



Pharmacogenetics from Swabbing to Outcomes

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Objectives

- Describe the interconnection of medications and health outcomes
- Define methods of using pharmacogenomics in interdisciplinary practice
- Provide information on opportunities for nurses and health professionals to collaborate in precision medicine to impact population health.

Why PGx?



Pharmacogenetics may identify patients who will have a very high or a very low likelihood of responding to a medication.

- We do this so a drug can be specific to patients who will respond & will have low possibility of ADRs.

Pharmacogenomics can identify the subset of patients with higher risk of serious adverse effect. So that these drugs can be avoided

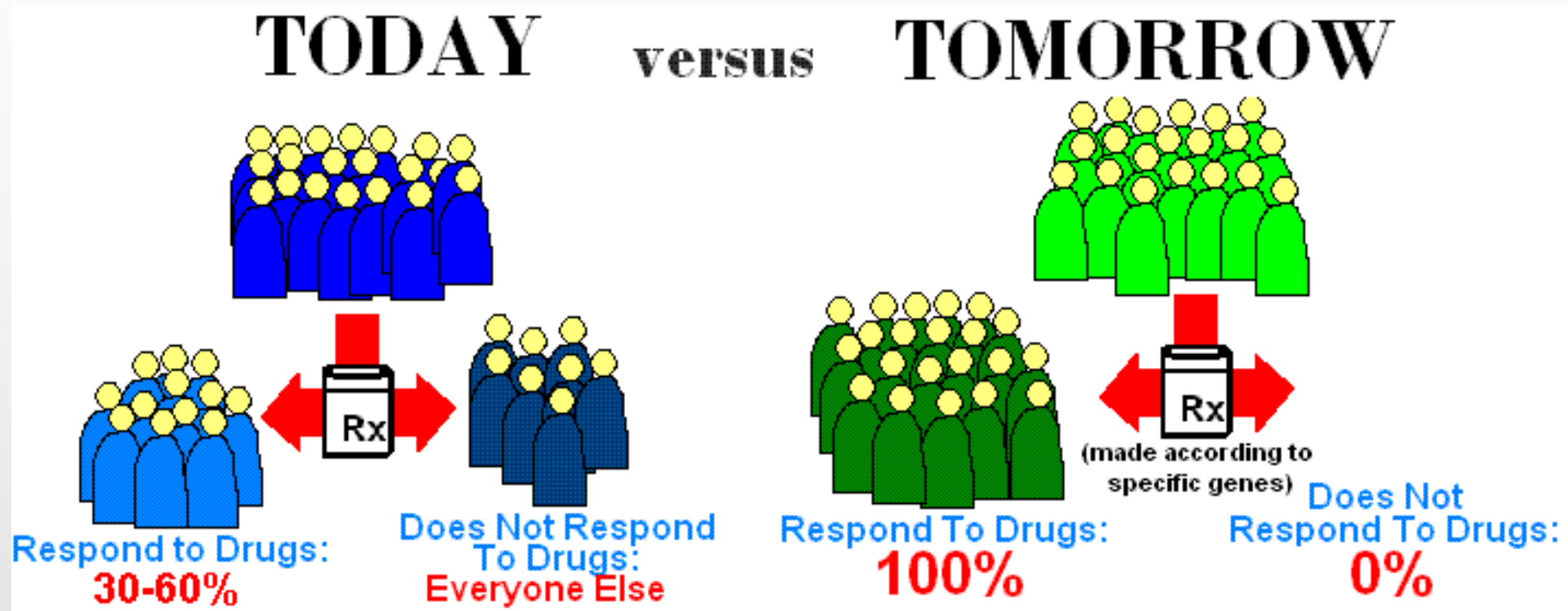
What is it?

- *PHARMACOGENETICS*: The effect of genetic variation on **drug response**, including disposition, safety, tolerability and efficacy.
- *PHARMACOGENOMICS*: **Employs the tools for surveying the entire genome** to assess multigenic determinants of drug response.

