



# FAQs: Everything you want to know about Canada's Rare Disease Strategy

## Q1. Why do we need Canada's Rare Disease Strategy?

First of all, we believe that now is the time to act to provide hope and treatment to Canadians and their families who are impacted by a rare disease. Right now, the approach to rare diseases is fragmented across the country and this means Canadian families with rare illnesses are facing extraordinary challenges. These include misdiagnosis, unnecessary surgeries, social isolation, financial hardship, lack of treatment options and early death. These are the same challenges faced by Canadians with "non-rare" conditions but the impact is often much more severe.

About 1 in 12 Canadians, two-thirds of them children, are affected by a rare disease. But because each specific rare disease affects only a small number of individuals, scientific understanding and clinical expertise may be limited and fragmented across the country. I encourage you to visit our website and read the Strategy because it tells some of these stories from the perspective of patients and their families. We are asking that patients with rare diseases get the same kind of care and consideration as those with more common illnesses.

## Q2. What are the main components of the Canada's Rare Disease Strategy and how will they help?

The strategy's main components start with are improving early detection and preventing the disease or preventing progression, getting right care to patients as early as possible, enhancing community support, providing sustainable access to promising therapies and promoting innovative research. The strategy components not only cover the spectrum from preventing disease to providing new cures and treatments but as importantly calls for their integration into a comprehensive framework so that research findings will lead to new treatments that can be efficiently available to patients along with other supportive care.

## Q3. We know that health spending has been on the rise and governments are trying to control costs. How much will it cost to implement the Strategy?

Canadians are already paying for rare disorders, but the question is whether we are getting good value. Patients are not getting diagnosed early, they're not getting the right treatments at the right time, and they lack other support from education, workplace, and social systems, Canada does not get the best value. Moreover, individuals and families are paying extraordinary amounts out of their own pockets, often for services that are available to those with common conditions. If people are properly screened, diagnosed and treated, there will be overall savings. Unnecessary surgeries, visits to multiple specialists, organ transplants, among other expenses, will be avoided. So a lot of cost for the Strategy does not have to be new money. Canada does need to make an

investment in rare diseases but it isn't how much but rather how the investment is made.

#### **Q4. The strategy calls for Centres of Excellence and a new Canadian Partnership for Rare Diseases. These will likely entail new costs.**

Canada has already demonstrated a commitment to Centres of Excellence as a way of stimulating innovation and better outcomes in many sectors, including health. A Canadian Partnership for Rare Diseases is based on proven beneficial models like the partnerships in cancer and mental illness. We only need to look at Europe if we want examples of effective Centres of Excellence in rare diseases. Finally, in Canada, Centres of Expertise in haemophilia, cystic fibrosis, and rare blood cancers are outstanding examples of cost-effectiveness of an integrated and comprehensive approach.

#### **Q5. Why should we implement this strategy when rare disorders affect just a tiny fraction of the population?**

Individually, each rare disorder may only impact a handful of Canadians but given that there are over 7,000 rare diseases, together, there are nearly 3 million Canadians living with a rare disorder. Impacts go far beyond each patient, as caregivers, employers and healthcare teams are all impacted. Moreover, even though 80% of rare disorders are genetic, many of these are "spontaneous" genetic mutations that can affect anyone. So, like cancer and mental illness, dealing with rare disorders is public health concern affecting all Canadians. Finally, new knowledge about rare disorders has spill-over benefits to more common diseases, such as understanding genetic determinants of subtypes of common conditions, such as cancer, cardiovascular disease, and inflammatory disease. So we can treat subgroups of patients with exactly the right therapy for their personal genetic make-up, the so-called personalized medicine revolution.

#### **Q6. Who should take the lead?**

Around the world, rare disease patient organizations have taken the initiative to drive everything from identifying the genes that cause their specific disease, sponsoring research into therapies for unmet needs, and engaging stakeholders to develop rare disease strategies. CORD has been calling for national leadership on this file for the last ten years. We have launched this strategy and taken the lead, and we're confident that Canada's elected governments are ready to step up and respond to this call to action. We'll support any effective leadership efforts from the federal and provincial governments that are aligned with our goals and actions.

