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To seek this information, please contact: info@ccso.ca

CRITICAL CARE SERVICES ONTARIO  
LuCliff Place, 700 Bay Street, Suite 1400  
Toronto, Ontario M5G 1Z6  
Telephone: (416) 340-4800 x 5577  
Email: info@ccso.ca  
Website: https://www.criticalcareontario.ca
Section 1.0 – Executive Summary

Rare diseases are defined as serious, chronic conditions that are debilitating or life-threatening. They affect 1 in 12 or nearly 3 million Canadians. To date, approximately 7,000 rare diseases have been identified and new rare conditions are being discovered each year. Since there are a relatively small number of patients with each rare disease, understanding of potential diagnosis and possible treatment options are often limited for many doctors. Patients with rare conditions are frequently referred to several specialists before a correct diagnosis can be made, and this delay in clinical care can have overall negative effects on patient’s health. Other challenges for these patients include limited availability of support in the community, access to participation in clinical trials and difficulty obtaining expensive medications and therapies.

Worldwide, many countries have recognized that people with rare diseases systematically experience barriers to accessing health care. In other jurisdictions, strategic plans have been developed to improve access to care and management for patients with rare diseases. These plans address the needs of the patient community, often include access to multiple care providers, and propose new or revisions to existing policies to streamline access and care delivery. All national plans have a fixed time-frame to achieve their goals.

In Ontario, various clinical programs and services have developed over time to address the diagnosis and management rare diseases, however patients and clinical experts report that services are not comprehensive or well-integrated. This has led to uneven delivery of care for this patient group. This is a reality for both paediatric and adult populations. Availability of clinical programs and services in non-urban areas is particularly limited.

In an effort to address equity and access challenges for this patient population, The Canadian Organization for Rare Disorders (CORD) prepared a report for rare diseases in Canada that describes the current gaps and challenges facing Canadians with rare diseases and makes recommendations on steps forward. Building on this work, the Ontario Ministry of Health and Long-Term Care (MOHLTC) struck a Working Group tasked with developing a rare disease framework for Ontario. The following report presents the work undertaken by this Working Group. The Working Group has made recommendations aligned with the strategic goals of the CORD report, adapted to the Ontario. It is important to acknowledge that caring for this patient population is uniquely dependent on research (frequently with limited published evidence).

This report provides a series of nineteen recommendations aimed to improve the delivery of health care for Ontarians living with a diagnosed or undiagnosed rare disease. The recommendations are not targeted at any one organization, specifically. It was the position of the Working Group that collaboration between multiple branches of the MOHLTC, co-ordination across various health care sectors, and leadership from sponsor organizations (hospitals, research, industry and patient advocacy groups) would be needed to achieve positive change in caring for Ontarian’s living with rare diseases.
The recommendations can be summarized into three main framework elements:

1. **To improve access to diagnosis and care for people living with rare diseases, establish a comprehensive network of health care organizations and providers.** System leadership should be provided from paediatric and adult main hubs (serving as the centre of reference), with formal links to regional sub-hubs (regional genetic service centres). Regional genetic service centres may act as clinical and educational resources to spokes (primary care and other specialists), in collaboration with rare disease patient organizations. This “hub and spoke” organization of services and providers will facilitate effective patient pathways, knowledge translation and ongoing education for the medical community and patients and their care givers.

   - Related recommendations: 3, 4, 5, 6, 7, 8, 10, 15, 17

2. **To support equity in service provision to people living with rare diseases (diagnosed and undiagnosed), ensure that existing diagnostic capacity and services provided are needs-based and designed to be inclusive and comprehensive.** Services include support for expensive drugs, complex home care and caregiver respite, complex care transitions between paediatric and adult settings. Awareness and capacity building with various agencies will be required to achieve equitable access to these services. Supporting organizational and MOHLTC policies may require review and revision to accommodate.

   - Related recommendations: 2, 9, 11, 13, 14

3. **Foster innovation in continued rare disease discovery, management and treatment.** This can be achieved by supporting existing registries and establishing an Ontario-specific rare disease registry, in addition to enhancing collaboration across research institutions, government and industry.

