Canadian Organization for Rare Disorders

MEDIA RELEASE

Canadians with rare diseases do not get timely diagnosis and care – with funding and the plan in place, it's time for political leadership

- *New Rare Disease Day survey shows three million Canadians with a rare disease suffer years-long delay to diagnosis and access to specialists, leading to irreparable harm, disability, and early death*
- *"We have the plan, we have $1 billion committed, and we have healthcare professionals ready to go … we just need governments to agree to act" – Durhane Wong-Rieger*

February 28, 2023 (Toronto, Ontario) — More than three million Canadians living with a rare disease have the added heartache of knowing that they, or their child, were diagnosed too late or didn't get the specialist care they needed to avoid irreparable harm or disability. These were among the distressing findings from the latest national survey by Ipsos Canada, commissioned by the Canadian Organization for Rare Disorders (CORD) and released today, Rare Disease Day.

The national Ipsos survey found that significant delays and issues arise for patients and their families in all key areas – including long delays in diagnosis, disease and knowledge information gaps, lack of access to drugs, and lack of coordinated and accessible care.

Canada is the only major developed nation without a comprehensive rare diseases strategy to help patients and families. Canadians have been waiting for years for one to be finalized and implemented. The federal government in 2019 committed new spending of $500 million a year for a national plan for drugs for rare disorders, starting in the current government year. With just one month left, the plan has not been finalized and no money made available to help improve healthcare services, access to drug, and patient group support.

"We know what has to be done, health leaders are ready to move, and we even have the funding allocated but while governments procrastinate, patients and families continue to suffer," said Durhane Wong-Rieger, President and CEO of CORD. "These new survey results confirm that rare disease patients

and families are being left behind, even as governments and health systems prepare to recover from the devastation of COVID."

The new Ipsos survey featured input from over 500 Canadian patients and their direct caregivers, highlighting their challenges and their perspectives on what emerging government strategies should focus on. The overwhelming majority support the creation of specialized centres networked to local healthcare professionals to reach all patients wherever they live in Canada.

Challenges with diagnosis

According to the survey, patients and caregivers felt that their healthcare professionals lacked knowledge of rare diseases, which delayed diagnosis and specialist care:

- Four out of five (80 per cent) do not think family physicians are aware and informed about rare diseases and 62 per cent say the same about pediatricians;
- Almost half (47 per cent) do not believe specialists have up-to-date knowledge about rare diseases;
- Respondents said they waited an average of 3.7 years to get an accurate diagnosis;
- In the process, they saw an average of 5.9 different healthcare professionals and received an average of 3.2 wrong diagnoses.

Information and knowledge gaps

Canadians impacted by rare diseases can often feel lost and in need of connection with others with rare diseases, but face obstacles in doing so:

- Almost two-thirds (63 per cent) say they do not have access to a care coordinator;
- There is a large lack of counselling support for patients with rare diseases, with almost three out of four (74 per cent) saying they did not have counselling support at the time of diagnosis and 70 per cent did not receive appropriate emotional and psychosocial support;
- Less than six out of 10 (58 per cent) feel they have access to all of the appropriate information they need and only 43 per cent say they were given a contact person from whom they could get additional information;
- Less than one in 10 agree that educational, disability and employment services are aware and informed about rare diseases.

Access to treatments

The large number of Canadians with rare diseases have issues related to their access to appropriate drug therapy for their condition:

- Only six out of 10 (60 per cent) say they have access to appropriate drugs for their condition and 63 per cent say they must pay out-of-pocket expenses;
- More than a third (36 per cent) say they are unable to access prescribed medicines because their share of the costs is too high;

- Two-thirds (68 per cent) of Canadians with rare diseases are aware of Health Canada-approved medicines for their condition but 43 per cent cannot easily access prescribed approved medicines;
- A third (32 per cent) are aware of off-label medicines that could help their condition but almost two-thirds (63 per cent) cannot access them.

Need for rare disease centres

Survey respondents were offered two alternatives that have been proposed as key attributes of a national rare disease strategy using the committed federal funding. One is the development of a new national list of drugs for rare diseases and the other is the creation of rare disease centres to specialize in rare disease treatment.

- Four out of five (80 per cent) respondents prefer the development of rare disease centres over a new national list of drugs (20 per cent);
- The most cited reason supporting rare disease centres was that they would offer "comprehensive support, a research centre with access to clinical trials, and be more efficient and accessible for patients";
- Among the minority (20 per cent) who supported a new national list of rare disease drugs, said that it would provide better drug coverage with lower costs and that drugs would be more accessible to all, regardless of location;
- A large majority (84 per cent) favour federal oversight of a rare disease strategy to set standards, facilitate inter-provincial care and improve access to treatment and care because such a policy would provide "more equal access across Canada" and establish "sets of standards and consistent policies." If the provinces maintain oversight, respondents said the federal government should set pre-conditions tied to federal funding.

"These survey results provide important guidance for Canadian policy makers in shaping Canada's rare diseases strategy which is so urgently needed," said Stephen Parrott, Board Chair of Canadian VHL Alliance which advocates for Canadians with VHL, a rare inherited disease that causes cysts and tumours which can be cancerous. "My two daughters have VHL and both lost their adrenal glands. They are now taking steroids for the rest of their lives. They are the lucky ones with VHL, many have it much worse. Canadians with rare diseases and their families have waited far too long and with severe consequences that in many cases could be prevented. We need this strategy implemented without further delay."

In-depth patient/caregiver interviews

The research also included in-depth one-on-one interviews with Canadians with a rare disease or their caregivers. Some of the comments include:

- "You had all these hopes and dreams and expectations of what you could do with your child, for your child, and it just kind of all comes crashing down and then on top of that, because it's a rare disease nobody knows anything about it. You're flying blind."

- "Treating a patient with a rare disease goes beyond access to rare and expensive drugs. Rare disease centres would address a wide variety of gaps that exist in the diagnosis and treatment of rare diseases."
- "It is sad that some people literally move their families across the country so they have access to better care. That is a discrepancy that shouldn't exist in a united country."
- "Everything's on your initiative and your efforts. For rare diseases, you are absolutely on your own."

About the Ipsos survey

To conduct the Ipsos-developed survey of Canadians impacted by rare diseases, CORD emailed an open link to its database. The link was also shared at the Rare Disease 2022 Fall Conference, Nov. 21-22, 2022. In total, 528 respondents completed the 15-minute online open-link survey between Oct. 26 and Nov. 28, 2022. Responses were received from every province, in both languages (90 per cent English) and distributed across urban (47 per cent), suburban (34 per cent) and rural (20 per cent) locations. By the nature of this sample, the data was not weighted. A credibility interval cannot be calculated when an open link is used and when the population universe is unknown. In a second phase, 10 in-depth interviews were conducted with patients and caregivers on their experiences navigating the system. Ipsos is the third largest market research company in the world, present in 90 markets and employing more than 18,000 people.

About Rare Disease Day

Rare Disease Day is the globally-coordinated movement on rare diseases, working towards equity in social opportunity, healthcare and access to diagnosis and therapies for people living with a rare disease. Since its creation in 2008, Rare Disease Day has played a critical part in building an international rare disease community that is multi-disease, global and diverse – but united in purpose. Rare Disease Day is observed every year on Feb. 28 or during leap years on Feb. 29 – the rarest day on the calendar. Rare Disease Day was set up and is coordinated by EURORDIS and more than 65 national alliance patient organization partners, including CORD. For details, visit www.rarediseaseday.org/

About 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. For more information, visit www.raredisorders.ca

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