#### **Q7. We have seen incidences where new drugs to treat these rare disorders can cost hundreds of thousands of dollars. Won't this put a huge strain on already underfunded provincial drug plans?**

Relatively high per-patient costs and prices won't have major budget impacts, as only a low number of Canadians will benefit from these treatments. In fact, drugs for rare disorders are a very small fraction of the overall pharmaceutical budget at around 3%, and a tiny fraction of overall health expenditure. Forecasts suggest very little change in that proportion in the coming years.

### **Q8. Hasn't the federal government taken action already on rare diseases?**

The federal government has committed to launching a regulatory framework for orphan drugs, and this is a step we're expecting very shortly, as they have been consulting with us for several years. The federal government is already stepping up in terms of research investments. We want to help Ottawa work with all health system stakeholders in a coordinated fashion. This strategy is an important step in that direction.

### **Q9. What can provincial governments do?**

This strategy sets out clear actions for the provinces to take to improve the quality of care for their citizens with rare disorders. Provinces already have infrastructure, services, even healthcare professional training that can be better coordinated, funded, and directed to serve the rare disease patients and families. Importantly, provincial and territorial health systems need to collaborate (work together, share resources, exchange best practices) to avoid duplication, unproductive redundancy, and discontinuities. They must work together to set up patient registries, professional training, patient and public education, and supportive services. They can also improve joint initiatives through existing agencies such as the Health Care Innovation Working Group and the Canadian Agency for Drugs and Technologies in Health.

### **Q10. How did CORD go about preparing this Strategy?**

This strategy is the result of cross-Canada consultations and contributions from a wide range of stakeholders, including governments, researchers, individual patient organizations, policy experts and others.

### **Q11. There are a lot of recommendations in the Strategy. Which ones are the most important?**

First, in terms of timing and priority, is the implementation of the federal Orphan Drug Regulatory Framework, because it is the cornerstone for a definition of rare diseases and will allow Canada to be partner in the research, development, and approval of rare disease drugs at the same time as Europe and the USA.

Second, along with the regulatory framework is the need for a tailored evaluation and funding approach to ensure timely and equitable patient access to orphan drugs. All countries are struggling with this issue and Canada can be part of evolving solutions and best practices. This is essential.

Third, a key-integrating factor is the Centre of Excellence, to generate and support research and patient care, which could be linked through a new Canadian Partnership for Rare Diseases or reference network. While this is not an easy component to implement, it needs to be planned for while we are planning for and implementing all others. Otherwise, we will never get to a comprehensive approach.

Fourth, provide dedicated and increased research funding for rare diseases, potential through Public Private Partnerships. At the same time, allocated resources for patient organizations and ORPHANET Canada is one of the few developed countries that do not provide some support for patient groups, to

contribute to the health system, to support the work with their patient community, and to do outreach and awareness.

And finally, adopt national program for Newborn Screening with clear guidelines for adding new diseases based on evidence and international best practices (USA) and implement state of the art DNA and genomic screening with “core sites” and national outreach.

### **Q12. There are a lot of patient organizations that are adopting strategies these days. Why should we pay attention to this one?**

Many patient organizations are striving for strategies because coordinated action, strategic frameworks, and partnerships are recognized as effective and cost-effective approaches. Beyond that, the rare diseases experience disproportionate and extraordinary challenges, including wide gaps in access to healthcare compared to Canadians with more common conditions. Almost every other developed country has recognized this need and adopted appropriate strategies and policies. Canada can and needs to do more, and that’s why we all need to pay attention and support this strategy.

### **Q13. What are your next steps on this strategy?**

CORD is going to do three things. First, we’ll work with stakeholders ready to take action immediately, and many are already doing so. Second, we’re going to take this on the road across the country in order to build awareness, gain feedback and earn the support of other health system stakeholders. We’re going to have a series of panels in several cities in the early fall. Finally, we’re going to monitor and report on progress and update this strategy regularly, so that we can track and celebrate progress.

**Canadian Organization for Rare Disorders**  
**151 Bloor Street West, Suite 600**  
**Toronto, Ontario M5S 1S4**  
**Tel: (416) 969-7464**  
**Email: [info@raredisorders.ca](mailto:info@raredisorders.ca)**  
**[www.raredisorders.ca](http://www.raredisorders.ca)**