   - Related recommendations: 1, 12, 16, 18, 19
Section 2.0 Background

2.1 Introduction

Rare diseases are defined as serious, chronic conditions that are debilitating or life-threatening. They affect 1 in 12 or nearly 3 million Canadians\(^1\). To date, approximately 7,000 rare diseases have been recognized, and new conditions are being discovered each year. Approximately 80% of rare diseases have a genetic basis\(^2\) and many symptoms of rare diseases present early in life and are diagnosed in childhood.

Patients with rare diseases face unique challenges due to rarity of their condition. As symptoms of a rare disease present, the first doctors these patients visit such as primary care providers, paediatricians emergency medicine and other specialists may not recognize the symptoms as a rare disease. For many patients with rare disease, the underlying etiology may remain unidentified, despite multiple investigations\(^3\). In fact, patients may be assigned an incorrect diagnosis and be referred to several specialist physicians until a correct diagnosis can be made. Because of the time and effort it can take to obtain a diagnosis and treatment plan, with the average time of 7 years, this process is referred to as the diagnostic “odyssey”. As the diagnostic odyssey is lengthy, the delay in obtaining appropriate clinical care, including treatment, may have serious consequences on the patient’s health\(^4\). Some patients live with diseases that have not yet been described, and without a diagnosis can endure the debilitating effects of the disease process. Since the large majority of rare diseases are genetic in origin, a correct diagnosis may impact not only the patient’s care but may have further implications for management and/or counselling of family members as well.

There are a number of challenges for primary care physicians managing a patient with a rare disease. The relative infrequency of these patients in their practice and relative lack of available resources on managing a given rare disease usually leads to a reliance on specialists to help co-manage the clinical needs of the patient. Patients and caregivers have reported that there is limited access to support groups, participation in clinical trials, clinical resources for specialized treatment and other resources such as social and financial assistance\(^5\). Lack of access to these resources is of particular burden to patients and their families living with complex needs that may include 24/7 monitoring, ongoing intensive physical and developmental therapies, accommodations in the education system and sometimes specialized, expensive medications.

In Ontario, expertise exists to discover, diagnose and manage rare diseases. Various clinical programs and services have evolved over time, but remain fragmented and challenging for clinicians and patients to access. This fragmentation has led to a disparity in equitable delivery of care for patients living with rare conditions. Specifically, inequities have been noted between available paediatric and adult rare disease programs, along with access to services between urban and non-urban areas of the Province\(^6\).

This Report presents a review of other jurisdictional approaches to rare diseases and similar initiatives within Canada. Current assets and potential gaps identified by the Working Group and through stakeholder engagement are documented. This work concludes with a series of system-level
recommendations for Ontario which align with the goals of the rare disease report developed by Canadian Organization for Rare Disorders (CORD).

Global Landscape
There is no universally accepted definition for rare diseases, or specific identification of which diseases are considered rare, and which are not. The designation of rarity for a disease is most commonly defined based on prevalence and incidence within a jurisdiction, or in some cases by a combination of factors based on severity and the existence or feasibility of alternative therapeutic options. A review of legislation, regulations, and policies related to rare diseases indicates that 18 OECD (Organization for Economic Co-operation and Development) countries have established national strategies to address rare disease populations. These strategies generally comprise of a set of integrated and comprehensive health and social policy actions to be developed and implemented at the national level. A time-frame is defined within which the identified objectives of the strategy should be achieved and accordingly resources are assigned to ensure effective development, implementation, monitoring and evaluation of the strategy.

