S Ontario Genomics



For years, Ontarians have been hearing about our crumbling health care system but there are many silver linings to the cloudy situation our hospitals are dealing with.

One of them is the groundbreaking work
Dr. Bekim Sadikovic at Lawson Health
Research Institute and London Health
Sciences Centre has been doing with
biotech company, Illumina. In their quest to
better detect rare diseases, Dr. Sadikovic
has created the clinically validated EpiSign™
test, which uses machine-learning
algorithms and compiles them into the
EpiSign™ Knowledge Database.



1 in 15

Canadian children born with a rare disease



1 in 4

Pediatric hospital beds is occupied by a child with a rare disease



4,000

Rare genetic diseases



EKD

EpiSign™ Knowledge Database The world's largest rare disorder DNA methylation database

HEALTHY **PEOPLE**, HEALTHY **ECONOMY**,
HEALTHY **PLANET** THROUGH **GENOMICS INNOVATION**



Ontario Genomics



Thanks to funding from Ontario Genomics, Genome Canada and their Genomic Applications Partnership Program (GAPP), the EpiSign™ project is helping doctors cut down on long and agonizing diagnosis wait times by efficiently bringing answers and treatment options to more of the 1 in 15 Canadian children born with a rare disease.

Not only does this mean peace of mind and quicker access to the right support for those children, it's critical relief to our health care system by speeding up the diagnosis process and easing the pressure on hospitals since 1 in 4 pediatric hospital beds are occupied by a child with a rare disease.

This incredible work has been an evolution. After all, projects like these don't happen overnight! It takes many years of collaboration and many rounds of funding. When Dr. Sadikovic's team applied for their first round of GAPP funding in 2019, they were able to detect 19 different disorders across 30 genes. Five years later, the

most recent version of EpiSign™ has 116 different indicators covering 126 disorders. This number will only continue to grow in the years to come and while this much needed innovation was created here in Ontario, it'll be a benefit to the rest of the world.



Dr. Bekim Sadikovic at Lawson Health Research Institute and London Health Sciences Centre











