Rare Disease Day is an occasion that inspires action and solidarity to address the challenges and opportunities of rare diseases. In Canada, approximately 1 in 12 people have a rare disease, many of whom are children. There are many different rare diseases that affect patients, their families, and caregivers; however, treatments are available for only a small percentage of rare conditions. To address the challenges faced by patients with rare diseases, in March 2023, the Government of Canada announced a National Strategy for Drugs for Rare Diseases (National Strategy), with an investment of up to $1.5 billion over 3 years. The National Strategy aims to increase the accessibility and affordability of drugs for rare diseases (DRDs) for patients across Canada by actioning across 4 broad pillars to seek national consistency in coverage for DRDs, support patient outcomes and sustainability, collect and use evidence, and invest in innovation. The first phase of the National Strategy will be implemented over 3 years — from 2023 to 2026 — and will involve coordinated actions by Health Canada, the provinces and territories, and various health system partners, including Indigenous partners, patient groups, clinicians, pan-Canadian health organizations, health research institutes, and industry.

Each pillar under the National Strategy is led by key health partners. CADTH-led initiatives are primarily related to improving the collection, use, and quality of evidence to inform and support decision-making by working with health systems partners on the following key initiatives:

- Monitor and identify new health innovations on the horizon in rare disease conditions to alert decision-makers and support systems readiness.

- Build on existing work and develop pan-Canadian guidance to support newborn screening programs, which play an important role in the early diagnosis of rare diseases. A Newborn Screening Advisory Panel has been established to develop guidance around various issues, including a common set of guiding principles for newborn screening in Canada, a proposed process and criteria for the addition or removal of conditions, and a recommendation for conditions for which newborn screening programs in Canada could screen. In addition, when appropriate, identify the potential need for additional evidence on emerging newborn screening tests through CADTH’s existing health technology assessment (HTA) infrastructure. This guidance is intended to foster greater consistency and timelier access to treatments.
• Provide agile and responsive advice to decision-makers through customized reviews, such as the recently completed non-sponsored review of eltrombopag used for patients with severe aplastic anemia — a rare blood disorder that affects approximately 2 people per million in North America and Europe.

• Undertake a series of activities that will help lay the foundation for improved evidence generation and access to real-world data from rare disease–based registries. Our goal is to help generate fit-for-purpose decision-grade real-world evidence to better address regulatory, HTA, and payer evidence gaps throughout the drug life cycle. These activities will include establishing an inventory of rare disease registries in Canada, establishing registry standards and guidelines, and testing existing registries for HTA readiness.

In summary, these initiatives set the foundation toward supporting those affected by rare diseases and help to advance the 4 pillars of the National Strategy for future phases.

References