# Pharmacogenetics

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# What is Pharmacogenetics?

- Scientists, physicians and the pharmaceutical industry are actively developing ways to customize medical treatments to suit our unique genetic signatures. The study of how genetic variations interface with drug response and disease risk is called pharmacogenetics.

# What is Pharmacogenetics?

- When physicians are given the tools to evaluate a patient's genetic make-up, they will be able to make more accurate diagnoses, and prescribe more efficient drug therapies with fewer adverse side effects.

- Differences between people extend beyond our outer physical features. How individuals respond to drugs for the treatment of cancer or other illnesses differs based on the activity and function of enzymes in the body. This information is available in each individual's genetic profile. Even today, genetics is being integrated into individuals' medical treatment plans.

# Example

- Purinethol is a common chemotherapy drug; it works by incorporating itself into rapidly dividing cancer cells and killing them.
- Most patients benefit from the drug, however treatment comes with a risk of severe, sometimes fatal side effects in certain people.
- In the 1990's scientists were able to discover why Purinethol affected some people badly.
- Because Purinethol is a toxic substance patients need to have enough TPMT enzymes in their body to inactivate Purinethol.
- 89% of people have full enzymatic activity and receive full dosage of Purinethol.
- 11% of people have partial enzymatic activity and can receive a lower dose.
- .33% of people have insufficient enzymatic activity and shouldn't receive any Purinethol.
- Pharmacogentic testing helps determine the correct dosage for each patient.

# Potential Benefits

- Improves Safety
  - If a genetic variant is found to be associated with an adverse reaction to a certain medicine, doctors could avoid prescribing the medicine to patients with this gene variant.

- Enhances Efficacy (ability to produce a desired effect)
  - Pharmacogenetics could allow doctors to prescribe medicine only for those patients most likely to respond.

# Potential Benefits Continued

- Adjusts Dosage
  - Genetic information could be used to adjust the dosage of a medicine, reducing the trial-and-error approach .

# Potential Drawbacks

- High cost of development.
  - High cost to consumer
- Identifying the genes that are affecting the patient takes time.
- The market for pharmacogentics may be too small.

# Ethical Information

- Allocation of Scarce Resources
  - Every year adverse reactions to drugs kill 100,000 Americans.
  - Over 2 million people have serious reactions to medication.
  - Critics say that it is a waste of time and money.
  - Who gets treated first?  Which ones are "worthy“?

- Fair Distribution of Burdens/Benefits
  - New drugs too expensive.
  - Conflicts of interest for those with investments.
  - Participants may not receive expected benefits.

# Ethical Information Continued

- Ethical Use of Individual Medication
  - Genes are not the only information needed to predict reaction.
  - Doctors may not test and continue to guess and check.
- Predominating Rights
  - Family history is needed.  Whose rights come first?

# Future Implications of Pharmacogenetics

- Basic Ideas/Thoughts
  - Clinicians and pharmaceutical companies are likely to continue favoring drugs that work well regardless of genetic differences. Where this is not possible pharmacogenetic medicines will become increasingly important.
  - Pharmacogenetic products are expected to enter mainstream healthcare in the next 15-20 years. This will happen as researchers discover subgroups of diseases based on different genetic factors and drugs are developed that target mutations in these genes.
  - Doctors will be able to use pharmacogenetic test results to help them decide the best treatment option. But they will continue to factor in other important influences, such as age, weight, diet and ethnicity, and they won't necessarily use a pharmacogenetic test in every prescription situation. Some people will find it hard to stick to a treatment regimen regardless of how it is prescribed, so patient adherence/compliance will continue to be an issue.

# Future Implications of Pharmacogenetics Continued

- Preparing the Profession
  - With the development of pharmacogenetic technology still being very new, the pharmacy profession should be looking to improve the education of its members. Rather than being taught on how to deliver a service, pharmacists need to develop a fundamental understanding of this technology so that they can suggest clinical practice for which it could provide maximum benefits to its patients.

# Future Implications of Pharmacogenetics Continued

- Role of the Pharmacist
  - A doctor's procedures include examining a patient, making a diagnosis, and prescribing a medication. Pharmacists ensure that the medicine has been prescribed appropriately; occasionally they may also be involved in making the prescribing decision. Once a diagnosis has been made and the decision to start drug therapy, the prescriber will need to determine if pharmacogenetic testing would aid drug or dose selection. If so, informed consent and a blood sample will need to be obtained from the patient. It is possible that the pharmacist will be responsible for some or all of these additional roles in monitoring and reviewing treatment. Pharmacists may also be involved in educating the public and other health professionals. Currently there are only a small amount of pharmacists in highly specialized roles that are involved in pharmacogentics, and this number is expected to jump in the near future.

# Future Implications of Pharmacogenetics Continued

- What Needs to be Done Now
  - As genetic research accelerates, basic education into the underlying principles is trailing behind. As the technology starts to impact more on primary healthcare, doctors, nurses and pharmacists will need more training in basic and applied human genetics, as well as guidance on how to offer and interpret key tests. Information technology needs to keep pace with pharmacogenetics. This means secure data storage and the development of computer programs that can analyze the influence of multiple factors on health including genetic data, age, gender, and other prescribed and over the counter medication. Health professionals will need training to manage and use this information. Well designed studies are needed to establish the relevance of pharmacogenetics to clinical practice, especially in the developing world. And as the field develops, regulations will be needed to follow scientific developments and their applications.

# Questions?