PROJECT INFORMATION

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Testing Two Patient Surveys for Diagnosing Rare Genetic Conditions

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Organization

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What was the project about?

To diagnose rare genetic conditions, doctors look at patients' genetic data and a phenotypic profile. A phenotypic profile is a record of all the physical traits of a condition. It uses a list of standard terms called Human Phenotype Ontology, or HPO. Doctors and clinic staff do a thorough exam with the patient to create the profile. The exam takes a long time and often more than one visit.

Patients may be able to create phenotypic profiles themselves using surveys. These surveys may take less time than clinic visits. But it is unclear whether patient surveys can provide enough details to correctly identify conditions.

In this project, the research team tested two surveys:

- **Phenotypr.** This survey asks patients to describe their symptoms and then matches the descriptions to plain language HPO or clinical HPO terms.
- **GenomeConnect.** This survey uses multiple choice questions to ask patients about their health and symptoms.

What did the research team do?

First the research team used a computer program to create phenotypic profiles from the surveys for all 7,344 known genetic conditions. For each condition, the program mimicked how 20 patients would respond to the surveys. The team checked these profiles

against published profiles for each condition. Published profiles include a standard set of typical symptoms for each condition.

Then the research team tested the surveys with 282 patients. Patients had 1 of 257 different rare genetic conditions. The team assigned the patients by chance to take one or both surveys. The team also interviewed 17 patients who took both surveys to see which one they preferred.

Patients, doctors, and a genetic counselor helped design the study.

What were the results?

Both surveys were able to accurately identify rare genetic conditions. When comparing published profiles with survey profiles, use of the plain language HPO had more exact matches.

Patients preferred GenomeConnect over Phenotypr.

What were the limits of the project?

It was hard to compare how well the surveys worked because the number of patients with each condition was small.

Future research could test the surveys with a larger group of patients with rare genetic conditions.

How can people use the results?

Clinics can use these results when considering how to create phenotypic profiles to diagnose rare genetic conditions.

To learn more about this project, visit www.pcori.org/HolmHaendel368.