

Outcomes of a two-year national rare disease gene discovery project

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As part of the Finding of Rare Disease Genes (FORGE) research project, Canadian researchers have developed an expertise in understanding the underlying biology of rare childhood disorders. The first public commentary of this nationwide study is available today in the *American Journal of Human Genetics*.

"When we launched this project, we predicted we might explain, or solve, 50 disorders; we've almost tripled that goal," said Dr. Kym Boycott, lead investigator of FORGE and clinician scientist at the Children's Hospital of Eastern Ontario (CHEO). "This was a leadership opportunity for Canada. Our team's rare disease expertise is now sought on an international stage – and it started with FORGE."

264 rare disorders were submitted by geneticists from across Canada and selected for the FORGE study. With a 55% success rate, the research team has already solved 146 disorders, including identification of 67 novel genes that had never been associated with a rare disease before.

The pan-Canada research team studied a wide range of childhood genetic disorders including neurodegenerative conditions and those that affect multiple systems in the body. They noted common biological pathways that crossed multiple rare disorders. This means that certain pathways may make better targets than others for designing therapies to treat [rare diseases](#), according to Dr. Boycott.

The technology they used to conduct this research is called exome

sequencing. It's a sophisticated, lab-based method of testing all genes at once vs. traditionally testing one gene at a time. Using this approach, the researchers were surprised by how many genes they identified that were already known to cause rare disease, but were missed in the same families using traditional testing.

"The rewarding part of this project for all involved has been the many lives we've impacted," said Chandree Beaulieu, project manager of FORGE and first author of the report. "Every result was reported back to the families who participated in the research. The information never stayed in a lab or database. This was highly motivating for the entire team."

Dr. Boycott explains that delivering an evidence-based diagnosis to families can influence disease management: by informing what progression of the rare disease might look like; adjusting treatment by adding or removing a particular therapy; and, being able to offer informed reproductive counselling.

The FORGE project has now rolled into an international research program called CARE for RARE, also led at CHEO. The team's objective is to continue the quest to identify more rare disease genes, but also to design and test therapies in the lab to treat specific rare diseases.

More information: For more information about CARE for RARE, visit: care4rare.ca/

Provided by Children's Hospital of Eastern Ontario Research Institute

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