Patient and Family Perceptions of Pharmacogenomic Testing at St. Jude Children's Research Hospital

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Background

- PG4KDS is an IRB-approved protocol that enrolls patients at St. Jude Children's Research Hospital (St. Jude).[1] www.stjude.org/pg4kds
- Each participant is pre-emptively genotyped for variations in genes that are known to affect drug therapy.
- Results for 14 pharmacogenes are placed in the patient's electronic health record (EHR) and uploaded to the patient portal along with an explanation of the patient's phenotype and its implications for pharmacotherapy.[1]
- To date, nearly 7,000 patients have been enrolled in PG4KDS. Age of participants ranges from 1 month to 51 years (median 8 years of age).[1.]
- 95% of patients have at least one high-risk result in their health record.[2]

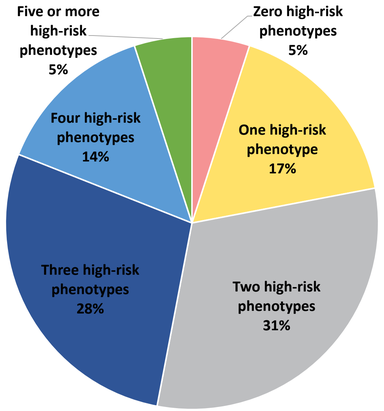

Figure 1. Breakdown of number of high-risk phenotypes for patients enrolled on PG4KDS out of fourteen total phenotypes reported per patient.

- Patients and families receive general education on pharmacogenomics and its potential implications on medication therapy during the PG4KDS consent process.
- It is important to educate the patient on the importance of their pharmacogenomic results as patients are often responsible for communicating results to outside providers.

Objective

Describe the perceptions of patients and/or legal guardians regarding pharmacogenomic testing provided at St. Jude.

Methods

- A survey was distributed via email to 523 families of patients actively enrolled on PG4KDS who had agreed to be contacted for participation in surveys through St. Jude.
- 128 individuals (24%) completed the survey. All responses were submitted by caregivers of minors.

Results

Have you received your child's pharmacogenomic test results?

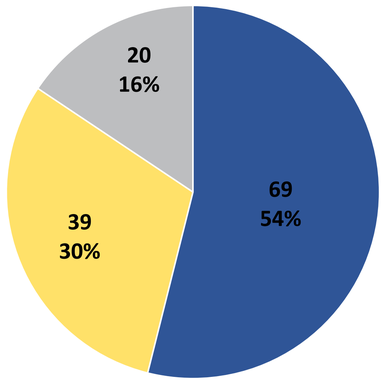

Figure 2. Guardian reported receipt of pharmacogenomic test results.

Did you understand the pharmacogenomic results when you read them on your own?

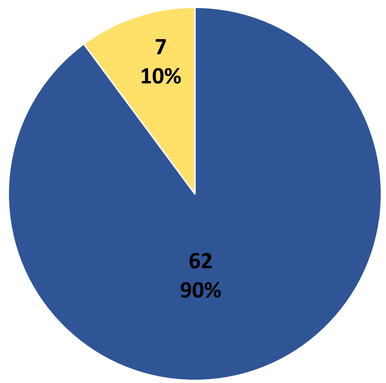

Figure 3. Guardian reported understanding of pharmacogenomic test results.

Has your child needed a modification in therapy based on their pharmacogenomic test results?

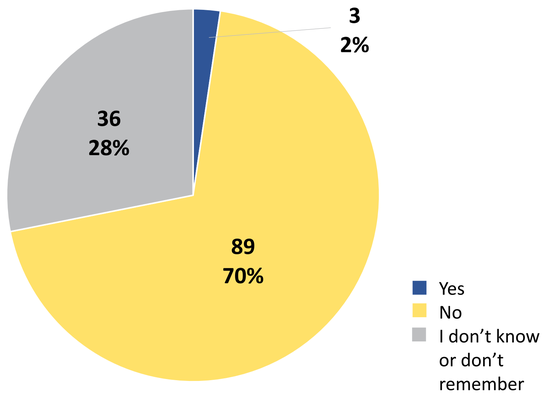

Figure 4. Guardian awareness of modifications made to patient's pharmacotherapy based on test results.

Have you shared your child's pharmacogenomic test results with their non-St. Jude physicians?

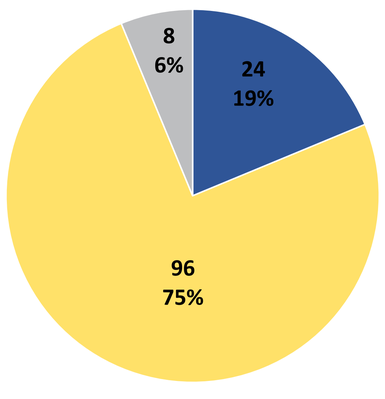

Figure 5. Guardian reported communication of results to physicians external to St. Jude.

If you have not shared the results with your child's non-St. Jude physicians, why not?	N (%)
Forgot/had not occurred to me	21 (22%)
Don't have results or don't know how to access them	31 (32%)
Have not considered it necessary	14 (15%)
Have not seen non-St. Jude providers since testing	26 (27%)
No reason given	4 (4%)

Table 1. Guardian reported reasons for not communicating pharmacogenomic results to physicians external to St. Jude (total percentage out of 96 respondents who answered "no" to sharing results with outside physicians).

Results cont.

Respondent Relationship to Patient	N (%)
Mother	117 (92%)
Father	9 (7%)
Grandmother	1 (1%)
Race	**N (%)**
White	112 (88%)
African American	7 (5%)
Asian/Pacific Islander	4 (3%)
Native American	1 (1%)
Not Reported	4 (3%)
Ethnicity	**N (%)**
Hispanic	8 (6%)
Non-Hispanic	120 (94%)

Table 2. Demographics of survey respondents.

Conclusions

- Guardians report an understanding of pharmacogenomic test results based on education provided during the consent process.
- Survey results indicate that caregivers may not be aware of most/all adjustments made to their child's therapy based on pharmacogenomic test results.
- A majority of guardians reported that they have not disseminated results to non-St. Jude physicians.
- A process is being developed at St. Jude for a pharmacist to return pharmacogenomic results in person to patients and their families.
- Improving patient and family knowledge of how pharmacogenomic test results are utilized may encourage continued use of these lifelong test results.

References

1. Hoffman, J. M., Haidar, C. E., Wilkinson, M. R., Crews, K. R., Baker, D. K., Kornegay, N. M., ... & Relling, M. V. (2014). PG4KDS: a model for the clinical implementation of pre-emptive pharmacogenetics. In *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* (Vol. 166, No. 1, pp. 45-55).
2. Haidar, C. E., Crews, K. R., Hoffman, J. M., Relling, M. V., & Caudle, K. E. (2022). Advancing Pharmacogenomics from Single-Gene to Preemptive Testing. *Annual Review of Genomics and Human Genetics* (Vol. 23, pp. 449-473).