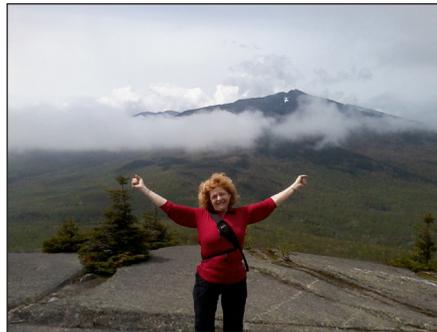




Canadian Organization  
for Rare Disorders

# Now is the Time: A Strategy for Rare Diseases is a Strategy for all Canadians

## EXECUTIVE SUMMARY: CANADA'S RARE DISEASE STRATEGY



### GOALS

1. Improving early detection and prevention
2. Providing timely, equitable and evidence-informed care
3. Enhancing community support
4. Providing sustainable access to promising therapies
5. Promoting innovative research

## Why Action is Needed Now

While each rare disease affects only a small number of individuals, there are more than 7,000 rare diseases that together affect 1 in 12, or nearly 3 million Canadians. However, awareness, knowledge and treatment of most of these diseases are still limited and fragmented across the country. As a result, individuals face a **host of extraordinary challenges**, including:

- misdiagnosis and/or long, difficult path to diagnosis
- no or limited treatments
- barriers accessing available rare disease therapies
- difficulties finding physicians or clinical centers with experience treating rare diseases
- challenges accessing medical, social or financial services or assistance
- feelings of isolation and of having been “orphaned” by the Canadian healthcare system.

All of these challenges lead to **increased morbidity, loss of life or poorer quality of life and increased costs to the family, the healthcare system and ultimately the Canadian economy.**

The Canadian Organization for Rare Disorders (CORD), as the umbrella organization for rare disease patients and patient organizations, acknowledges the courage, commitment and creativity shown by the community to date in tackling these challenges. However, so much more needs to be done by all of us. This is why CORD is saying “now is the time” for governments and all stakeholders to coordinate their efforts as proposed in Canada’s Rare Disease Strategy. All patients deserve the **same timely and quality health and social care regardless of what they suffer from or where they live.** As importantly, this Strategy benefits all Canadians, not just those with rare diseases.



## Strategic Goals and Action Plan

Further to extensive stakeholder engagement, CORD is proposing the following **five-goal strategy**, which includes recommended actions directed **at improving the lives of people with rare diseases.**

*Goal #1: Improving early detection and prevention*

1. Adopt a national approach to **newborn screening**
2. Implement **early detection and preventive services** across Canada

*Goal #2: Providing timely, equitable and evidence-informed care*

3. Improve education and capacity of **healthcare providers** related to rare diseases, including genetic counsellors
4. Address **gaps in social care programs** for people with rare disorders
5. Develop provincial guidelines to ensure **appropriate accommodation** for people with rare diseases in the **workplace**
6. Provide people with rare diseases the same **coverage for healthcare services** (e.g., physiotherapy) as people with more common diseases

7. Establish **Centres of Excellence on rare diseases** to generate and support research and patient care, develop and implement clinical practice guidelines, develop and provide professional and patient education to general healthcare practitioners and the public, and develop and support extended diagnostic, clinical and educational services, for example, through telemedicine or satellite specialized clinics
8. Explore the creation of a **national registry for all rare diseases**, and support new and existing **disease-specific registries**
9. For diseases where specialized clinics and virtual clinical networks may not be feasible, ensure better integration of care for patients with rare diseases into existing **Complex Care Clinics or medical homes**
10. Adopt measures to facilitate **linkages between healthcare administrative databases** across the country to support health service delivery to patients with rare diseases

*Goal #3: Enhancing community support*

11. **Rare disease-specific patient organizations, as well as CORD and the Regroupement Québécois des Maladies Orphelines, should be adequately funded** to achieve their missions, which include involvement in research initiatives, knowledge translation, policy development, education, engagement and support initiatives for patients
12. Increase resources to optimize the utility of **Orphanet for all stakeholders**

*Goal #4: Providing sustainable access to promising therapies*

13. Implement a **regulatory framework** for orphan drugs
14. Explore **adaptive clinical trial designs** for market authorization and post-market phases of therapies
15. Enhance and formalize the **role of patients in the market authorization process** and post-market evidence-generation and provide resources to support the participation of rare disease patient groups in this process
16. Establish a **separate, more flexible health technology assessment process** tailored to the specific attributes of orphan drugs
17. Provide increased **support to assist rare disease patient groups in engaging in health technology assessment reviews**, including in preparing patient input submissions
18. Develop a consistent **funding approach** to ensure timely and equitable patient access to orphan drugs

*Goal #5: Promoting innovative research*

19. Provide **dedicated and increased funding for rare disease research** and the Centres of Excellence on rare diseases
20. Establish a new **Canadian Partnership for Rare Diseases** to help coordinate a national rare disease research agenda and Centres of Excellence on rare diseases, among other actions recommended throughout this strategy

For the full version of Canada's Rare Disease Strategy, see: [www.raredisorders.ca](http://www.raredisorders.ca)

## Looking Ahead for Positive Change

CORD will be **monitoring progress against these five goals** with all stakeholders. Our shared success will be celebrated when policymakers from all levels of government and the rare disease community can point to **specific initiatives that have improved the lives of Canadians** affected by these diseases.

## Background Information

### What is a rare disease?

In Canada, while no formal definition has yet been established, Health Canada has been informally defining the term as: *“a life-threatening, seriously debilitating, or serious chronic condition that only affects a very small number of patients (typically less than 5 in 10,000 persons)”*.<sup>1</sup> It is estimated that they affect 1 in 12 Canadians and that about two thirds of those living with rare diseases are children.

### About 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.

CORD believes that by working together on these five goals, we can, collectively, **bring real positive change** in how we care for people with rare disorders in this country.



### About Canada’s Rare Disease Strategy

We are grateful to the Canadian Institutes for Health Research, Genome Canada, BIOTECanada, Canada’s Research-Based Pharmaceutical Companies (Rx&D), Care4Rare, PRISM, the Rare Disease Foundation and so many other organizations and individuals who have contributed to the working documents and consultations, from which CORD developed this strategy. This strategy is endorsed by CORD’s board of directors but is not intended to represent the official position of any of these other organizations.

1. See [http://hc-sc.gc.ca/ahc-asc/media/nr-cp/\\_2012/2012-147a-eng.php](http://hc-sc.gc.ca/ahc-asc/media/nr-cp/_2012/2012-147a-eng.php)