PSW Pharmacogenetics Interest Group

Genomic Literacy

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Introduction

Health Literacy

"The degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions"

Health Numeracy

"The degree to which individuals have the capacity to access, process, interpret, communicate and act on numeric, quantitative, graphic, biostatistical data, and probabilistic health information to make health decisions"

Genomic Literacy

"The capacity to obtain, process, understand and use genomic information for health-related decision making"

Identifying Low Health and Genomic Literacy

- Education level does not always tell the full story
- Age and physical appearance are NOT a reliable measure
- **Individuals may hide or adapt literacy challenges**
  - 67% of patients with low literacy never told spouse
  - > 50% never told children
  - 19% ***never told anyone***

Davis T, et al. *CA: A Cancer Journal for Clinicians*. 2002. 53(3):134-149.

Implications of Genomic Health Literacy

- Like other forms of literacy, higher genetic literacy is expected to enhance informed decision-making in genetic testing

- Conversely, **low genetic literacy** is expected to **negatively impact** understanding of the genetic test

*Can lead to inappropriate follow-up or **noncompliance with recommended care**, such as additional testing and preventive clinical screenings*

Rasouley HM, et al. *J Genet Couns*. 2021;30:742–754.

Rapid Estimate of Adult Literacy in Genetics (REAL-G)

- Modeled after a widely used rapid estimate of adult literacy in medicine (REALM) test

- Involves watching a pre-recorded video about genomic literacy and genetic counseling and answering a series of questions

Sixth grade reading level identified as cut off for lower scores

Erby LH, et al. *American Journal of Medical Genetics*. 2008; 146A(2),174–181.

Genetic Literacy Fast Test (GeneLiFT)

- Designed to facilitate rapid screening of genetic literacy

- Test takers asked to self rank genomic knowledge, evaluate the accuracy of statements about genetic testing, and tested on the recognition of genomic terminology

Association with genetic knowledge was independent of health literacy, numeracy and education

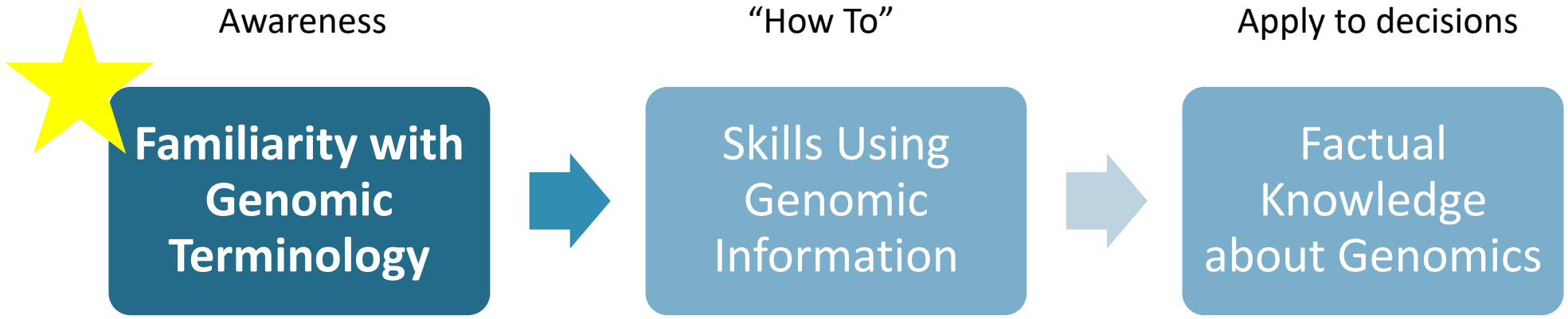
Genomic medicine concepts were poorly understood, even after adjusting for education level

Rasouley HM, et al. *J Genet Couns*. 2021;30:742–754.