Globally, government agencies have identified the following areas of focus in their national strategies aimed at improving the delivery of health care for the rare disease population:

- Develop and disseminate information to raise awareness of the impact of rare diseases on patients, families, health professionals and their communities.
- Develop regulatory frameworks for access to treatment services, patient advocacy and research stimulation.
- Improve access to early diagnosis, timely intervention, expert advice and, coordinated care for rare disease patients and developing referral pathways for rare disease patients to facilitate efficient care delivery.
- Improving provision of treatment options for rare disease patients by establishing regulatory strategies to improve the availability of orphan drug treatments (e.g. research incentives, marketing authorization procedures) and developing strategies to improve access to orphan drug treatments (e.g. price regulation, reimbursement through national health plans).
- Provide educational resources and knowledge exchange opportunities to health professionals to allow them to better identify, manage and treat rare diseases.
- Support research initiatives with the objective to better understand and treat rare diseases. Some of the steps taken in this direction may include developing national and international multidisciplinary research partnerships, effective knowledge translation activities, adequate funding/grant opportunities for promising research projects, incentivizing pharmaceutical companies for drug development, increasing epidemiological knowledge of rare diseases and their impact and, developing and contributing to rare disease registries.
- Develop, sustain and support integrated peer support networks, patient organizations and interest groups to ensure that rare disease patients, their family/caregivers do not feel isolated and have a resource for advice and support to make informed decisions about their condition.
The scope of this review is limited to published and gray literature available in English. An overview of individual jurisdictional strategies is presented in Appendix D.

**Canada – Rare Disease Initiatives**

To support the development of an Ontario Rare Disease Strategy, The Ministry of Health and Long-Term Care (MOHLTC), communicated with Ministries responsible for healthcare in other Canadian Provinces and Territories to assess rare disease service models in those jurisdictions. Currently, a comprehensive rare disease strategy does not exist in any other Canadian Province. Most provinces provide consultation, screening, assessment, diagnosis, treatment and counselling for individuals with or at risk of rare disorders (genetic, metabolic and other), similar to the programs that currently exist in Ontario. All provinces have newborn screening programs in place but the disorders being screened are not the same. Many provinces offer disease-specific programs for the diagnosis and management of rare diseases that are more common in general or regionally.

On a national-level, Alberta, Quebec and Ontario are co-leading a Working Group by the Provincial and Territorial Health Ministers (established in Fall of 2014). The mandate of this Working Group is focused on improving inter-jurisdictional consistency in assessment and coverage of expensive rare disease drugs, and a pricing strategy that is fair to both payers and drug manufacturers⁹.

Orphanet is a reference portal for information on rare diseases and orphan drugs, for all audiences. Its main aim is to help improve the diagnosis, care and treatment of patients with rare diseases. Orphanet-Canada is dedicated to providing data and a platform for knowledge translation tools to the rare disease community. It collects information regarding specialized clinics, medical laboratories, ongoing research and patient organizations. This data collection is managed by information scientists and different sources and inclusion criteria are used to ensure the quality of the information in the database. Orphanet is governed by various committees, which independently supervise the project in order to ensure its coherence, evolution and viability. Orphanet is supported by a consortium of nearly 40 countries and coordinated by the French National Institute of Health and Medical Research (French INSERM) team.

Rare disease patient organizations have also undertaken planning efforts. CORD released its rare disease report in 2015. CORD is an umbrella organization representing patients with rare diseases and patient organizations. CORD collaborates with governments, researchers, clinicians and industry to promote research, diagnosis, treatment and services for all rare disorders in Canada.

**2.2 Rare Diseases Working Group**

**Context**

In May 2015, CORD released *Now is the Time – a national rare disease strategy for Canada*. The CORD Report, along with other recent work on specific rare disease pathways to improve access to services for patients living with rare diseases, precipitated MOHLTC interest in establishing a co-ordinated care
delivery approach to rare diseases in Ontario. To this end, in 2016, the Minister of Health and Long-Term Care announced the establishment of a Working Group to develop a strategic framework for the Province.

The strategy in the CORD Report outlines five strategic 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

All discussions, stakeholder engagement, and the resulting recommendations by the Working Group, included in this report are organized in alignment with strategic goals outlined by CORD, adapted to the Ontario context.

CORD Strategy – Mapping to Ontario Framework
Membership and Mandate

Critical Care Services Ontario (CCSO), at the request of the MOHLTC, established the Rare Disease Working Group. Co-Chaired by Dr. Ronald Cohn, Paediatrician-in-Chief, The Hospital for Sick Children and Scott McIntaggart, Senior Clinical Vice-President, University Health Network, the 12-member Working Group included 25% patient, caregiver and patient advocate representation in addition to clinical and administrative representation from the Ontario health care system. A list of all Working Group Members has been included in Appendix A. The clinician and provider expertise was drawn from the areas of diagnosis, management and treatment of rare diseases, including (but not limited to) physician geneticists, primary care providers, paediatric and adult specialists in rare diseases.

