



Conscience Announces Support for Two Projects Advancing Earlier Diagnosis and Treatment of Childhood Rare Diseases

CureSPG50 and SickKids to pursue Phase 1/2 clinical trial of potential gene therapy for CLN7 Batten disease in Canada

ThinkRare to expand AI-driven approach to help identify children with undiagnosed rare genetic diseases

Toronto, ON, Canada, October 6, 2026 — [Conscience](#), a Canadian non-profit dedicated to enabling drug discovery through open science and collaboration for the advancement of accessible treatments, is proud to highlight two recently approved projects focused on earlier diagnosis and intervention while advancing the development of therapies for rare diseases in children. The first project is a multi-centre Phase 1/2 clinical trial of a gene therapy for CLN7 Batten disease, a fatal ultra-rare genetic disorder affecting children, conducted by [Elpida Therapeutics](#). [CureSPG50](#) and [The Hospital for Sick Children](#) (SickKids) have partnered with Elpida to bring the clinical trial to Canada. The second project is the deployment of [ThinkRare](#) across Canadian pediatric hospitals. This AI-driven, rule-based algorithm, which was developed at the [CHEO Research Institute](#) and used in clinics at [CHEO](#), analyzes electronic medical records to help identify patients who may have an undiagnosed rare genetic disease. The projects are being supported through two of Conscience's programs: Developing Medicines through Open Science ([DMOS](#)) and AI-Driven Medicines ([AIM](#)), respectively.

"Rare diseases can be incredibly difficult to diagnose and even harder to treat, especially in children. Too often, promising scientific advances struggle to reach patients because conventional drug development and funding models are not designed for very small patient populations and the perceived limited market opportunities that they represent" said Peng Fu, CEO of Conscience. "New approaches are needed to move breakthrough science forward more efficiently, derisk early innovation, and help ensure commercial viability so that patients can ultimately benefit. At Conscience, we are building a portfolio of initiatives that harness the power of AI and open science to address these barriers. Supporting these two projects reflects this broader vision:

bringing together the right tools, data, expertise and partners to shorten the path from scientific discovery to meaningful treatments for patients.”

”Rare diseases sit at the intersection of everything Conscience was built to address: high unmet need, limited commercial incentive, and enormous potential for collaboration to change outcomes for patients,” said Anne Fortier, VP, Drug Discovery and Development at Conscience. ”AI and open science are proving to be powerful partners across the continuum of care, from enabling earlier diagnosis to advancing new gene therapies. We’re excited to see both of these projects progress.”

Developing Medicines through Open Science (DMOS) Project: Clinical Development of AAV9/MFSD8 for the treatment of CLN7 disease

CLN7 Batten disease is caused by mutations in the MFSD8 gene. These mutations prevent the body from producing a fully functional protein needed to support healthy neurons in the brain. As the disease progresses, this disruption can affect how cells function and lead to developmental delays, vision loss, seizures, loss of mobility and, ultimately, death by late childhood. There are currently no treatments available for children living with CLN7 Batten disease.

With support from funders, including \$768,823 from Conscience over a two-year period, CureSPG50 and SickKids have partnered with Elpida Therapeutics to bring their Phase 1/2 clinical trial of a gene therapy for CLN7 Batten disease to Canada. The therapy is designed to deliver a functional copy of the MFSD8 gene directly to the brain, enabling cells to produce the protein needed to support healthy neuronal function. A Phase 1 clinical trial involving four patients was completed previously, demonstrating that the gene therapy was safe and providing early evidence of potential clinical benefit. The proposed Phase 1/2 clinical trial would expand the study to an additional nine children living with CLN7 Batten disease, bringing the therapy one step closer to potential regulatory approval and, ultimately, access for patients beyond the clinical trial setting.

“For families facing CLN7 Batten disease, hope is not about simply gaining more time. It is about giving a child the chance to keep walking, talking, playing, learning and being themselves for as long as possible,” said Terry Pirovolakis, founder of Elpida Therapeutics. “We believe gene therapy has the potential to fundamentally change the trajectory of this devastating disease. Our goal with this trial is bigger than demonstrating that a treatment can be delivered safely, we want to show that we can meaningfully alter the course of CLN7 and give families something they have never had before: a real reason to believe their child’s future can be different. And if we can prove that model here, we believe it can help open the door for children living with many other ultra-rare neurodegenerative diseases.”

Pirovolakis's work in rare disease began after his son Michael was diagnosed with SPG50, an ultra-rare neurodevelopmental disease. He founded CureSPG50 and worked with biomedical experts to develop Melpida, a novel gene therapy for SPG50. Its success led to the creation of Elpida Therapeutics, a 501c3 US Charity applying the same approach to other rare diseases, like CLN7 Batten disease.

AI Driven Medicines (AIM) Project: ThinkRare: Harnessing AI for the early identification of rare genetic diseases

Developed at the CHEO Research Institute, ThinkRare uses AI to analyze electronic medical records and identify patients who may have an undiagnosed rare genetic disease. Flagged patients at CHEO can then be reviewed by genetics specialists and referred for appropriate genetic evaluation, with results validated through detailed medical-record review and subsequent diagnostic follow-up.