Scaling Genomic Literacy

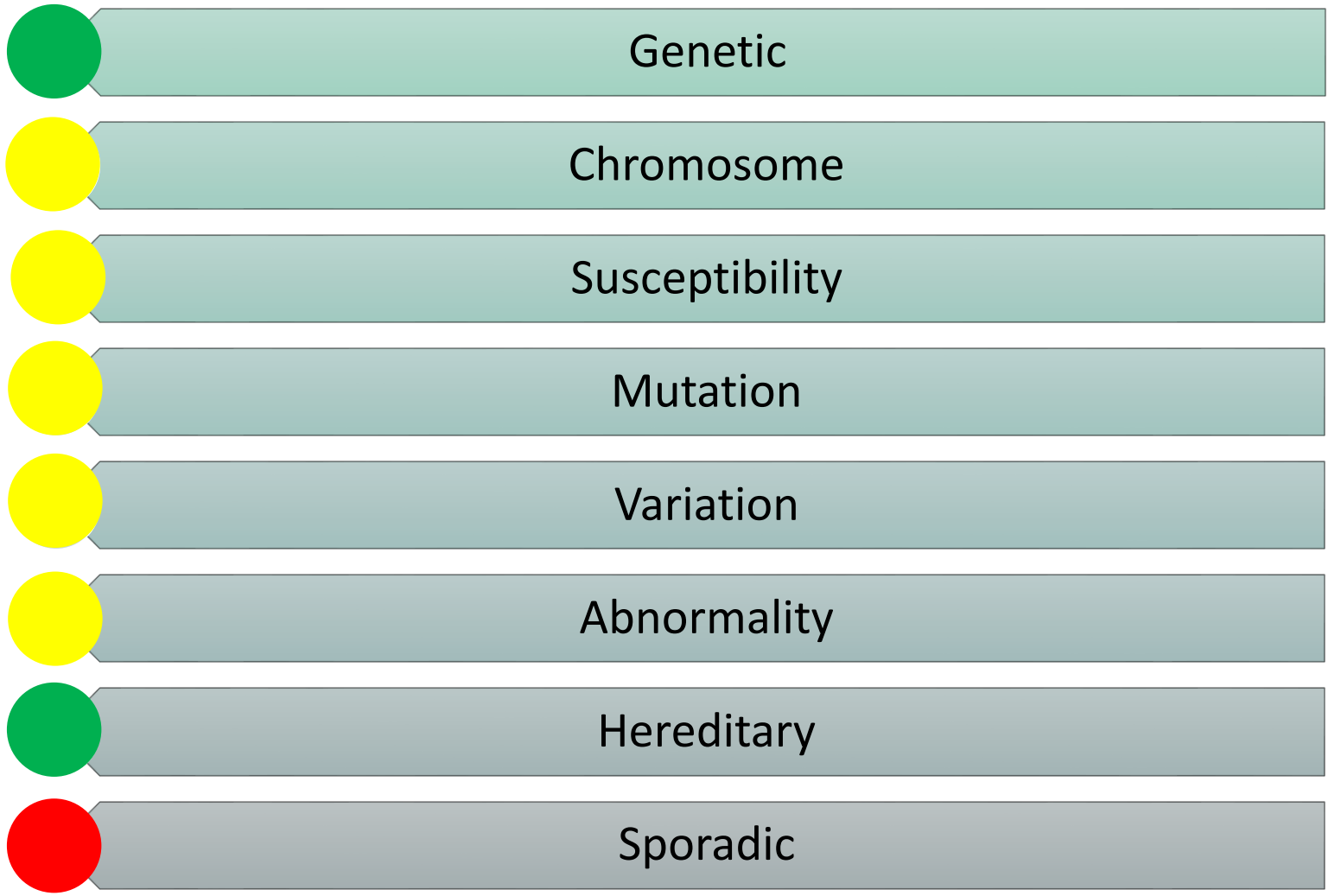
- Individuals need a combination of familiarity, skills, and factual knowledge to have a lay understanding of topics, including genomics

- Consumer panel evaluated genomic literacy in the United States, adjusting for education level



Abrams LR, et al. *PLoS ONE.* 10(10): e0141532.

Genomic Terminology – “Familiar” Terms



Abrams LR, et al. *PLoS ONE*. 10(10): e0141532.

Genomic Literacy in Oncology

- "**Precision Oncology**" testing framework analysis revealed:
  - Almost **two thirds** of patients reported never having heard of or not knowing anything about the term "**Precision Medicine**"
  - **33 terms** used to describe biomarker and genetic testing for cancer patients in patient education materials
  - Example – "**Biomarker testing**" also referred to as:
    - *Molecular testing, tumor profiling, somatic testing, genomic testing…*

Martin NA, et al. *JCO Precis Oncol*. 2021;5:1563-1567.

Genomic Literacy in Oncology

Recommended consensus umbrella testing terminology:

"Biomarker testing"

- Preferred term for testing for somatic acquired alterations and in the DNA of cancer cells, or when referring to other biomarkers

"Genetic testing"

- Preferred term for inherited variations of genes, and genetic testing for inherited cancer risk for germline (sperm/egg) variants

Martin NA, et al. *JCO Precis Oncol*. 2021;5:1563-1567.

Genomic Testing: Implications for Health Literacy

- Patient and provider communication
- Informed consent
- Evaluation of risks and benefits of testing
- Goals of testing
- Cost of testing and reimbursement
- Interpretation of test results
- Delivery of test results
- Patient education, education materials, and resources

Patient Education and Communication

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Patient Education Tips

- ✓ Ensure caregivers are present
- ✓ **Focus on the top 3 to 5 key pieces of information!**
- ✓ Slow down
- ✓ Repeat
- ✓ Summarize
- ✓ Use visual aids
- ✓ Use plain language
- ✓ Provide clear written education materials

