# New research areas in personalised medicines

1. There are new research areas driving personalised medicine forward:

## Molecular genetics

Current research is telling us a lot about diseases at the cell and molecular level. There is evidence that specific changes to genes (variations) can affect the way cells function and how a disease may develop. Diseases with similar symptoms may be diagnosed as the same disease. However, they may be caused by different genetic variations.

## Epigenetics

This growing area helps us to understand variation in disease. Epigenetics is the study of how genes can be turned on or off, or modulated (turned up or down) without changes to their DNA sequence. Epigenetic changes may influence how a patient responds to **treatment**, since the changes can occur in response to environmental or lifestyle factors, such as UV light exposure, diet, smoking, or stress. Ultimately, a personalised medicine would take account of an individual's epigenetics.

## Biomarkers and medicines development

Processes that happen at the level of cells and molecules can be measured using 'biomarkers'. Some examples of biomarkers are:

- Physiological measures such as blood pressure or

temperature
- biological substances (‘biochemicals’), such as enzymes or hormones
- gene changes
- images from Magnetic Resonance Imaging (MRI)

To create targeted treatments, biomarkers are increasingly used in medicines development. This is expected to:

- improve outcomes for patients: doctors should able to choose medicines that work well for each individual, and with a lower risk of serious side effects
- improve the efficiency of medicines development, making non-clinical and clinical trials more efficient, less time consuming and safer

# Pharmacogenetics or pharmacogenomics

One type of biomarker that is becoming widely used is an individual’s genetic or genomic information. The study of how genetics and genomics affect an individual’s response to a treatment is known as pharmacogenetics and pharmacogenomics. These help to “tailor a treatment” according to an individual’s genetic configuration. A number of medicines on the market today require a genetic test before the medicine is prescribed, to ensure the treatment is safe for the individual patient. For example:

- HIV patients are tested for a genetic variation known as ‘HLA B*5701’ before the medicine abacavir is prescribed because the variation is associated with an adverse reaction to the medicine.

In addition, the information provided with some other medicines on the market advises doctors how to use a patient’s genomics information when they prescribe the medicine. Genomics information can help to decide if that medicine is

the best option for that patient, and what would be the best dose. For example:

Trastuzumab (Herceptin) is gene-targeted monoclonal antibody directed against the Human Epidermal growth factor Receptor-2 (HER-2) and approved for the treatment of early-stage breast cancer. HER-2 is overexpressed in approximately 20 % of breast cancers, causing an excessive signaling to the cell interior that drives the breast cancer cells to grow faster and faster. Only patients who have tested positive for high HER-2 levels will benefit from treatment with trastuzumab. HER-2 exists on the surface of some breast cancer cells and is routed in the cell membrane. Trastuzumab works by attaching itself to the HER-2 on the surface of breast cancer cells and blocking them from receiving growth signals. By blocking the signals, trastuzumab can slow or stop the growth of the breast cancer and is an example of an immune targeted therapy.

# Biobanks

Studies that use biobanks are especially important for the development of personalised medicine, and biobanks are increasingly used in clinical trials for new medicines. Biobanks are basically large, organised sets of blood and/or tissue samples donated by patients and healthy volunteers. They also include carefully collected data on the donors' clinical condition, lifestyle (diet, smoking, etc.) and other factors. Biobanks allow the cells and molecules of large numbers of samples to be studied, and for this information to be **linked** with clinical and other data. Combining information in this way is helping us understand why individuals vary:

- in which diseases they develop
- in how severe their diseases are, and
- in how they respond to treatment.

The more samples available, the more effective such studies

can be. Biobanks are being set up in many countries. ‘EuroBioBank’ is an example of biobanks from different countries being linked, to make even more data available for research (in this case, into rare diseases).

# References


1. How Herceptin affects breast cancer cells” by **beyondthedish.wordpress.com** is licensed under a Creative Commons Attribution-NonCommercial-NoDerivs 3.0 Unported License.
   See: https://beyondthedish.wordpress.com/2012/06/04/smart-bomb-successfully-treat-advanced-breast-cancer-in-clinical-trials/


# Attachments