Public Health England

Genomics overview

Public Health England drives forward infectious disease genomics

At the recent celebration of the NHS's 65th birthday, the Secretary of State for Health announced that the government's major drive to introduce high-tech DNA mapping for patients will start with cancer, rare diseases and infectious diseases. Prior to this announcement the Prime Minister had announced that the personal DNA code (known as a genome) of up to 100,000 patients or infections in patients will be decoded, or sequenced, over the next five years – "The 100,000 Genome Project". The primary purpose of this is to improve understanding of disease, leading to better and earlier diagnosis and personalised healthcare. Based on expert scientific advice, the Department of Health (DH) has initially prioritised sequencing of lung and paediatric cancer, rare diseases and infectious diseases.

What is a genome?

The word 'genome' is used to describe all the hereditary information of an organism (including humans, animals, plants and infectious 'bugs'). The hereditary information is coded within DNA which is inherited by children from their parents and is why children share traits with their parents, such as skin, hair and eye colour.

Similarly bacteria and viruses causing infectious disease pass their DNA from one generation to another so we use this to understand how different bacteria are related and whether they are the same strain. We can also tell whether they carry genes for antimicrobial resistance or other properties of the organism.

DNA is in each cell of an organism and is a long chain of millions or even billions of 'nucleotides'. Nucleotides can be one of four different molecules represented by A, C, T or G. Different patterns in the sequence of these four molecules define the differences between species and the members of those species.

Modern day DNA sequencing

Determining the sequence (or order) of nucleotides in DNA from any organism has been very difficult to do. The first complete human genome sequence was reported in 2001. This required collaboration between expert centres across the globe, and it took 13 years to complete and cost hundreds of millions of dollars. However, recent developments in DNA sequencing technology have produced a revolutionary increase

in the number and speed at which genomes can be sequenced and with a massive reduction in cost. This means that there is the opportunity to use whole genome sequencing as a key method in identifying and comparing bacteria and viruses causing disease in humans and animals.

Infectious disease sequencing in Public Health England

The work being undertaken by Public Health England in collaboration with the NHS and academia, follows recommendations to invest in genomics for the study of infectious disease by Professor Sally Davies, Chief Medical Officer, who said:

"By putting firm foundations in place through Genomics England, this technology will let us make ground-breaking discoveries about how diseases work, who could be susceptible to them, how we can treat them and what treatments might work. Earlier diagnoses will help to reduce uncertainty and stress for patients and families involved."

Part of the investment by Public Health England has been to establish a state of the art, high-throughput sequencing capability at the PHE Colindale site. This is to provide the 'PHE Pathogen Genomics Service' which is soon to be launched. In order to deliver this service, PHE has also invested in recruitment of highly qualified and trained laboratory and bioinformatics staff to deliver and provide analysis of the data. PHE is now developing methods for:

- rapid identification of bacterial and viral pathogens (organisms causing disease)
- comparing pathogens at a forensic level so that we can understand their relationship (i.e. linked in an outbreak or emergence of new strains)
- being able to predict whether a particular strain is going to be more infectious or cause more serious disease

Next Generation Sequencing (NGS) can potentially do all of this in one process, whereas several different methods would have traditionally been required and, most importantly, offers a faster service for the patient and a more cost effective process for the taxpayer. NGS is changing the way we investigate outbreaks of infectious disease. Any significant outbreak is now almost certainly to be investigated using NGS which will provide a rapid response with a high level of detail and this, combined with good epidemiological information, is a powerful tool.

Further information:

www.gov.uk/government/news/dna-mapping-to-better-understand-cancer-rare-diseases-and-infectious-diseases

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