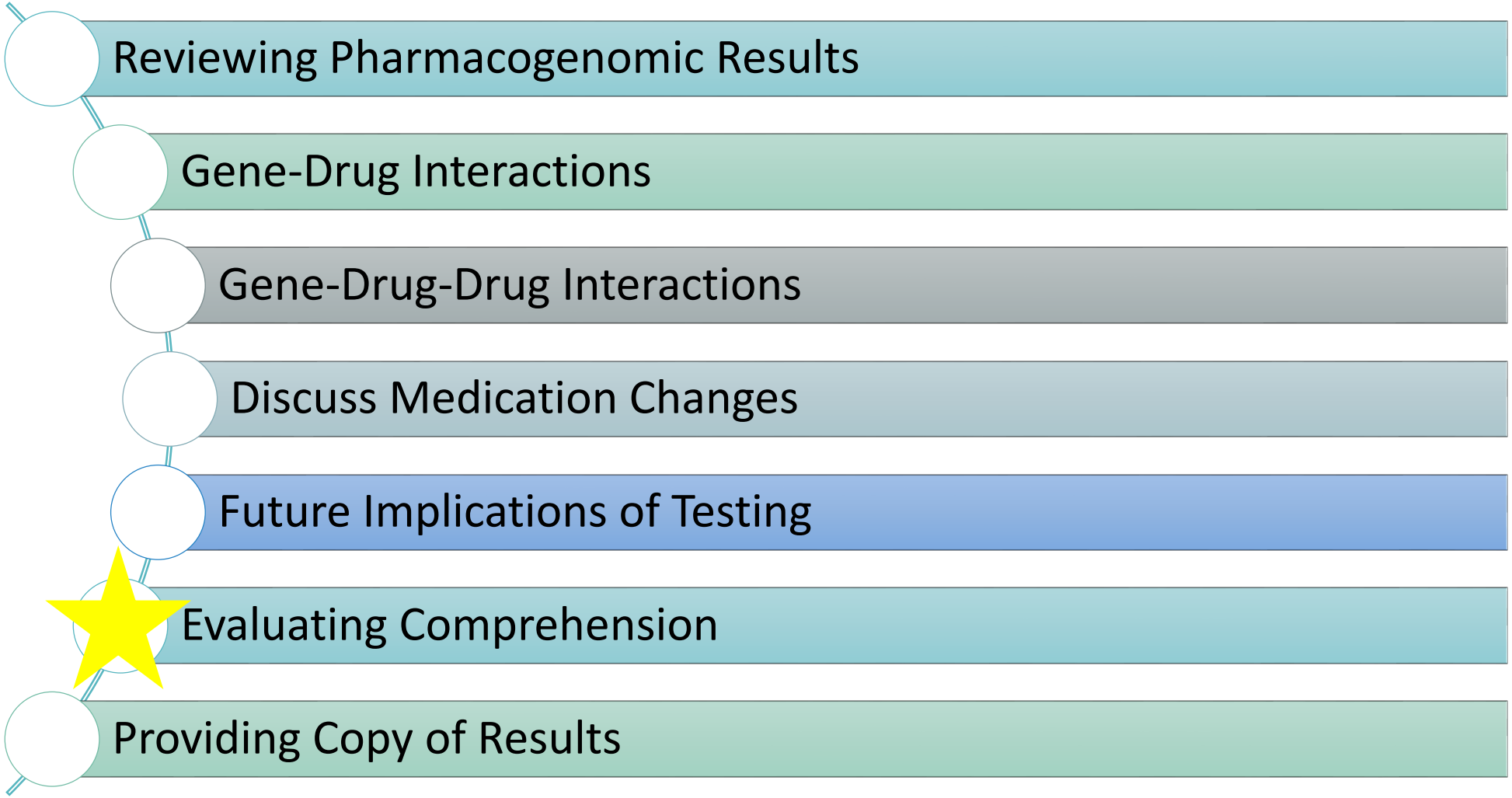
Mills R, et al. *Pharmacogenomics Pers Med*. 2013;6:105-112.

Literacy Evaluation - Pause for …

- ❑ Few or no follow up questions
- ❑ Unable to provide names and indications for medications
- ❑ Adherence issues with medications, testing, or referrals
- ❑ Missed appointments
- ❑ Incomplete forms filled out

Davis T, et al. *CA: A Cancer Journal for Clinicians*. 2002. 53(3):134-149.

