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Commentary

Sequencing of Personal Genomics and it's Benefits

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ABSTRACT

Personal genomics, also known as consumer genetics, is the branch of genomics concerned with the sequencing, analysis, and interpretation of an individual's genome. During the genotyping stage, various techniques are used, such as Single-Nucleotide Polymorphism (SNP) analysis chips (typically 0.02% of the genome) or partial or full genome sequencing. Once the genotypes are determined, the individual's variations can be compared to the published literature to determine trait expression likelihood, ancestry inference, and disease risk. Personal genomics, in its most basic form, is the analysis and interpretation of information from an individual's genome. In this chapter, we broaden the definition by emphasizing three key differences between personal genomics and traditional clinical genetic testing.

Keywords: Genome; Personal genomics; Genetic testing; Tumour

DESCRIPTION

In this we have to broaden the definition by emphasizing three key differences between personal genomics and traditional clinical genetic testing. First, personal genomics genetic information is derived from across the genome rather than from a single position, gene, or genomic region. Second, whereas traditional genetic testing is triggered by specific circumstances, such as an individual undergoing a diagnostic work-up or a family history of disease, personal genomic testing occurs with or without such specific circumstances. Third, personal genomics eliminates the middleman that is typically involved in traditional testing, providing a person with direct access to his or her own genomic information. These three elements are inextricably linked. Because the information is genome-wide, some of it may have clinical utility, some may only have personal utility, and some, at least for the time being, may have no utility at all. As a result, testing does not have to be tied to a specific application. Furthermore, the individual undergoing the testing derives personal value from the vast amount of information provided.

Sequencing personal genomes

The technology that enabled the Human Genome Project is becoming more affordable, and as a result, genetic analysis is becoming more accessible to a larger population. Hundreds of sequencing machines worked for years to complete the first human genome sequence. A single machine can now sequence the entire human genome in a matter of days. In 2016, the cost of sequencing a human genome was around $1,000 (US), and companies are still competing to lower the cost. Clinical applications for genome sequencing have already emerged, most notably in the diagnosis of rare childhood conditions and informing cancer therapeutics.

Genome-wide information

Personal genomic information can be genotypes for thousands or millions of SNPs, exon sequences (the "exome"), or a whole genome sequence. Personal genomic information can be expanded to include RNA, tumour DNA, epigenetic, or human microbiome data, but the focus here is on non-tumor DNA data. Technological advances have reduced the cost of genome-wide testing; today, one can obtain genotype data for over one million SNPs for less than the cost of a single genetic test. At the same time, the cost of a full genome sequence is rapidly declining. As the costs of genotyping and sequencing decrease, testing for a single SNP will become less economical.

Although it is becoming more common in consumer-directed genetic testing, genome-wide genetic testing is still uncommon in clinical medicine. Clinicians who use genetic testing tend to

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concentrate on one or a few tests with proven clinical utility that are relevant to their specific area of expertise or professional service. Any large amount of genetic data poses significant challenges to physicians and other healthcare providers.

The value of personal genetic information varies across the genome and from person to person. Its usefulness varies over time for each individual. A wide range of clinical and personal utility is the foundation for a diverse set of reasons for an individual to obtain personal genomic testing.

Personal genomic information direct access

As genome-wide testing becomes more common in clinical care, it is likely that individuals and their physicians will receive their personal genomic information at the same time, via a centralized system. A growing number of genetic tests are available to look for known genetic mutations linked to specific diseases. Genetic tests typically characterize only one gene (or specific parts of one gene), and their availability is dependent on scientists' ability to link well-characterized diseases to specific genes. These tests have proven to be relatively simple for conditions with specific genetic causes, such as Huntington's disease or cystic fibrosis. In contrast, progress in predicting a person's risk for complex and multifactorial diseases like diabetes and heart disease has been more difficult.

Other benefits

In 2018, police arrested Joseph James De Angelo, the Golden State Killer or East Area Rapist, and William Earl Talbott II, the prime suspect in the 1987 murders of Jay Cook and Tanya Van Cuylenborg. These arrests were based on private genomics uploaded to GED match, an open-source database that allowed investigators to compare DNA recovered from crime scenes to DNA uploaded to the database by suspect relatives. Family Tree DNA changed its terms of service in December 2018 to allow law enforcement to use their service to identify suspects of "a violent crime" or to identify the remains of victims. The company confirmed that it was collaborating with the FBI on at least a few cases, indicating that GED match was no longer the only one doing so. Using the same method, nearly 50 people suspected of assault, rape or murder has been apprehended since then. Using GED match, personal genomics has also enabled investigators to identify previously unknown bodies.