PGx Gene Review

- In general CYP enzymes are involved in about 75% of drug metabolism
- TMPT: Enzyme carries out a chemical reaction
- **ATM gene** is involved in **cell cycle control, apoptosis, gene regulation, oxidative stress, and telomere maintenance**
- The VKORC1 gene provides instructions for making a **vitamin K epoxide reductase enzyme**.
- **Factor V Leiden**: mutation of a clotting factor
- **MTHFR**: assists in metabolizing folate
- (Centers for Disease Control and Prevention, 2022)

Improvement is Needed



- Integrating pharmacogenomics into clinical practice to assist in drug selection and dosing has the potential to improve the outcomes of treatment, reduce the risk of drug-induced morbidity and death, and be cost-effective. Cleveland Clinic Journal of Medicine, Feb. 2020.
- With the NHS working towards the integration of personalized medicine, in particular, aiming for whole-genome sequencing to be standard for specific conditions by 2020, it is important for pharmacists and healthcare professionals to be aware of the principles of pharmacogenomics and its evolving application into clinical practice. The Pharmaceutical Journal Oct. 2017
- The advent of inexpensive genotyping and sequencing and the development of increasingly sophisticated EHR systems holds the promise that implementing pharmacogenomic variant information will become a routine part of the practice of Genomic Medicine. HHS Aug. 2019

Studies Indicate

- [Clinical Pharmacogenetics Implementation Consortium \(CPIC®\)](#) is an international consortium of individual volunteers and a small dedicated staff who are interested in facilitating use of pharmacogenetic tests for patient care (2023).
- **Position Statement: The American Society of Health System Pharmacists (ASHP) believes pharmacogenomic testing can improve medication-related outcomes across the continuum of care in all health system practice settings. (Am. Journal of Health, 2022)**
- Precision medicine carries the potential of improving health outcomes at both the individual as well as population levels. (Frontiers in Genetics, 2023)

Further Resources

Reasons why:



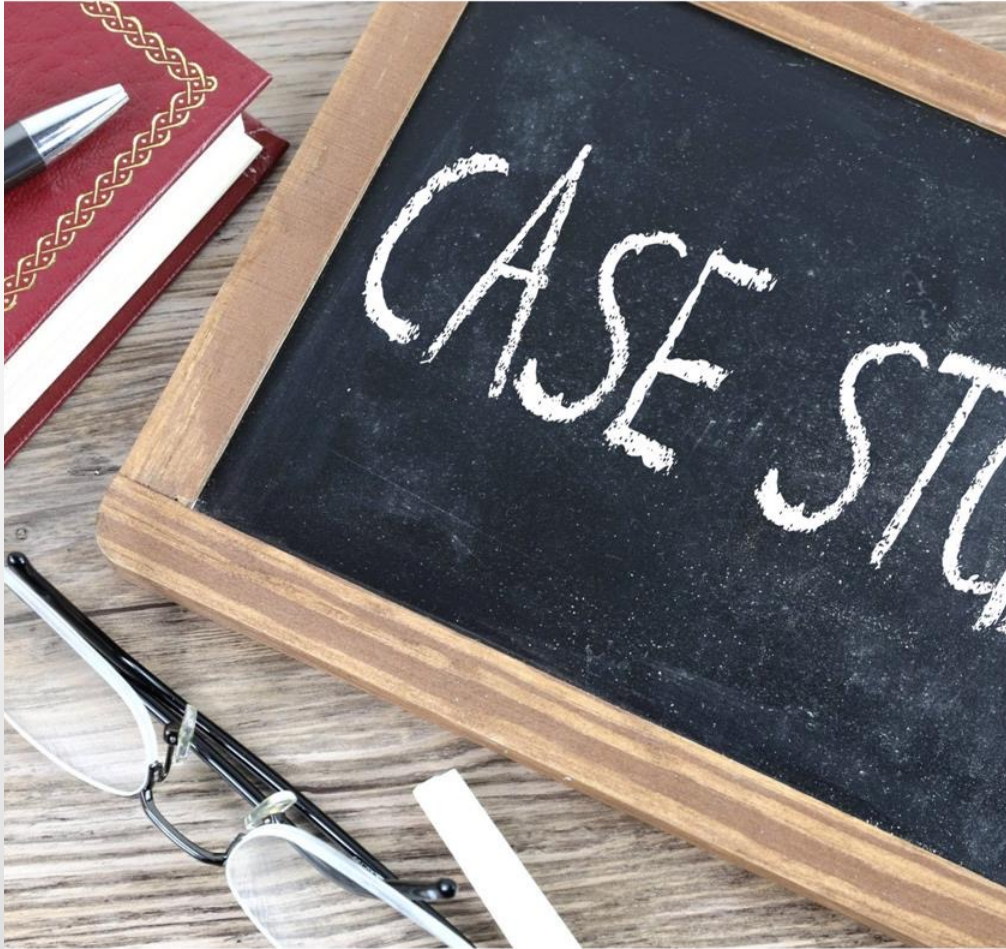
- To predict a patient's response to drugs
- To develop “customized” prescriptions
- To minimize or eliminate adverse events
- To improve efficacy and resident compliance
- Pharmacogenetic test need only be conducted once during the lifetime.
- To improve the accuracy of determining appropriate dosage of drugs