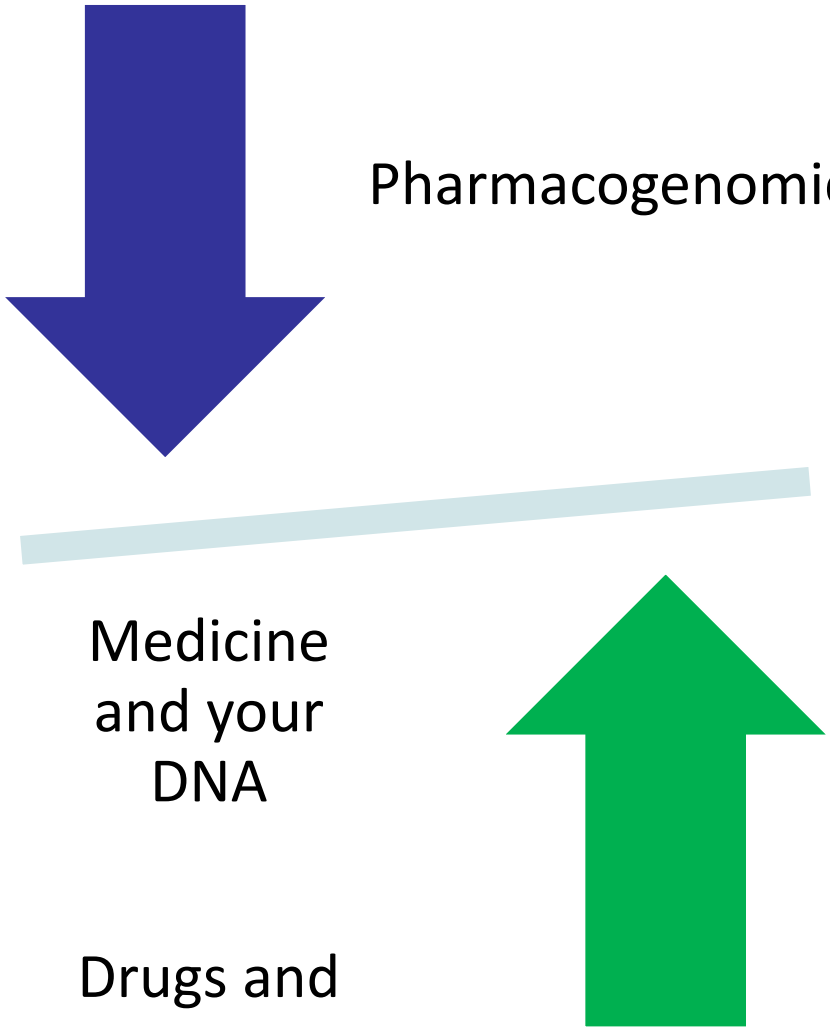
Pharmacogenomic Counseling Session

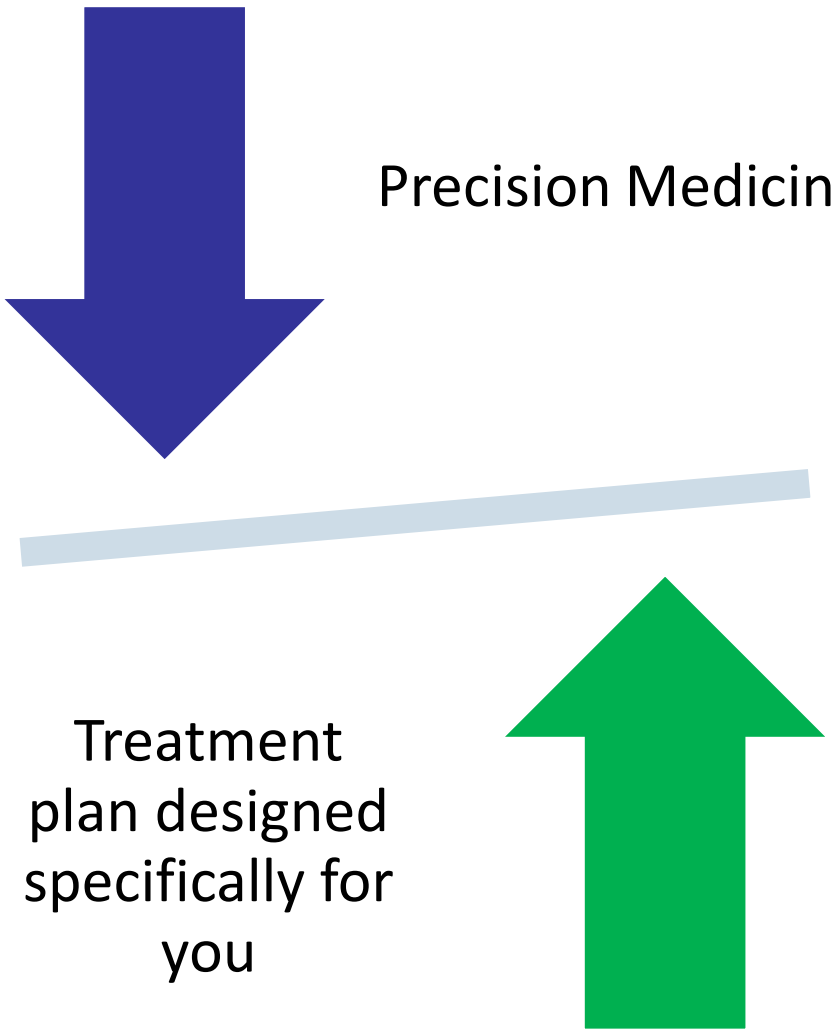


Ho T, et al. *Am J Health-Syst Pharm*. 2022;79:1634-1644


“Right Patient, Right Drug, Right Dose, Right Time…”
But What are the Right WORDS?

