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Finding of Rare Disease Genes in Canada (FORGE Canada)

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Research Funding Program:
Advancing Technology Innovation through Discovery

Genetic disorders of children are individually rare but collectively frequent, affecting the lives of approximately 500,000 children in Canada. These disorders cause a variety of medical problems including birth defects, intellectual disability, difficulty with growth and organ failure. Most genes that cause these conditions have not yet been found, mainly because gene-discovery studies are difficult to perform when DNA from only a small number of affected children is available.

Researchers have created a large network of Canadian doctors and scientists who will now have access to next-generation sequencing. Through this national collaboration, researchers will be able to rapidly identify many genes responsible for genetic disorders that affect children in this country and throughout the world. The Canadian Pediatric Genetic Disorders Sequencing (CPGDS) Consortium (www.cpgdsconsortium.com) has 150 members and will ensure that Canada becomes a world leader in this exciting field. The Consortium brings together doctors from all genetics centres across Canada, internationally-recognized Canadian scientists with expertise in finding genes, and teams from the three Genome Canada Science and Technology (GC S&T) Innovation Centres (Montreal, Toronto, Vancouver), which have already set up the new sequencing technology.

The CPGDS Consortium will:

1) Assist doctors to identify patients with rare childhood diseases. Because the Consortium has members from all the medical genetic clinics in our country, for any given disorder we will be able to enroll children and families from across Canada. Therefore, even for very rare conditions, we will be able to find disease-causing genes. So far, over 100 genetic disorders that affect Canadian children have been submitted for study.

2) Sequence the genomes of patients to identify disease-causing genetic changes.

3) Set up a national data coordination centre to streamline and improve existing large-scale sequence analysis tools. This will improve our ability to distinguish genetic changes that cause disease from ones that are normal variants contributing to human diversity.

4) Create national ethical guidelines for analyzing sequence data from entire genomes and for sharing results with families.

The consortium will allow for rapid gene discovery of rare childhood-onset disorders, with immediate and long-term health benefits for Canadian families. Discoveries in this project aim to lead to genetic tests that will allow earlier and more precise diagnoses. Better diagnoses will allow Canadian health care teams to reduce or prevent patient complications, to develop tailored treatments, and to provide more accurate reproductive counselling to families. In the long term, identification of disease genes is an essential step toward the development of drugs that will one day improve the lives of affected children.