Learning Portal Lite: Pharmacogenomics

This is a one-page summary; see the full version online

This specially commissioned tutorial is to support the pharmacy workforce across all sectors who currently have **limited experience** working with pharmacogenomic information.

Remind me of the science

Refresh your memory of the basics with the NHS England Genomics Education website which has a range of excellent resources including short videos and a genomics glossary to help you get back up to speed quickly. BBC bitesize also has some useful content.

Genomics and the NHS

Genomics can play a key role in personalised and precision medicine and has a wide range of potential applications within healthcare. In England, 7 regional Genomic Medicine Service Alliances have been established to co-ordinate the implementation of genomics into clinical practice: the devolved nations have different models.

Taking and sequencing a sample

Biological samples for genomic testing are typically blood, saliva or tissue biopsy. Sequencing is used to 'read' the genomic information and may involve specific genes of interest or a whole genome. Familial hypercholesterolaemia is an example condition that may be identified with genomic screening.

Making decisions about medicines

Pharmacogenomics is concerned with the way in which an individual's genetic attributes affect their likely response to medicines. It can explain differences in efficacy and toxicity. An example is the variation in the activity of cytochrome p450 enzymes such as CYP2D6 which is responsible for metabolising codeine to its active metabolite morphine. However currently only 3 tests are listed in the National Genomic Test Directory (NGTD):

1. **DPYD gene –** to predict toxicity to systemic chemotherapy such as 5-fluorouracil
2. **Thiopurine methyltransferase (TPMT) and nudix hydrolase 15 (NUDT15) in patients with ALL –** to predict toxicity with 6-mercaptopurine
3. **Aminoglycoside exposure posing a risk to hearing (R65)**

Testing for the human leukocyte antigen variant HLA-B*57:01 in patients needing abacavir predated the creation of the NGTD, but is routinely done.

Information sources

If you are starting out try the product SmPC, look for an MHRA Drug Safety Update or the National Genomics Education Programme's GeNotes site. More advanced sources include the Clinical Pharmacogenetics Implementation Consortium.

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