The Working Group was tasked with development of a strategic framework for rare diseases for the province, by adopting, enhancing and incorporating key components of the rare disease report created by CORD. The Working Group was tasked with submitting a Report in early 2017, outlining the proposed strategic framework, incorporating the identification of current initiatives and assets in the 5 key areas for focus in Ontario:

1) Diagnostic capacity and early detection;
2) Timely access to equitable and evidence-informed care, including supports to primary care providers in identifying and managing rare diseases;
3) Access to Complex Care and rehabilitation;
4) Access to clinical trials and potential promising therapies; and
5) Innovative research.

Process

Over the course of several months, the Working Group held a series of meetings, and work by members was undertaken between meetings. Additional stakeholder engagement and qualitative input was achieved through 16 one-on-one interviews, and participation at a patient/caregiver support group meeting. The stakeholders included patients and caregivers, clinicians, researchers and health care administrators. All stakeholders were assured confidentiality regarding their contributions. The input gathered was refined and themed, and presented to the Working Group for input. A series of recommendations were generated based on a review of the existing assets and gaps in the care-provider continuum. A high-level Asset Map (presented below) was developed to identify how the key areas of the proposed rare disease strategy may relate to programs already in existence, for consideration in the implementation of the recommendations.
Section 3.0 – Ontario’s Five Key Areas of Focus

The following section of this report is organized by the five key areas of focus and provides a review of the existing assets, the main gaps identified by the engaged stakeholders and Working Group members, and offers system-level recommendations from the Rare Disease Working Group.

For context, it is important to define an asset. Assets are programs and initiatives that are provincial in scope, and that have a current mandate to provide or advance the provision of care for the rare disease population. These programs may meet the needs of the rare disease population exclusively, or may serve this population as a subset of care provision to a broader population (e.g. complex care). The assets lists are not exhaustive, and it is recognized that regions and single health care providers are also serving the rare disease population. The purpose of listing the assets, however, is to ensure that any future planning that is to occur takes into account existing programs and initiatives.

3.1 Diagnostic Capacity and Early Detection

Assets

There exist a number of resources in the province that assist with the diagnostic capacity and early detection for rare diseases. A brief description of each is provided below.

i. Genetic Testing Advisory Committee (GTAC): Provides a formal evaluation process to assess the validity and effectiveness of new and existing genetic tests that may assist with diagnosis and detection of rare diseases. Genetic tests under consideration undergo a well-defined and in-depth evaluation process followed by a final report and recommendations that are presented to MOHLTC. Genetics Advisory Committee will operate until January 30, 2017, at which point it will be replaced by ii) and iii).

ii. Consultation and Advisory Group for Genetics in Ontario (CAGO): This group is tasked with providing policy advice directly to the labs and genetics branch at the MOHLTC.

iii. Ontario Genetics Advisory Committee (OAGC): This is a standing subcommittee of Ontario Health Technology Advisory Committee (OHTAC) at Health Quality Ontario (HQO). Its role is to provide advice to HQO in providing HTA and evidentiary support for decisions around genetic testing in Ontario.

iv. Recommendations for the use of Genome-Wide Sequencing for Undiagnosed Rare Disease in Ontario: A Working Group was convened to develop standardized criteria for the use of Genome-Wide Sequencing (GWS) in Ontario by the Genetic Testing Advisory Committee (GTAC). GWS is a genetic test used to identify undiagnosed rare disease conditions by annotating known disease causative DNA variants on a genome-wide scale; other variants, not relevant to patient’s primary indication, may also be identified. The recommendations provided by the Working Group are under consideration by the Ministry.

