NHGRI Published New Vision for Genomics
Genomics Reaches the Clinic: From Basic Discoveries to Clinical Impact

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Today, more than ever, basic science research provides significant opportunities to advance our understanding about the genetic basis of human disease. Close interactions among laboratory, computational, and clinical research communities will be crucial to ensure that genomic discoveries advance medical science and, ultimately, improve human health.
Imperatives for genomic medicine

Opportunities for genomic medicine will come from simultaneously acquiring functional knowledge of genome function, insights into disease biology and powerful genomic tools. The following imperatives will capitalize on these opportunities in the coming decade.

**Making genomics-based diagnostics routine.** Genomic technology development so far has been driven by the research market. In the next decade, technology advances could enable a clinician to acquire a complete genomic diagnostic panel (including genomic, epigenomic, transcriptomic and microbiomic analyses) as routinely as a blood chemistry panel.

**Defining the genetic components of disease.** All diseases involve a genetic component. Genome sequencing could be used to determine the genetic variation underlying the full spectrum of diseases, from rare Mendelian to common complex disorders, through the study of upwards of a million patients; efforts should be directed towards organizing the necessary sample collections.

**Comprehensive characterization of cancer genomes.** A comprehensive genomic view of all cancers will reveal molecular taxonomies and altered pathways for each cancer subtype. Such information should lead to more robust diagnostic and therapeutic strategies for cancer care.

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**Practical systems for clinical genomic informatics.** Thousands of genomic variants associated with disease risk and treatment response are known and many more will be discovered. New models for capturing and displaying these variants and their phenotypic consequences should be developed and incorporated into practical systems that make information available to patients and their healthcare providers, so that they can interpret and re-interpret the data as knowledge evolves.

The role of the human microbiome in health and disease. Many diseases are influenced by the microbial communities that inhabit our bodies (the microbiome). Recent initiatives (http://www.human-microbiome.org) are using new sequencing technologies to catalogue the resident microflora at distinct body sites, and studying correlations between specific diseases and the composition of the microbiome. More extensive studies are needed to build on these first revelations and to investigate approaches for manipulating the microbiome as a new therapeutic approach.
Collaboration between NHGRI & Wellcome Trust

Goal: Consider processes, databases, and other resources needed to:

- Identify clinically relevant variants
- Decide whether they are actionable and what the action should be
- Provide information for clinical use
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