NeuroPGx – Pharmacogenetic Testing in Pediatric Neurology:

A Pragmatic Study Evaluating Clinician and Patient Perceptions

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Background

- The field of pharmacogenetics has grown exponentially, over the past years; however, use of these tests in practice has been limited;
- Pediatric epilepsy patients, in particular, could benefit from pharmacogenetic testing as clinically relevant gene-drug associations for antiepileptics have been reported.

Objectives

- The primary objective of this study was to evaluate clinicians' perception of pharmacogenetic testing.
- Patients' and community pharmacists' perceptions were also evaluated in order to assess all participants in the clinical pharmacogenetic testing process.

Methods

- This is a prospective observational mixed-methods study
- This study was conducted at the Centre Hospitalier Universitaire Sainte-Justine in Montreal, Quebec, from March 2021 to August 2021
- Neurologists from the study center were given access to a pharmacogenetic panel (Precision Rx; Dynacare; Laval, Qc) for their pediatric patients with epilepsy who had a follow-up appointment within the study period.
  - The results report also provided a pharmacist's recommendations for the management of the relevant psychotropic medications
- The study included three evaluation methods:
  - 1) hospital pharmacists and neurologists participated in focus groups regarding pharmacogenetic testing;
  - 2) patients who received pharmacogenetic testing during the study period completed surveys to assess their perception of these tests; and
  - 3) community pharmacists, who received a copy of these test results, responded to a survey on their perception of the tests

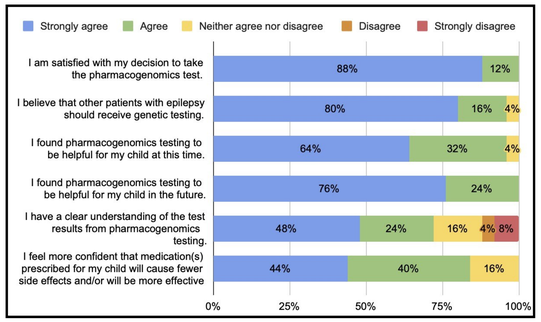
Results

Focus Groups

- Eight clinicians, including four hospital pharmacists and four neurologists, were included;
- All participants had previously used the Precision Rx test in practice, with the exception of one pharmacist who was given access to a sample report;
- Participants' exposure to pharmacogenetic testing prior to the study was limited;
- Three major themes were identified, along with their respective subthemes.

Themes	Subthemes and Summary of Findings
Receptiveness to pharmacogenetic testing	**Utility of pharmacogenetic testing**: A majority of clinicians agreed that pharmacogenetic testing was useful in their practice, although few had the opportunity to modify treatment decisions based on the results during the study's timeframe. **Clinicians' comprehension of test results**: Time was required to fully understand test results, but participants believed the results reports were clear and comprehensible. **Communication with the parents**: Some parents expressed concern regarding the potential impact of testing on their child's future health insurance coverage. However, they were very interested in the results and appreciated the follow-up provided.
Test characteristics	**Presentation of test results**: Some clinicians found the report too long and found some information provided concerning other drug classes less relevant for children with epilepsy, such as antivirals. The completeness of results was appreciated by others. **Which genes to test**: A panel would be more advantageous than testing specific genes. **Time to obtain results**: Opinions regarding the acceptable delay to obtain results were variable: some stated that there is no urgency in receiving test results whereas others would prefer to have them within a week. **Cost of testing:** Test reimbursement was identified as a barrier to pharmacogenetics.
Integrating pharmacogenetic tests into practice	**Which patients to test**: Selecting specific patient populations for testing would be favoured over broad systematic screening. **Workflow integration**: Some stated that tests were simple to integrate in their practice; however, others found that the process was burdensome, particularly to obtain parents' consent and explain the test initially. **Long-term responsibility**: Test results would be useful to multiple specialties and health care professionals; therefore, implementation would require structure and cohesion on an organizational level.

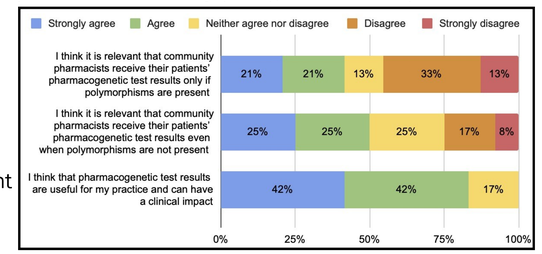
Patient Surveys



- Twenty-five of 26 parents responded to the survey (96%)
- The main advantages of testing perceived by parents were:
  - to reduce the number of attempts to find the appropriate treatment (45%; 13/25);
  - to increase medication effectiveness (28%; 8/25); and
  - to decrease adverse events (21%; 6/25).
- Parents' opinion on what is a reasonable cost for pharmacogenetic testing varied

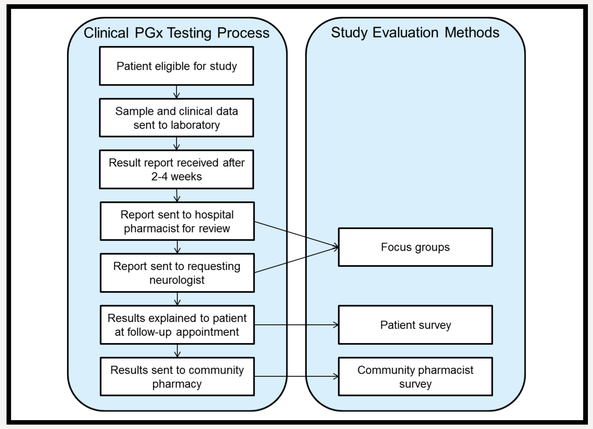
Community Pharmacist Surveys

- Twenty-four of 26 community pharmacists responded to the survey (92%), and few had previous academic training on pharmacogenetics (25%; 6/24);
- All responders stated that they understood the pharmacogenetic report either partly (21%; 5/24) or completely (79%; 19/24);
- It remains unclear whether community pharmacists think test results should be sent to them systematically or only if mutations are present.



Test Documentation

- Test results were documented in 85% of patients (22/26);
- As results were sent by email and manually added into the file; there was a greater chance of results being misclassified or missing from the patient file.



Discussion and Conclusion

- Our study concretely brought forward the use of pharmacogenetic tests directly to clinicians and patients and showed that both clinicians and patients generally favour the implementation of pharmacogenetic testing in the field of pediatric epilepsy;
- Certain facilitators are required for these tests to become more commonly prescribed, including:
  - The reimbursement of these tests by insurance;
  - The inclusion of clinical decision support or pharmcists' interpretation; and
  - The establishment of an organizational structure to ensure efficient long-term use of test results;
- This study has some limitations, notably due to its small sample size, its short follow-up period and the use of a single pharmacogenetic panel; the results can not be extrapolated to other settings or other types of pharmacogenetic tests;
- Local integration of pharmacogenetic testing in practice is an essential step to further clinicians' comfort and knowledge of these tests, to eventually improve patient care and safety on a broader scale.

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