v. Maternal Child Screening Committee: Screening in the maternal-child population, is an important practice aimed at early identification of significant health risks to allow timely intervention and improved outcomes. This committee provides recommendations for maternal-child screening
practices, assists with policy development and stakeholder consultation with regards to screening programs and prioritization of recommended tests

vi. **Newborn Screening Ontario (NSO):** Located at the Children’s Hospital of Eastern Ontario (CHEO), this provincial program leads and administers newborn screening for the province. NSO tests for 29 diseases, including Metabolic diseases, Endocrine diseases, Sickle Cell Disease, Cystic Fibrosis and Severe Combined Immune Deficiency. NSO coordinates an extensive provincial network to deliver this care, ensuring that every newborn receives stat screening and care. NSO’s mandate includes research, development, education and innovation. Screening for a 30th target, Critical Congenital Heart Disease, was launched in February 2017 using pulse oximetry testing at the bedside. The impact of screening for current listed conditions is reviewed and evaluated by the NSO advisory council which includes observers from the MOHLTC and Ministry of Children and Youth Services (MCYS). Analyses have been performed, or are in progress, to study the health care utilization and other system outcomes by linking NSO data at Institute for Clinical Evaluative Sciences (ICES).

vii. **Better Outcomes Registry and Network (BORN):** This is the provincial pregnancy, birth and childhood registry aimed to collect, interpret, share and protect data about pregnancy, birth and childhood in the province. This asset supports screening systems by providing a critical information and knowledge translation system. As a prescribed registry under Personal Health Information Protection Act (PHIPA), it has the authority to collect, use and disclose information without consent for the purposes of quality improvement and facilitation of care. For example, the data from this registry is also accessed by NSO and helps in identifying missed screens for newborns that may be at risk. Other examples of how BORN supports quality improvement include prenatal screening (including non-invasive prenatal testing) quality assurance, perinatal care dashboards for the province, and national quality support for assisted reproduction clinics (Canadian Assisted Reproductive Technologies Register Plus). BORN is located at CHEO, and has created direct links to many hospital and primary care electronic health record systems.

viii. **Regional Genetic Centres:** Regional genetic programs exist at centres across Ontario and provide diagnostic and early detection services such as genetic testing to confirm or rule out a suspected condition or assess the probability of developing or passing a genetic disorder.

ix. **Care for Rare:** This is a nation-wide research program aimed at the development of evidence-based approaches to identify and treat rare diseases. Presently the program does not provide clinical diagnostic services, however the approaches being developed will enable translation of these approaches in future.

x. **Matchmaker Exchange:** This is an international collaborative initiative which provides a robust and a systematic approach to rare disease gene discovery by connecting multiple databases using a common application programming interface. The exchange currently connects 7 international databases and includes all of the Care for Rare research data connected via the Phenome Central database which resides at The Hospital for Sick Children.
Main Gaps Identified

- While Ontario does well compared to peers with screening programs significant system fragmentation exists for those seeking a diagnosis for rare diseases external to the existing screening programs.
- In some circumstances, it can take years to obtain a diagnosis, including multiple visits to specialists, emergency departments, and diagnostic tests.
- There is no systematic protocol for undiagnosed patient follow-up. Other jurisdictions run Undiagnosed Programs (such as the National Institute of Health in the USA) for patients with conditions that have not yet been discovered.
- Novel genome sequencing technology (Whole-Exome Sequencing, Whole Genome Sequencing) is mainly used for research purposes and rarely for clinical diagnostics in Ontario, despite decreasing the diagnostic odyssey, with the exception of the small subset that are approved for testing under the Exceptional Access Program and are sent to clinical laboratories in the USA for analysis. In addition, majority of the genetic testing panels (containing many genes) are sent to the USA, which adds to the healthcare costs. Out of Country (OOC) services program in Ontario provides access to standard of care diagnostics if the service does not exist in Ontario or if the volume of testing means that it would not be cost effective to do testing in the province. This becomes even more vital in the currently existing system.
- The diagnosis and management of rare disease conditions, genetic and non-genetic (such as auto-immune diseases), is often completed by specialists and sub-specialists outside of Regional Genetic Centres (e.g. Rheumatologist, Neurologist, Epileptologist, Cardiologist, Physiatrist, etc.). There is little organization and knowledge related to these resources and individual degrees of expertise.

