Global Genomic
Medicine Collaborative

Breakout Group 6

Translation / clinical impact

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International Cohorts Summit

Global Genomic Medicine Collaborative

Breakout Group 6: Translation / clinical impact

Discussion question 1: What are the opportunities for translation of cohort findings to improved clinical care and population health?

Advance medical practice

- Change diagnosis, prognosis, therapeutic decision making
- Mendelian disease: facilitate genomic interpretation; expand variant catalogs across multiple ancestries and include phenotypes
- Pharmacogenomics
- Genetic risk scores

Drug development

- Find new targets, validate existing targets, find the right patients for study

Generic opportunities

- Public understanding/increase health literacy
- Develop exemplars for teaching (e.g. human knockouts)
- Evidence generation: e.g. exploit EHRs and associated expertise to do large simple trials: within the context of the “learning healthcare system”

Population health and policy

- New knowledge → policy (precedents of smoking, trans-fats)
- e.g. Premarital counselling to avoid recessive disease

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Discussion question 2: What are the major barriers to clinical and population health translation and how can they be dealt with?

- **Variable healthcare delivery systems**
- **Disparities at many levels**
- **Lack of diversity in present datasets**
- **Lack of evidence for clinical utility**
- **"The handoff" – from data/evidence generation to implementation**
- **Potential regulatory barriers (e.g. regulation of CDS)**
- **Behavioral change is hard; behaviors are major sources of error and confounding**
- **Reimbursement, clinical costs**
- **Privacy regulations vary across countries**
- **Ethics approval; varying consent mechanisms**
- **Academic territorialism**
- **Barriers (perceived and real) around engaging industry**

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Other comments / discussion points

Need to identify exemplar projects that could benefit each member of a 'cohort of cohorts' -- suggestions:

1. **International human knockout project.** Create a catalog that includes other capabilities (e.g. recontact; EHR-based phenotyping; basic biology, including actually establishing that a gene is knocked out). **Opportunity/Barrier**: who pays for the catalog, recall, etc.
2. **Standardize implementation of return of results (general and local factors).** Share approaches to communication of risk information. Capture outcomes Maybe start with FH or cancer.
3. **Country (cohort)-specific risk prediction using standardized methodology.** Start with cancer and cardiovascular disease. Assess GRS and non-genetic risk factors in the same framework across ancestries (Kadoorie, UKB, Mexico City …). could share summary data initially, then individual level data …