

# Five ways of making diagnoses and targeting treatment



## 1 Refining diagnoses and understanding the basis of diseases

Genomic information can assist in making a diagnosis or identifying a subtype of a condition. For instance, new laboratory techniques have allowed us to understand why some children develop dysmorphic syndromes, and to identify subtypes of breast and other cancers.

Genetic factors influence the susceptibility to common conditions such as bipolar disorder, coronary artery disease, Crohn's disease, rheumatoid arthritis, and types 1 and 2 diabetes. These factors are being identified through research studies involving thousands of patients.

## 2 Individualising clinical care

New medicines tailored for patients with a particular genetic make-up are becoming available. A well-known example is breast cancer, where molecular testing is used to subtype the cancer and then choose the best treatment option. Targeted drugs are also available for some types of breast, colorectal and lung cancer, leukaemia and melanoma. But cancer is not the only disease where genomics is making a difference: better vaccines are being produced, including those that treat a disease rather than just preventing it.

## 3 Predicting the effect of drugs

Research is starting to provide the basis for predicting individual responses to an increasing range of drugs. Genomic information is used to predict which patients will develop adverse reactions to azathioprine, carbamazepine and abacavir (a drug used against the HIV virus), or those who may need the dose varying (for instance, warfarin).

## 4 Diagnosing infections and tracking epidemics

Examining the genes of infective organisms allows rapid diagnosis and tracking of infections. Examples include tracking the bird flu virus, antibiotic-resistant infections (such as tuberculosis and MRSA), and E coli food poisoning.

## 5 Developing targeted therapies

Researchers have designed molecules that stop or alter the effects of alterations in the genome. Examples include compounds to treat chronic myelogenous leukaemia, breast, colorectal and lung cancer, and melanoma.

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