Alternative Options





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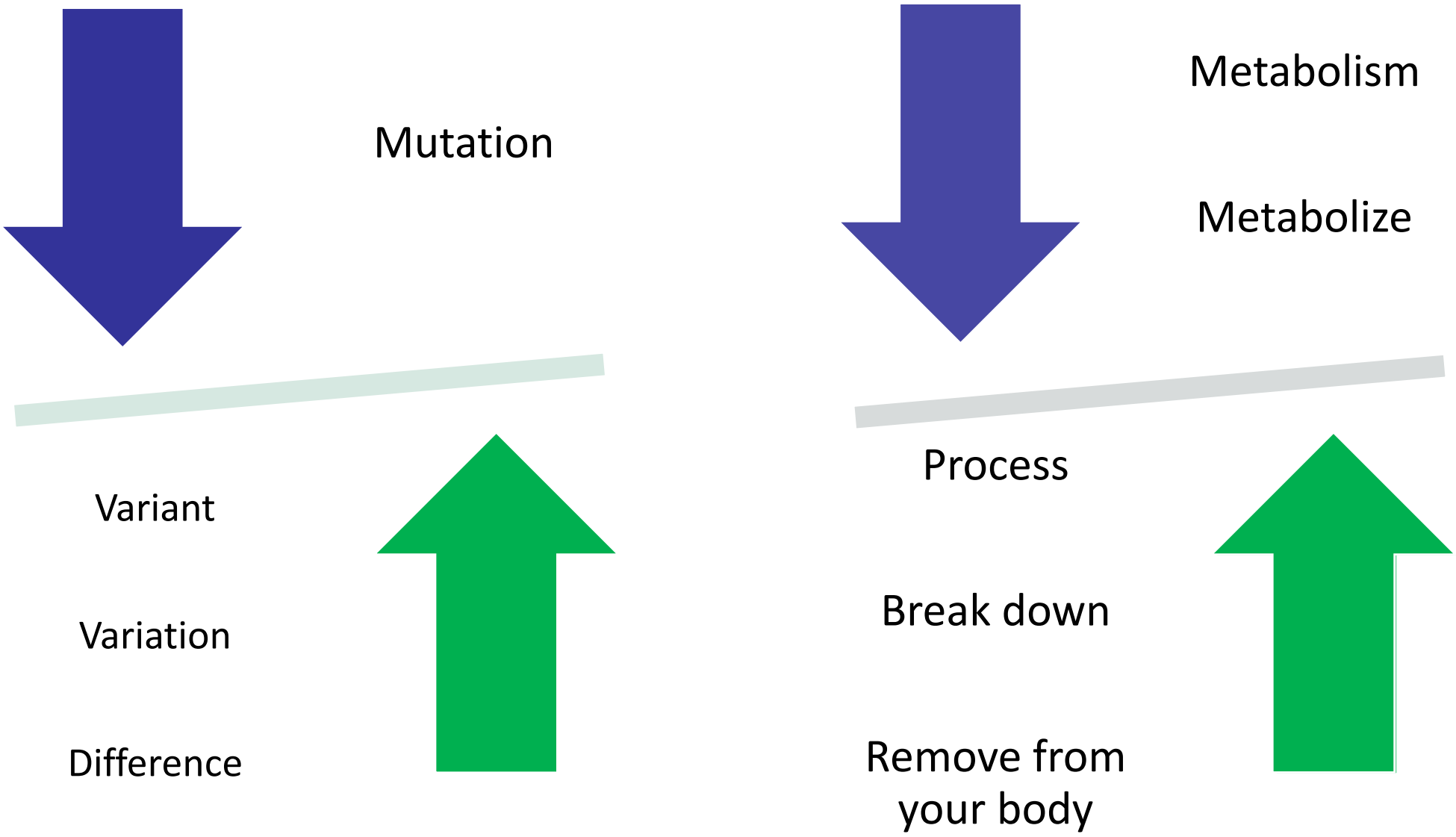
Alternative Options



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Helpful Phrases

- “Your DNA may impact how you respond to medications”
- “Genomic test results can help choose the best medications at the correct doses for you”
- “Do not make any changes to your medications based on gene testing results without talking to your health care team first”

Ho T, et al. *Am J Health-Syst Pharm*. 2022;79:1634-1644.
Mills R, et al. *Pharmacogenomics Pers Med*. 2013;6:105-112.

Patient Communication Example

Genotype

- This result is a variation in the DNA you inherited, which impacts how your body breaks down and reacts to medicines

Phenotype

- "Poor metabolizer" means your body may not break down or process this medication effectively
- It may not work as intended and you may have side effects

