MEDIA RELEASE

Canada's $1.5-billion Rare Disease Drug Strategy generating 'return on investment' for patients and society but more needs to be done

November 25, 2025 (Ottawa, Ontario) – At the midpoint of the federal government's three-year (2024-27) National Strategy for Drugs for Rare Diseases, the Canadian Organization for Rare Disorders (CORD) is pleased to report that children and adults living with nine different debilitating rare conditions are being treated with drugs that significantly improve and, in some cases, save their lives.

The nine drugs are among at least 12 to be funded by provinces and territories through bilateral agreements in Phase 1 of the national strategy. They are treatments for different types of serious rare disorders:

- **Primary hyperoxaluria type 1 (PH1):** A genetic liver enzyme defect leading to kidney damage
- **Von Hippel-Lindau (VHL) disease:** A condition causing multiple tumours and cysts in different organs
- **Neurofibromatosis type 1 (NF1):** A genetic condition that causes tumours to grow on the nerves
- **Fibrodysplasia ossificans progressiva (FOP):** An ultra-rare disorder where muscles and connective tissues turn into bone, progressively restricting movement and breathing
- **Diffuse large B-cell lymphoma (DLBCL):** DLBCL is an aggressive non-Hodgkin lymphoma that grows quickly and requires urgent treatment
- **Mycosis fungoides/Sézary syndrome (MF/SS):** These are rare blood cancers attacking the skin
- **Large B-cell lymphoma (LBCL):** A fast-growing blood cancer causing rapidly enlarging lymph nodes, fevers and fatigue
- **Bardet-Biedl syndrome (BBS):** A genetic condition disrupting the brain's appetite signaling, causing insatiable hunger and severe obesity
- **Homozygous familial hypercholesterolemia (HoFH):** An inherited disorder that causes extremely high levels of "bad cholesterol," increasing the risk of early-onset heart disease

Dr. Jennifer Adams, whose daughter was diagnosed at 18 months with PH1, calls Oxlumo a "life-changing medication." As a result, her daughter's "quality of life has improved, hospital visits were reduced, and it has likely eliminated the need for costly dialysis or transplants in the future."

Unfortunately, for these diseases and indeed most rare conditions, patients are often diagnosed too late to obtain maximum benefit from treatment.

“A significant portion of the federal funds is supposed to be used for screening and diagnosis,” said Durhane Wong-Rieger, President and CEO of the Canadian Organization for Rare Disorders. “The patient community is committed to working with provinces and territories to set up initiatives to meet the objectives for timely diagnosis and data collection in real-world use to achieve optimal outcomes.”

Survey shows impact on patients and the health system

This lack of screening and diagnosis services results in extensive use of health system resources by patients seeking help for unexplained conditions. A new national survey conducted by Ipsos for CORD found that respondents reported needing an average of 14 visits with general practitioners and specialists before receiving an accurate diagnosis. Additionally, one in four (25 per cent) had at least one inpatient hospital stay in the past year.

“This is very concerning, considering a study in the U.S. that shows that earlier access to diagnosis and treatment is not only life-changing for patients but it reduces long-term costs for families and health systems,” said Dr. Wong-Rieger. “Similarly, a European study has shown that accelerating access to treatments for rare diseases moves the burden away from families.”

As a result, while the first year and a half of Phase 1 of the National Strategy for Drugs for Rare Diseases has focused on funding critical drugs through public plans, the remaining half should be committed to expanding early screening, ensuring timely access to treatment and investing in the specialist networks and data infrastructure that sustain long-term value.

Such actions would set Canada up well for Phase 2 of the national strategy beyond 2027, when the current deals expire. Canada should continue to support direct funding for rare disease drugs but also invest in comprehensive, coordinated care models that are key to reducing inequities and improving outcomes. Governments and health systems should include stronger real-world data collection, incorporating many aspects of healthcare costs to gain a full picture.

“These actions would ensure we can optimize the return on investment of this strategy for patients, their families and Canadian society as a whole,” added Dr. Wong-Rieger.

CORD hosted a breakfast event today on Parliament Hill with federal politicians and government officials to underscore the importance of continuing and enhancing the benefits Canadians are receiving from the national rare disease strategy.

“It’s an investment Canada can’t afford to stop if we want to ensure Canadians with rare disorders get the care they need and deserve,” added Dr. Wong-Rieger.

About the Canadian Organization for Rare Disorders (CORD)
CORD is Canada's national network for organizations representing all those with rare disorders. CORD provides a strong common voice to advocate for health policy and a healthcare system that works for those with rare disorders. CORD works with governments, researchers, clinicians and industry to promote research, diagnosis, treatment and services for all rare disorders in Canada. About 1 in 12 Canadians, or about 3.5 million people, are affected by one or more of thousands of rare disorders. Two-thirds of them are children. For more information, visit www.raredisorders.ca

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