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
- Pharmacogenomics
- Evidence
- Policy

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
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- Evidence
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- Communications
Five Working Groups Top Ideas

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