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
- Pharmac
- Evidence
- Policy

What did we miss?

Leadership must be multinational… need volunteers!

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
  - Defnition 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?