## Communicating Pharmacogenomic Test Results to Pediatric Patients and Caregivers

# "PEDIATRICS PGx TESTING"

## A Counselling Best Practices for Healthcare Practitioners

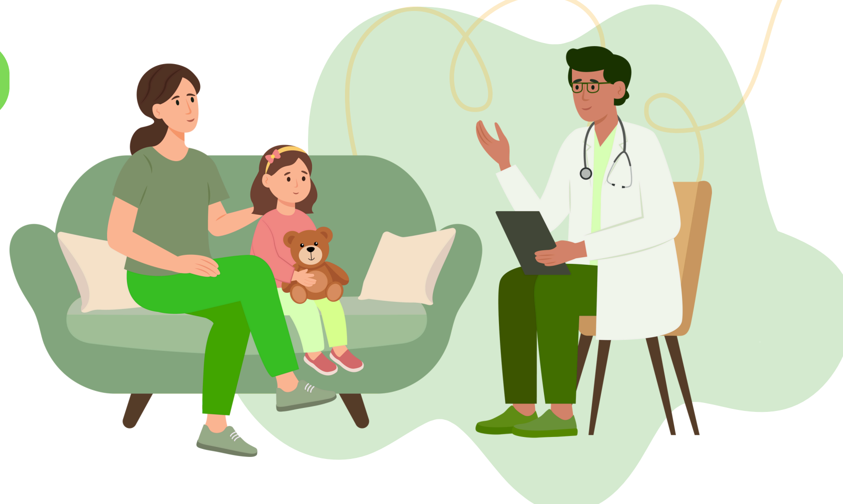
**Pharmacogenomic (PGx) testing** is an evidence-based strategy to optimize medication selection and dosing based on an individual's genetic information. It has been implemented in many healthcare systems worldwide to help inform therapeutic decisions for mental health, pain, oncology, gastrointestinal disorders, and more. With its increasing prevalence in pediatrics, communication of the results to young people and their caregivers is a crucial component in delivering effective care to families.

**"PEDIATRICS PGx TESTING"** is a communication tool that uses mnemonics to assist healthcare practitioners in delivering a thorough pre-test and post-test counselling to pediatric patients and their caregivers.

## PRE-TEST

### "PEDIATRICS"

- **P** — **Purpose**
  Educate on the purpose and benefits of PGx testing.
- **E** — **Expectation Management**
  Discuss the evidence of gene-drug interactions in the context of the patient's unique situation, including the impact on current and future medication use.
- **D** — **Drawbacks**
  Discuss limitations, including ethno-geographic variability. Ensure the understanding that PGx testing is not a disease risk assessment or diagnostic tool.
- **I** — **Incidental Findings**
  Explain the concept of incidental/unexpected findings and their potential discovery via PGx testing.
- **A** — **Ask for Understanding!**
  Open the floor to explore what questions that arise about any discussed points thus far.
- **T** — **Testing Procedure**
  Explain the physical testing procedure (saliva, swab, etc.), timeline, and format to expect results (phone call, portal, etc.).
- **R** — **Risks**
  Address potential risks (e.g., psycho-social impacts of results, misattributed paternity, disease risk).
- **I** — **Insurance and Costs**
  Communicate expected costs and possible insurance coverage. Discuss and address concerns regarding potential insurance and employment discrimination.
- **C** — **Consent/Assent**
  Discuss consent/assent process.
- **S** — **Sharing of Data**
  Discuss who will have access to their results and the systems (e.g., PIPEDA) and laws (e.g., GNA) in place to protect their data.



## POST-TEST

### "PGx TESTING"

- **P** — **Purpose**
  Reiterate the purpose, benefits, and limitations of PGx testing.
- **G** — **Go Over Results**
  Interpret the results based on the patient's unique situation. Emphasize that the default "normal" may not capture all variants. Answer any questions regarding test concepts or the results.
- **X** — **Expected Changes**
  Communicate initial changes that can be made to the child's current medication schedule, emphasizing that no medication changes should be made without consulting a provider.
- **T** — **Time and Future Implications**
  Discuss how the relevance of results may change over time. Emphasize the utility of the results for future medication decisions.
- **E** — **Education and Emotions**
  Discuss potential secondary findings and familial implications. Prepare for possible emotional reactions and provide appropriate resources and referral if necessary.
- **S** — **Sharing of Results**
  Encourage sharing of results with providers responsible for their current or future care. Discuss if and where results will be stored and/or shared.
- **T** — **Tools/Supplemental Education**
  Provide families with resources (info-graphics, video links etc.) that may enrich their understanding of the results.
- **I** — **Information Summary**
  Provide a portable copy (e.g., wallet card) of the results which they can share with providers in their care.
- **N** — **Next Appointment**
  Schedule a follow-up to answer any new questions and to review their medication management plan.
- **G** — **Game Plan**
  Conclude with a summary of results and their meaning. Ask them to "teach-back" the main points to ensure understanding.

