
- Policy

Leadership must be multinational… need volunteers!

What did we miss?
Products – International Steering Group

- Monitor working groups
- Identify directions
- Facilitate communications and interchange
  - Observatory, website, materials warehouse
  - Information clearinghouse
  - Push notices and news releases; newsletter
  - Repeat meeting?
  - Liaise with other relevant groups
  - Identify other key members
- Communications working group
Products – Working Groups

- IT/bioinformatics
- Education/workforce
- Pharmacogenomics
- Evidence
- Policy
- Communications
Five Working Groups Top Ideas

- **IT**: Define key elements to be stored in EMR
- **IT**: Global resource for actionable variants
- **Educ**: Define workforce needs
- **Educ**: Develop existing/new educational tools that can be widely shared
- **Evid**: Develop systems to capture evidence—federated network, standardized APIs (e-tools)
- **Evid**: Identify poolable/extendable projects
- **PGx**: Global eradication of SJS/TEN
- **PGx**: PGx card
- **Policy**: economics, cost-assessment
Day 2 Summary – International Projects

• Global Medicine Alliance: already begun many of components identified here, developing countries potentially more nimble

• A few break-out group reports for *Pers Med* special issue? (GMA)

• Sequencing/genotypic data will likely not replace phenotypic measures, will be used in combination (Newborn Sequencing)

• Regulatory issues in using new technology in different setting (NBS)
Day 2 Summary – International Projects

- Press releases in US have implications overseas, how to improve communications (NBS)
- Potential for international pilot projects to join networks as affiliate members (IGNITE)
- Bringing together international guidelines
Day 2 Summary – Next Steps

- Post slides, presentations on NHGRI site
- Draft summary and executive summary, distribute, comment
- Draft white paper: presenters, break-out group leaders
- Volunteer for working group leadership and membership
- Convene steering group and working groups
- Consider follow-up meeting
- Pursue EC observatory/catalog
- Identify other members
- Ensure global accountability
IT/Bioinformatics/CDS Top Ideas

- Define key elements to be stored in EMR
- Identify most robust and generalizable solutions for potential wider adoption (e.g., CDS, variant databases, informatics pipelines)
- 10 Global resource for actionable clinical variants
- Collection/aggregation of variant/phenotype associations
- Define necessary federated databases to implement genomic medicine
- Phenotype ontology (including inventory)
- Clearinghouse of implementation guidelines
Education/Workforce Top Ideas

- Three types: genomics professionals, other healthcare providers, public
- Genomics professionals:
  - Collect data on genomic professional workforce and training in different countries
  - Conduct formal workforce studies
  - Share competencies and training paradigms
  - Genomics academy
Education/Workforce Top Ideas

- Other healthcare providers:
  - Deploy new educational tools
  - Develop region-specific teaching materials, perhaps common templates
  - Use existing professional workforce to educate other physicians/providers

- Public:
  - Provide clearinghouse for information
  - Consider novel educational paradigms
  - Customize culturally-specific materials
  - Extend DNA Day to be international educational event
Evidence Generation Top Ideas

• Evidence epistemology (pre-evidence)
  • Catalog evidence generating projects
  • Standards for a test
  • Definition of evidence
  • Encourage adoption of genomic medicine applications with sufficient existing evidence

• Opportunities for action
  • Identify countries/systems willing to enable patient data sharing
  • Develop systems to capture evidence—federated network, standardized APIs (e-tools)

• Next steps: discuss areas of overlap with others particularly poolable or extendable projects
Pharmacogenomics Top Ideas

• Endorse desire for quality of evidence base for PGx implementation

• Emphasize cheap drugs with treatment failure or extreme ADRs (metformin, vaccines)

• Add drug/PGx component to all iPS initiatives (lack of basic mechanistic science)

• Global effort to develop value around cancer NGS-- expensive test generates use of very expensive drugs

• Global eradication of SJS/TEN via PGx–systematic approach
Policy Top Ideas

• Engaging stakeholders
• Data sharing
  • Privacy
  • Informed consent
  • Legal issues
• Regulatory oversight
  • Map activities and issues addressed, gap analysis
  • Encourage network of networks (consent, data-sharing groups, etc)
• Cost benefit assessments– improve capacity for doing them; engage payers
Day 1 Summary – Additional Points

• Develop global ClinGen resource of actionable variants
• Pilot implementation projects
• Policy – standardized informed consent (model)
• Global EMR phenotype standards – not impossible
• What about children?