Unlike screening tools designed to detect a single condition, ThinkRare is designed to identify thousands of rare diseases simultaneously, using expert-curated clinical criteria combined with patterns in electronic medical records to help genetics specialists identify patients who may otherwise go undiagnosed. In a proof-of-concept study, ThinkRare demonstrated a negative predictive value of 99.9 per cent, indicating a high level of confidence that patients who are not flagged are unlikely to have been missed by the algorithm.

ThinkRare has already helped shorten diagnostic journeys for patients with rare diseases, including one patient who had undergone 153 outpatient visits in the past 17 years without receiving an accurate diagnosis. ThinkRare was able to quickly identify and diagnose the patient, highlighting the need for more efficient diagnostic tools for rare diseases. In another example, ThinkRare helped identify a rare disease in a three-month-old infant, demonstrating the algorithm's potential to support earlier diagnosis and intervention and help prevent prolonged diagnostic journeys.

At CHEO, ThinkRare has identified 23 patients who subsequently received rare disease diagnoses. Among patients referred for further assessment and testing, the algorithm achieved a 70 per cent diagnostic yield, helping clinicians reach diagnoses that can inform more targeted medical management and care.

With \$1 million in total funding from Conscience over a one-year period, the CHEO Research Institute plans to expand ThinkRare across additional institutions, with the potential to identify hundreds to thousands of children living with undiagnosed rare diseases. The initiative will also help build a collaborative network of clinicians, researchers, and data scientists.

Implementation plans are already in development at McMaster Children's Hospital, Alberta Children's Hospital, and Stollery Children's Hospital. Next steps include developing disease-specific algorithms, connecting ThinkRare with rare disease registries such as the emerging CONNECT Registry, and studying the economic impact of earlier diagnosis to help inform health policy and government investment.

"Rare diseases present a unique data challenge. While individually rare, they affect millions of people worldwide, and the signals that could lead to a diagnosis are often buried within complex clinical records," said Ivan Terekhov, Director of Research Informatics, AI and Technology at the CHEO Research Institute. "ThinkRare was developed at the CHEO Research Institute to help uncover those signals using AI, enabling genetics experts at CHEO to identify children who may otherwise go undiagnosed. With Conscience's support, we can expand this approach to more pediatric hospitals across Canada, helping more children and families find answers sooner and demonstrating how data and AI can accelerate rare disease diagnosis at scale."

For more information about the DMOS program, visit the [website](#). To learn more about the AIM program, visit the [website](#).

Additional Quotes

"Canadian researchers and innovators are using cutting-edge science and artificial intelligence to tackle some of the most difficult health challenges facing children and families," says the Honourable Mélanie Joly, Minister of Industry and Minister responsible for Canada Economic Development for Quebec Regions. "By advancing new approaches to diagnosing rare diseases earlier and developing promising treatments, projects like these are helping turn Canadian innovation into better health outcomes and new hope for patients. The government will continue supporting the talent, research and collaboration that strengthen Canada's life sciences sector and improve the lives of Canadians." — The Honourable Mélanie Joly, Minister of Industry and Minister responsible for Canada Economic Development for Quebec Regions

About Conscience

Conscience is a non-profit focused on enabling drug discovery and development in areas where open sharing and collaboration are key to advancement towards accessible treatments. It does so by encouraging and funding the open sharing of knowledge and tools, the use and improvement of artificial intelligence, and the development of policies that break down barriers of traditional drug development. Powered by a network that includes academics, industry, technologists, policy experts,

and public support, Conscience seeks to drive innovation by turning drug discovery and development into a team sport. Its open science model brings unique value in areas where market solutions are limited, offering alternatives to traditional intellectual property models to make new accessible medicines so no one is left behind. Through key initiatives, such as its DMOS (Developing Medicines through Open Science) program and CACHE (Critical Assessment of Computation Hit-finding Experiments) Challenges, Conscience is accelerating the path to treatments for those who need them most. For more information, visit conscience.ca.

About the DMOS Program

The Developing Medicines through Open Science (DMOS) program supports drug development in areas of unmet medical needs where open sharing and collaboration are key to advancement towards accessible treatments. Using an open science model, the program aims to foster collaborations that undertake preclinical and clinical work to develop drug candidates in areas of unmet medical need, establish proof of concept for an open science path to drug development and further translate innovations into affordable medicines, generate economic activity, and support small and medium-sized enterprises (SMEs) in Canada. With total funding of \$15M CAD, this program supports projects focused on life-threatening or severely debilitating diseases that have demonstrated strong target validation and tractability to enable clinical proof of concept and undertake either IND-enabling preclinical studies or human safety and efficacy studies. To learn more about the program, including eligibility criteria, please visit the [website](#).

About the AIM Program

The AI-driven Medicines (AIM) program aims to drive transformative research that leverages computational or digital-based methods and tools to address critical, real-world challenges in the discovery and development of accessible and affordable medicines. This initiative focuses on supporting the development of innovative methodologies and tools to accelerate the drug discovery and development pipeline, bridging the gap between advanced computational theory and practical therapeutic applications. To learn more about the program, including eligibility criteria, please visit the [website](#).

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