

Genetic testing

Science fiction is becoming science fact. Mr Sergey Brin, co-founder of Google, was recently in the news when a scan done by 23andMe (founded by his wife) revealed that he had the G2019S mutation, associated with an increased risk of Parkinson's disease, as well as a family history of Parkinson's disease. Genetic profiles of single nucleotide polymorphisms (SNPs) can also be obtained from deCODE genetics, an Icelandic company.

A number of genetic tests can now be made before drugs are prescribed. Cetuximab is an epidermal growth factor receptor (EGFR) antagonist presently used in head, neck and colorectal cancer. The prescribing of trastuzumab requires identification of human EGFR-2 tyrosine kinase receptor over-expression. Identification of functional *KRAS* and EGFR over-expression is required for panitumumab prescription for advanced colorectal cancer.

The presence of the BCR-ABL translocation (or Philadelphia chromosome) is a diagnostic feature of chronic myelogenous leukaemia. Genotyping for HLA-B*5701 establishes a possibly lethal hypersensitivity to abacavir (a nucleoside analogue reverse transcriptase inhibitor). Sexual health clinics are using nucleic acid amplification tests to detect the presence of *Chlamydia*. The disease risk associated with mutations is often poorly quantified; however over 80 health-related genetic tests are already available direct to the consumer in the US [1].

1. Goddard KA, Public Health Genomics. 2009;12:92-104.

Scientists map genome for two cancers

Research published in *Nature* on 16 December 2009 has identified all mutations in melanoma and lung cancer cells, which has allowed them to produce the first complete cancer genomes. The researchers found more than 23,000 mutations in the lung tumour and more than 33,000 in the melanoma.

Professor Mike Stratton of Wellcome's Sanger Institute's cancer genome project said the aim now was to produce genetic maps of all types of cancer. There are more than 100 cancers in all, and each genome mapping process requires several months of work and costs tens of thousands of euros.

The first 50 cancers are to be mapped by scientists in the International Cancer Genome Consortium, launched in 2008, which includes the US National Institutes of Health and groups from Australia, Canada, China, France, India, Japan and Singapore. Separately, Dr Elizabeth Rapley of the Institute of Cancer Research said she expects that "similar insights into other cancer genomes are likely to follow... As more cancer genomes are revealed by this technique, we will gain a greater understanding of how cancer is caused and develops, improving our ability to prevent, treat and cure cancer".

doi:10.1038/nature08629/08658

Licence extensions for trastuzumab and abatacept

On 18 December 2009 the EMEA's Committee for Medicinal Products for Human Use adopted a positive opinion on an extension of licence for trastuzumab (Herceptin) to include the treatment of patients with HER2 positive metastatic adenocarcinoma of the stomach or gastro-oesophageal junction who have not received prior anticancer treatment for their metastatic disease in combination with capecitabine or 5FU and cisplatin.

It also issued a positive opinion on licence extension for abatacept (Orencia), to include polyarticular juvenile idiopathic arthritis in paediatric patients from six years of age who have had an insufficient response to other DMARDs including at least one TNF blocker.

NPSA third quarter data summary on patient safety incident reports 2009

The UK National Patient Safety Agency (NPSA) has published the latest quarterly data summary analysing patient safety incident reports received by the National Reporting and Learning Service between October 2003 and the end of September 2009. This includes details of the volume of incident reports received, the sector they are from, the type of incidents and the level of harm caused. Healthcare leaders are invited to use the report as a tool to improve patient safety by comparing the data with data reported within local organisations.

The theme of this report is "Learning from reporting". It looks at how and why we need to improve the quality of data reported. In September 2009, the National Reporting and Learning Service published the first edition of the Data Quality Standards. This report provides an outline of the standards and highlights actions to take to implement them. So make 'improving patient safety' your pharmaceutical new year's resolution!

www.nrls.npsa.nhs.uk/resources