The GenomeConnect Patient Portal: Patients as Partners in Advancing Genomic Knowledge

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BACKGROUND

Genomic Interpretation
- Technological advances allow for identification of an increasing number of genomic variants.
- Process of interpreting variants remains complex especially if variants are novel or rare.

The Clinical Genome Resource (ClinGen)
- A resource funded by the NIH.
- Aims to create centralized resource to define the clinical relevance of genes and variants.
- One effort involves encouraging laboratories, clinicians, researchers, and patients to share genetic and health data (Figure 1).

Data Sharing
- Sharing genotype and phenotype data aids in variant interpretation.
- Many laboratories are now actively contributing genotype data to public databases.
- Laboratories often do not have detailed phenotype information associated with variants.
- Access to detailed phenotype information about the patients in whom particular variants were found would facilitate the interpretation process.
- Patients themselves are a logical source of phenotype information.

GenomeConnect, a web-based patient portal, aims to:
- Provide a mechanism for patient-centric data sharing
- Support patients as a valuable partner in advancing genomic medicine

Figure 1: Sources of Genetic and Health Information to aid in Variant Interpretation

GENOMECONNECT

Open to anyone who has had genetic testing regardless of test results or diagnosis

Figure 2: Process of GenomeConnect Participation

Patient-Entered Phenotype Information
- Survey collects detailed health history
- Patient-friendly language mapped to HPO terms
- Additional surveys assigned based on initial health survey answers

Curated Variant Information
- Patients upload genetic testing reports
- Genetic counselors curate reports and extract variant information

Ability to Re-contact Participants
- Provide additional surveys
- Match participants with one another
- Provide information about research opportunities

GENOMECONNECT PARTICIPATION TO DATE

Figure 3: GenomeConnect Enrollment (October 2014-January 2016)

GENOMECONNECT PARTICIPATION

Genetic Testing

Figure 4: Types of Genetic Testing Participants Report

- Majority have had testing due to symptoms they experienced (50%) or to confirm a diagnosis (25%)

Referral Sources

Figure 5: Referral Sources for GenomeConnect

- 22% of participants are members of patient groups
- Only 5% report being referred to GenomeConnect by such a group

CONCLUSIONS AND FUTURE DIRECTIONS

Benefits of GenomeConnect
- GenomeConnect brings value to all involved parties:
  - Detailed phenotype and genotype information aids in variant interpretation
  - Connects individuals, laboratories, and researchers

Future Efforts
- Increase participation and engagement in GenomeConnect
- Partner with patient advocacy registries to increase their sharing of genotype and phenotype data

Clinical Genome (ClinGen) Resource
More Info: www.clinicalgenome.org
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