Action

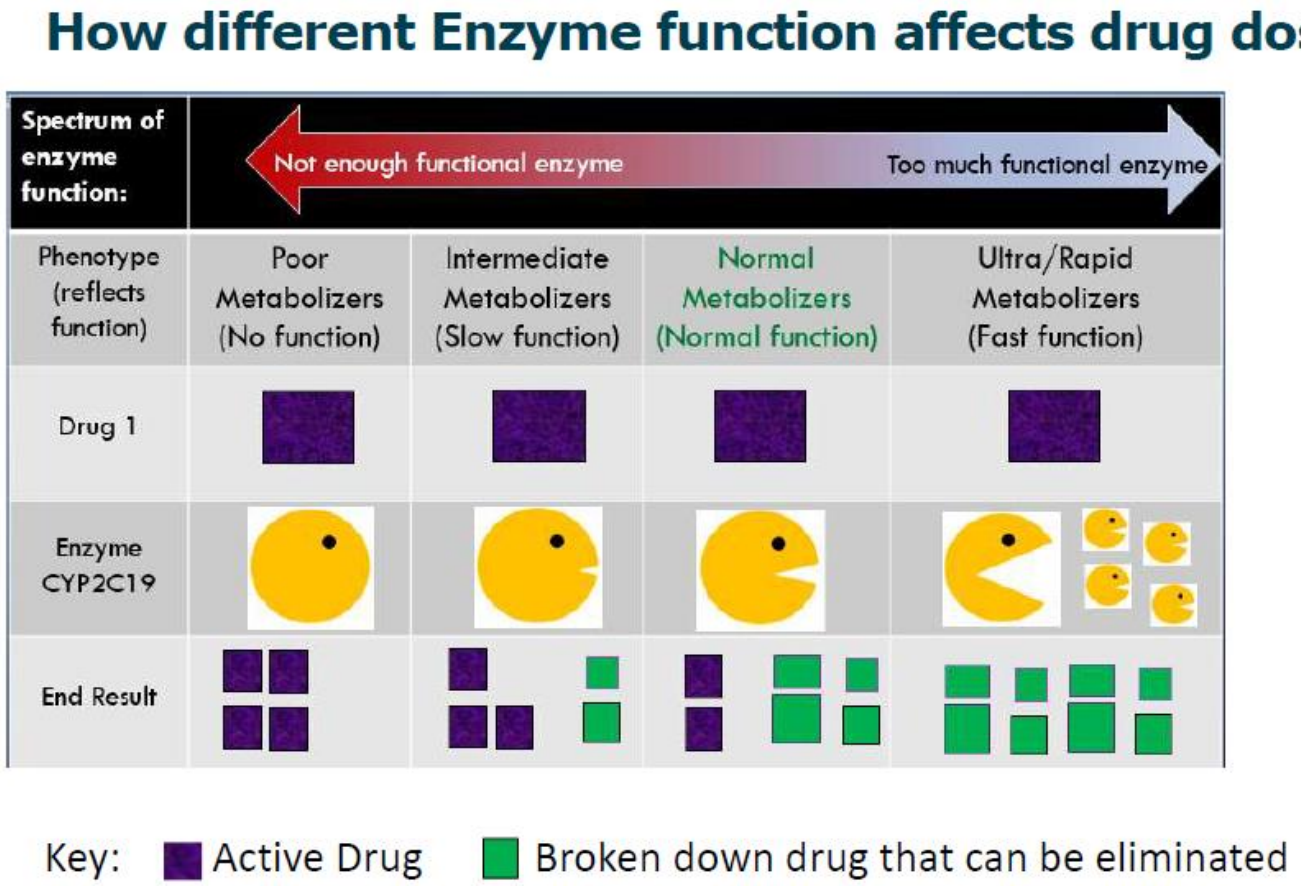
- Check with your doctor for the final plan; there may be a change in your medicine

Mills R, et al. *Pharmacogenomics Pers Med*. 2013;6:105-112.

Designing Genomics Educational Handouts

Short chunks of limited text
Clear formatting
Use graphics
Provide visual cues
Include pictures
Reiterate main take-aways
Provide definitions or glossary

Marshfield Clinics Example – “Pac Man”



Patient-Pharmacist Reflection

- What signs will you look for to indicate genomic literacy?
- What words or phrases will you use to make sure you are explaining the test results clearly?
- What are the top key concepts you want the patient to walk away with?
- How will you double check to ensure comprehension?
- How would you document this interaction in the electronic health record?
- What patient resources can you recommend for additional genomic testing information?

Provider Education and Communication

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Genetic Terms and Definitions

Term	Definition
Allele	A version of a gene
Diplotype	A haplotype pair
Gene	Physical unit of inheritance
Genotype	Individual collection of genes; or diplotype
Haplotype	DNA variations inherited together
Heterozygous	Two different inherited alleles
Homozygous	Two of the same inherited alleles
Phenotype	Observable traits based on genetics
Polymorphism	Variations of a DNA sequence

National Human Genome Research Institute. Talking Glossary of Genetic Terms.

Standardized Consensus Terms

Gene Category	Phenotype Terminology
Drug Metabolizing Enzymes	○ Ultra-rapid metabolizer ○ Rapid metabolizer ○ Normal metabolizer ○ Intermediate metabolizer ○ Poor metabolizer
Drug Transporters	○ Increased function ○ Normal function ○ Decreased function ○ Poor function
HLA (Human leukocyte antigen) Molecules	○ Positive ○ Negative

Caudle KE, et al. *Genet Med*. 2017;19:215-23.

Provider Communication Example

Genotype

- This patient has the *CYP2C19 *17/*17* genotype, which means the patient is an ultra-rapid metabolizer of voriconazole

Phenotype

- The probability of maintaining therapeutic trough levels is low
- This could result in an increased risk of fungal infection

Action

- Recommend choosing an alternative agent, such as isavuconazole

Moriyama B, et al. *Clin Pharmacol Ther*. 2017;102(1):45-51.