Why so difficult to implement?

- Despite considerable research activity, pharmacogenetics are not yet widely utilized in clinical practice.
- Dose adjustment on the basis of renal or hepatic dysfunction can be accepted by clinician.
- But there is much more hesitation from clinician to adjust the dose on pharmacogenetic ground.
- This can be due to resistance to accept or can be due to unfamiliarity with the principles of genetics.



Case Study



- Use of the information
- Recommendations and Why
- Future use of the data

Goals



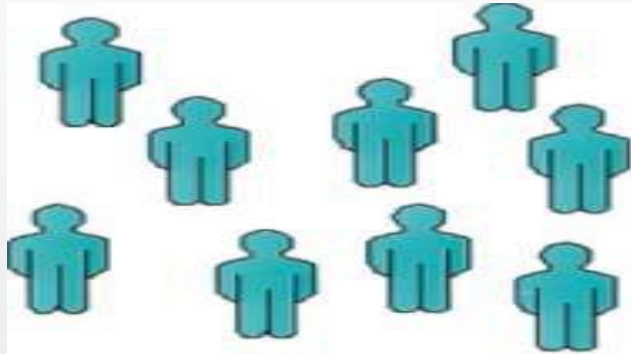
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Isn't this the best for our patients and public health?

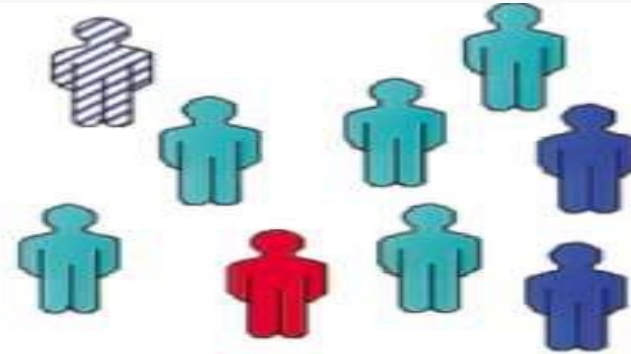
- Maximize drug efficacy
- Minimize drug toxicity
- Predict patient's response to treatment
- Have options that may assist in the patient's well-being
- Opportunities for practice are many

Precision Medicine: the Future is Bright

- One size DOES NOT fit all



Current Approach
One size fits all



Individualized Approach
*Drug and/or dose chosen
for each patient*



Questions?