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 be continued…

Products – Working Groups

- IT/Bioinformatics

- Education/workforce

- Pharmac

- 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

- <u>6 IT</u>: Define key elements to be stored in EMR

- <u>IT</u>: Global resource for actionable variants

- <u>3 Educ</u>: Define workforce needs

- <u>Educ</u>: Develop existing/new educational tools that can be widely shared

- <u>2 Evid</u>: Develop systems to capture evidence–federated network, standardized APIs (e-tools)

- <u>Evid</u>: Identify poolable/extendable projects

- <u>4 PGx</u>: Global eradication of SJS/TEN

- <u>PGx</u>: PGx card

- <u>7 Policy</u>: economics, cost-assessment