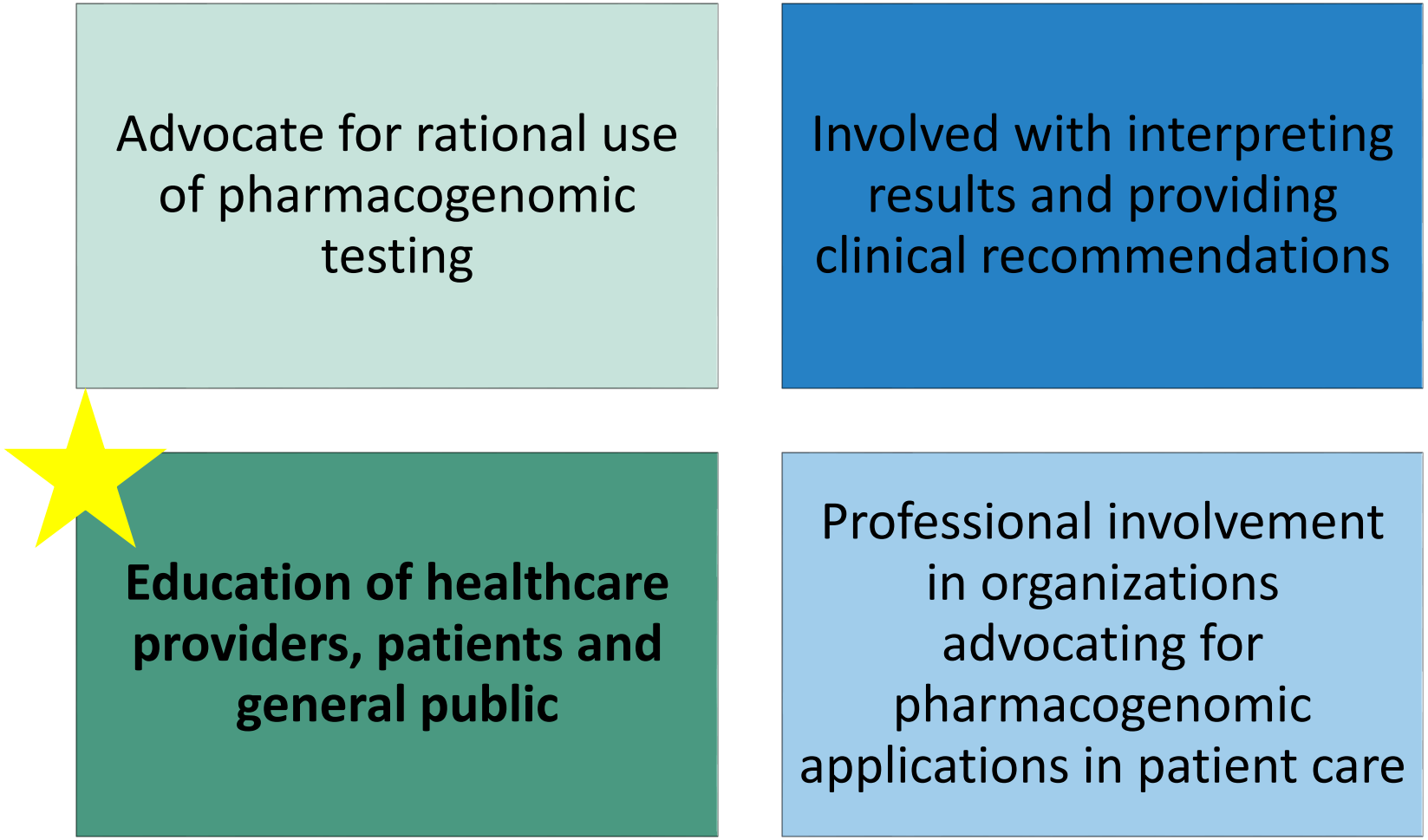
Provider-Pharmacist Reflection

- What is your first step when preparing to communicate genomic information?
- What resources can you use to determine the recommendation to a provider as a result of a genomic test result?
- How will you communicate your recommendation to the provider?
- How does this communication differ from patient education? How do you evaluate understanding?
- How will you document the intervention in the electronic health record?
- What follow up is needed with the patient?