Working Group Recommendations

1. Sponsor the development of an Ontario-based Rare Disease registry for the following:
   a. Tracking patient outcomes;
   b. Establishing linkages to research endeavors and clinical trials;
   c. Connecting patients and families to disease specific patient support groups;
   d. Evaluating the program.

   Such a system should leverage existing systems in Ontario such as BORN, ICES, NSO, etc. This registry may connect to a Pan-Canadian registry, and international databases and registries through portals such as Phenome Central, Canadian Inherited Metabolic Diseases Research Network (CIMDRN) and Matchmaker Exchange.

2. Develop well-defined criteria for novel genome sequencing technology (Whole-Exome Sequencing, Whole Genome Sequencing) to ensure timely access for testing and diagnosis of rare diseases. For non-geneticists ordering testing, a mechanism to access education and genetic counselling to support the diagnostic process (pre and post-test evaluation and diagnosis) at the Regional Genetic Centres should be established to ensure that patients are
receiving appropriate information. Where applicable, genetic counselling should include discussions about familial testing for rare diseases.

3. Identify, prepare and maintain an inventory of existing specialized clinics/services, evolving research programs and rare disease support groups and other initiatives across the province. Publish the information on government websites and other websites such as Orphanet Canada, to assist patients, caregivers and, primary care providers to support connections within and outside the health care system.

3.2 Timely Access to Equitable and Evidence-based care, including supports to primary care providers in identifying and managing rare diseases

Assets
i. **Newborn Screening Ontario (NSO):** This program has been described in Section 3.1. The initiative is also focused on timely access to treatment and management of the 29 identified rare disease conditions to improve the outcome and save lives. To serve this aim, all samples received are treated as urgent, the data entry team strives to record all information on the same day the sample is received and positive screens are communicated immediately. NSO coordinates a set of six “Disease Specific Working Groups” to share knowledge and coordinate practice between the specialists at the five Regional Treatment Centres (located at the Pediatric Academic Health Sciences Centres at Hamilton, Kingston, London, Ottawa and Toronto).

ii. **Complex Care for Kids Ontario (CCKO):** This strategy is implemented by Provincial Council of Child and Maternal Health (PCMCH) to improve care for children with medical complexity and their families. The strategy aims at making care more effective and efficient, along with improving service delivery and quality of life for the patient-families.

iii. **Regional Genetic Centres:** Regional genetic programs exist at centres across Ontario and provide timely access by providing genetic counselling and assessment for the inherited disorders, advising the consequences and probability of developing or transmitting the conditions to patients or family. These centres also provide access to physician geneticists to discuss potential treatment options or formulate a comprehensive care plan for management of the condition. Of note, the Northern Regional Genetics Program (NRGP) is a network that includes five genetic counselling programs which provide services to families located in Northeastern and Northwestern Ontario. As part of the NRGP, Health Science North's Genetics Laboratory offers specialized services to these families.

iv. **Rare Disease Specialty Clinics:** Several specialty clinics for specific rare disease conditions serving children and/or adults exist or are under development in the province. There are variable funding strategies to support these clinics. Some of the clinics also propose centralizing care coordination to support primary care providers in sub-urban or rural areas through e-consults. A few examples of these clinics are: Phenylketonuria Clinic, Hemoglobinopathy Clinic, Neurofibromatosis Clinic, 22q Deletion Syndrome Clinic, Sickle Cell Clinic, Rett Syndrome Clinical Network, new Ehlers-Danlos Syndrome (EDS) Clinic, etc.
v. **Genetics Medical Education Program**: The genetics curricula for medical education programs at the undergraduate and postgraduate levels across universities in the province have been updated to focus on identification and management of rare diseases and interpretation of genetic testing. Further there exist continuing medical education programs designed to educate primary care providers for diagnosing rare diseases e.g. Family Medicine Genetics Program at Mount Sinai Hospital.

vi. **Genetics Education Canada Knowledge Organization (GEC KO)**: This program aims to build genetics competencies in primary care, specialty medicine, and other health professions by increasing genetics literacy and supporting translation of genetics research. The program also enables development, collection, dissemination and evaluation of genetics educational materials.

vii. **Professional Networks, Societies**: The professional network societies such as Canadian College of Medical Geneticists (CCMG) and the Ontario Medical Association - Genetics Section, Canadian Association of Genetic Counsellors (CACG), play an important role in training, credentialing and providing continuing education to professionals who work in the field or those who are interested in specializing in it. Besides knowledge dissemination and updating members with the latest advancements, these organizations also establish standards for education and training programs offered.