# Communication Tips

Communicating pharmacogenomic (PGx) information with pediatric patients and their caregivers can be optimized using the following communication techniques (*adapted from these references).

## Determine communication preferences

Ask caregivers prior to counselling whether they would like to be counselled prior to or alongside the child and the depth of content (big picture vs details, etc.)

## Choose appropriate language

Tailor explanations to align with health literacy, numeracy, and developmental level, and avoid terms that increase the risk of misinterpretation.

**Avoid** terms such as “mutant”, “abnormal”, or “poor metabolizer”.
**Instead,** use terms such as “working” or “non-working”, or “fast” or “slow” drug metabolism.

## Include the patient

Emphasize a shared decision-making model when counselling about PGx.

## Manage Expectations

Families can have unrealistic expectations of what PGx testing can offer (e.g., it can determine "right drug and right dose"). Explain that PGx testing is a tool that can reduce some of the uncertainty that is inherent with drug therapy.

## Use visual aids

Provide graphs, info-graphics, or videos to supplement the discussion and improve understanding. See “Resources”.

## Use “teach-back”

Ask caregivers and/or patients to explain using their own words to confirm understanding.

## Review consent practices

It is a common practice to obtain caregiver consent and pediatric assent/consent. Allocate sufficient time to review the consent form with the family!

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# Resources

Check out these helpful resources for more information regarding pharmacogenomic testing and tools to implement in your practice.

**Cincinnati Children’s Hospital**
**Patient Education**
https://www.cincinnatichildrens.org/service/g/genetic-pharmacology/education
A video outlining PGx testing can be found following this link, along with patient handouts that correlate to specific gene results.

**ClinPGx**
**Pediatric Pharmacogenomics**
https://www.clinpgx.org/page/PedsIntro
ClinPGx provides a helpful dashboard which provides PGx information based on pediatric studies or relevant to pediatrics.

**Sequence2Script**
**Translate PGx Data into Clinical Action**
https://sequence2script.com
A tool for healthcare providers to translate PGx test results into clinically useful recommendations.

**PharmDOG**
**Pediatric Pharmacogenomics**
https://pharmdog.clinpgx.org
A tool that provides genotype-based prescribing guidance for practitioners.

**Genome BC**
**Pharmacogenomics**
https://www.genomebc.ca/infobulletins/pharmacogenomics
This page contains PGx information sheet for clinicians and patients and a conversation guide.

**St. Jude’s Children’s Hospital**
**Pharmacogenomics**
https://together.stjude.org/en-us/treatment-tests-procedures/tests/pharmacogenetics.html
This page outlines the basics of pharmacogenomic testing concepts in relation to pediatric care.

**National Human Genome Research Institute**
**Pharmacogenomics**
https://www.youtube.com/watch?v=QtnyYxmhsoM
Demonstrates how to utilize PGx resources, such as the CPIC and DPWG guidelines.

**Maruf and Colleagues (2020)**
**Pharmacogenetic Testing Options Relevant to Psychiatry in Canada**
https://pubmed.ncbi.nlm.nih.gov/32064906
Compares the available PGx tests in Canada.

# More Information

Visit the **MARUF LAB** website to learn more about ongoing PGx projects and download a copy of the tool.

***References:** Allen *et al.* (2022); Avard *et al.* (2009); Donnelly *et al.* (2024); Haga *et al.* (2014); Haga (2019); Hernandez-Martinez *et al.* (2023); Ho *et al.* (2022); Madadi *et al.* (2011); Mills *et al.* (2013); Pervola *et al.* (2019); PsychPGxLab (2025); Stancil *et al.* (2021); Weitzel *et al.* (2019); Zierhut *et al.* (2017). **GNA**, Genetic Non-Discrimination Act; **PIPEDA**, Personal Information Protection and Electronic Documents Act; **CPIC,** Clinical Pharmacogenetics Implementation Consortium; **DPWG**, Dutch Pharmacogenomics Working Group. Corresponding author: abdullah.maruf@umanitoba.ca
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