Role of the Pharmacist

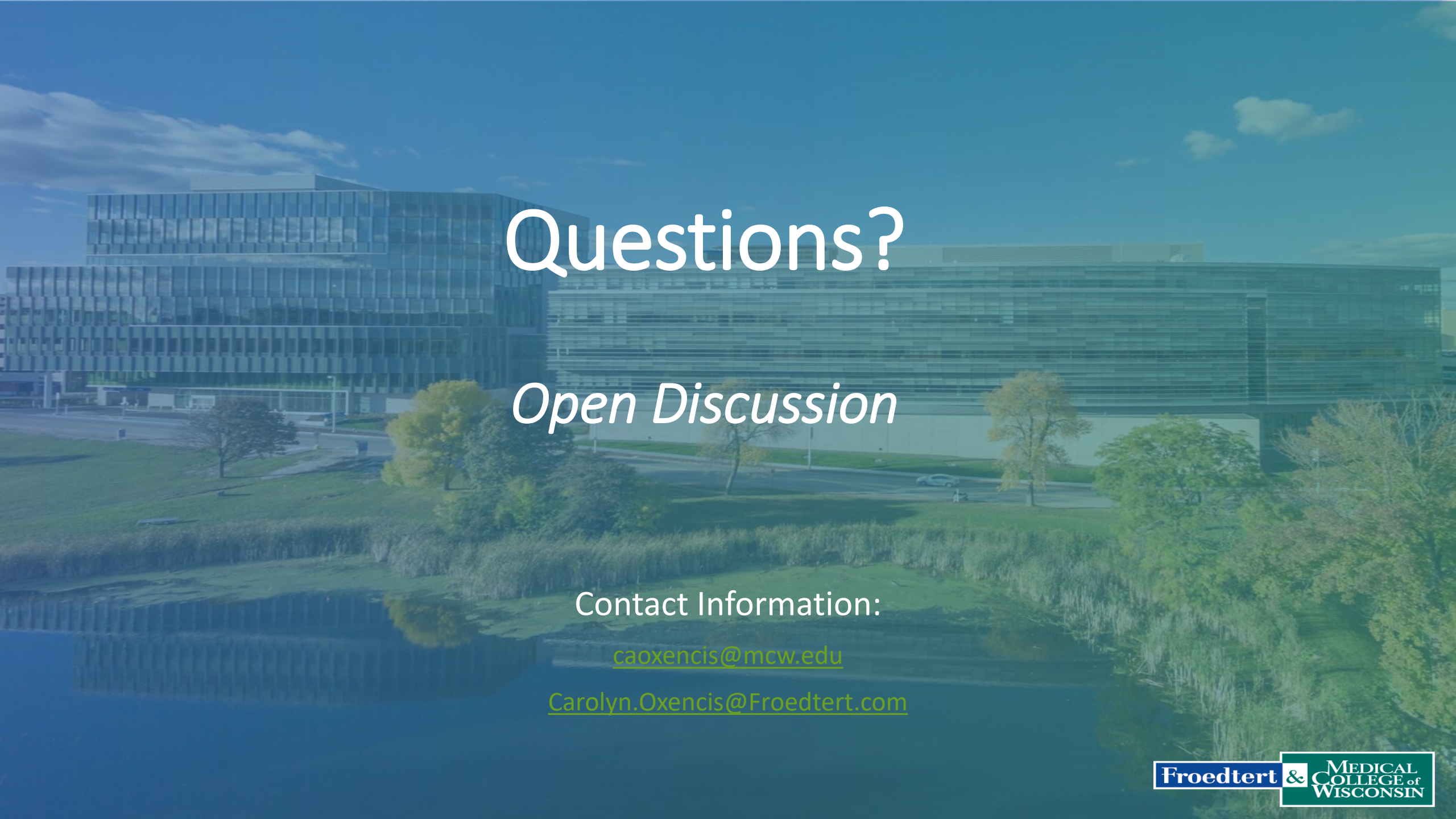
- ASHP position statement
- AJPE pharmacist core competencies in genomics

"Drug experts need to be pharmacogenomic experts"



Summary

- Communication is key – **WORD CHOICE MATTERS**
- In order to explain genomics to providers and patients, you must understand and feel **comfortable** and **competent** with pharmacogenomics in your clinical practice
- Spread awareness of the importance of **genomic literacy**



Questions?

Open Discussion

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Resources for Providers

- Table of Pharmacogenomic Biomarkers in Drug Labeling
  - **FDA (Food and Drug Administration)**
  - https://www.fda.gov/drugs/science-and-research-drugs/table-pharmacogenomic-biomarkers-drug-labeling

- Evidence-based clinical practice guidelines for pharmacogenomic results
  - **CPIC (Clinical Pharmacogenetics Implementation Consortium**)
  - https://cpicpgx.org/

- Pharmacogenomics Knowledge Resource
  - **PharmGKB**
  - https://www.pharmgkb.org/

Resources for Patients

Source	Resource
National Human Genome Research Institute	**Fact sheets about genomics** • https://www.genome.gov/about-genomics/fact-sheets **Pharmacogenomics FAQ** • https://www.genome.gov/FAQ/Pharmacogenomics **Introduction to Genomics** • https://www.genome.gov/About-Genomics/Introduction-to-Genomics **Talking Glossary of Genetic Terms** • https://www.genome.gov/genetics-glossary
US National Library of Medicine	**MedlinePlus Genetics** • https://medlineplus.gov/genetics/
Pharmacogenomics Knowledgebase	**What Is Pharmacogenomics?** • www.pharmgkb.org/whatIsPharmacogenomics/genetics-101
St. Jude Children's Research Hospital	**Pharmacy and Medicines Educational Materials** • www.stjude.org/treatment/patient-resources/caregiver-resources/patient-family-education-sheets/pharmacy-and-medicines.html

Ho T, et al. *Am J Health-Syst Pharm*. 2022;79:1634-1644

Genetics Professionals Resources

- **The National Society of Genetics Counselors (NSGC)**
  - https://www.nsgc.org
    - Find a genetic counselor

- **The American College of Medical Genetics and Genomics (ACMG)**
  - https://www.acmg.net
    - Find a genetics specialist
    - Find a genomics clinical tool