**Main Gaps Identified**
- Family physicians and specialists may not be well-equipped to address the needs of rare disease patients, even after diagnosis.
- Lack of recognition and absence of coordinated care (individualized medical protocols) for patients at an increased risk of medical complications across various organ systems spanning multiple medical specialties.
- Patients and families report that centres with genetic clinical and research programs such as CHEO, SickKids (and other Pediatric Academic Health Sciences Centres at London, Hamilton, Kingston) or specialized clinics like University Health Network (UHN) and those offered in the community, provide comprehensive care. However, these can be difficult to access outside urban areas and waits can be long.
- In general, access to clinical and support services becomes disparate at the time of transition to adult care, irrespective of location\(^\text{11}\).
- Access to genetic counselling for patients and families is uneven, with less access to counsellors reported outside major academic hospitals.
- Specialized disease clinics are generally not connected to patient organizations, or other community-based support services and programs, including rehabilitation, specialized education, home modifications and psychosocial support services.

**Working Group Recommendations**
4. Include rare disease education within individual curricula (with a focus on interpretation of genetic testing and management of rare disease) in undergraduate and post-graduate medical education and physician training programs.
5. Establish criteria for “Centers of Reference” in rare diseases to facilitate improved coordination of care and access to specialized knowledge for the management of rare diseases. “Centres of Reference” may provide rare disease diagnosis, treatment and management leadership and work collaboratively with Regional Genetic Centres. Rare disease system leadership activities may include current rare disease knowledge translation in diagnosis and management and the provision of consultative advice for patient care. Leverage existing resources within hospitals currently offering rare disease focussed programs such as SickKids, CHEO and UHN.

6. Develop a “hub and spoke” style model between Regional Genetic Service Centres, primary care, and appropriate specialist providers, and education and capacity building programs such as Project ECHO, to improve the provision of care in sub-urban and rural areas. Regional Genetic Centres may further provide support as a connection to genetic counselling services, consultative advice between providers and co-management of patients, where appropriate. Consider leveraging telehealth systems to facilitate remote consultations.

7. Develop and disseminate accredited clinical and patient education tools such as Clinical Practice Guidelines and patient self-management tools. Ensure education tools support cross-provider knowledge transfer and patient management throughout the lifespan. Extend education beyond clinical setting to the community, connecting with patient groups and schools.

3.3 Access to Complex Care and Rehabilitation

Assets

i. Rare Disease Specialty Clinics: Several specialty clinics for specific rare disease conditions serving children and/or adults exist or are under development in the province. Some of the clinics also propose centralizing care coordination to support primary care providers in sub-urban or rural areas through e-consults. A few examples of these clinics are: Phenylketonuria Clinic, Hemoglobinopathy Clinic, Neurofibromatosis Clinic, 22q Deletion Syndrome Clinic, Sickle Cell Clinic, Rett Syndrome Clinical Network, Pediatric Neuromuscular Clinics, new EDS Clinic, etc.

ii. Complex Care for Kids Ontario (CCKO): This program has been described in Section 3.2. It is aimed at child and youth population that demonstrate a need for complex healthcare and are medically fragile and/or technology dependent. Rare disease patients who persistently demonstrate most complex and challenging healthcare needs may participate in this program.

iii. Ontario Special Needs Strategy: This strategy is led by MCYS. It aims at supporting children and youth with special needs to receive timely and effective services they need at home, at school, in the community and as they transition to adulthood. The initiative works with service providers and educators across Ontario to develop these services. The three key areas being

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a Project ECHO (Extension for Community Healthcare Outcomes) is a collaborative model of medical education and care management that empowers clinicians everywhere to provide better care to more people, right where they live. The heart of the ECHO model™ is its hub-and-spoke knowledge-sharing networks, led by expert teams who use multi-point videoconferencing to conduct virtual clinics with community providers. In this way, primary care doctors, nurses, and other clinicians learn to provide excellent specialty care to patients in their own communities.
worked upon are: identifying kids’ needs earlier and connecting them to the right help sooner, coordinating service planning, making the delivery of rehabilitation services seamless.

The assets discussed below are not specifically designed to serve the needs of the rare disease population but may be of help to rare disease patients with comorbidities such as developmental delay, physical and, mental disability.

iv. Community Care Access Centres (CCAC): This program provides care at home, school or in community and helps connect to other services that exist in the community. The program serves varied segments of the population such as seniors, adults, children and their families who need support and care, and is not specifically designed for the rare disease population.

v. Developmental Services Ontario (DSO): The DSO agencies help adults with developmental disabilities find services and supports in their community.

vi. Ontario Disability Support program (ODSP): The program offers income and employment supports such as financial assistance with living expenses, benefits including prescription drugs and vision care, and assistance with finding and keeping a job to individuals with disabilities.

Main Gaps Identified

- Several government-funded supports exist providing complex care and rehabilitation, particularly for children and youth. In the community, these resources are difficult to access as there are limits and waits for various supports from DSO, CCAC, ODSP, CCKO etc. Families often self-fund care.
- In the adult system, there are less resources and less co-ordination available, leaving the load of navigation on the patient and/or families. This can be particularly difficult for families that transition from paediatric to adult stage wherein certain support services may no longer be available.
- There exists a lack of coordination of care for paediatric patients in area of complex care and rehabilitation.
- Patients and their caregivers expressed concern about caregiver burden when caring for patients with complex needs. Of chief concern was sustainability of care and potential burnout.

Working Group Recommendations

8. Explore capacity to leverage Community Health Centres (CHC) and Family Health Teams (FHT) as networks for care and management for patients with complex presentations. Engage CHC and FHT networks as ‘Spokes’ of the ‘Hub and Spoke Model’ that connect to “Centres of Reference” for providing specialized care for rare disease patients. Clearly outline the roles of each level of the ‘Hub and Spoke Model’ to ensure the right expertise and access is in place. Identify and enlist primary care provider champions (especially in sub-urban and rural areas) with specialized interest and/or knowledge of rare diseases to manage patients with complex needs through programs such as Project ECHO. Incorporate patient and family engagement with rare disease
patient organizations and support groups, to provide psychosocial support, establish connections and joint programs of awareness, education, and support.

9. Review policies and program criteria for complex care service delivery for organizations offering programs for patients with complex needs. Focus patient inclusion/exclusion criteria on patient’s needs, versus diagnosis. Ensure education of the health discipline providers regarding rare diseases to facilitate patient care (e.g. physiotherapist, occupational therapist), to prevent a diagnosis from becoming a barrier in care.

10. Allocate personnel with specialized rare disease knowledge as patient navigators (e.g. genetic counsellor, social worker) to assist patients with complex needs, and assist caregivers to obtain adequate and appropriate supports. This has been successful locally in other models such as adolescent transition clinics, cancer care, geriatric emergency management, and at a system-level for chronic disease management (i.e., Health Links).

11. Design programs for patients with complex needs with a Family-centered care approach. Specifically ensure that patients graduating from the paediatric care setting to the adult setting, have appropriate clinical and social support transitions to ensure continuity of clinical and support service delivery through the patient’s lifespan.

3.4 Access to Clinical Trials and Potential Promising Therapies

Assets

i. **Centre for Genetic Medicine, The Hospital for Sick Children**: The program focusses on discovery, translating knowledge into clinical applications, and enhancing genetic/genomic education for health-care professionals, patients and families. Key areas that pertain to clinical trials, research and promising therapies include, examining new therapies and gene-based diagnostics for their cost effectiveness and the ability to improve clinical outcomes. Along with, a clinic research program utilizing Whole Genome Sequencing (WGS) to identify causes for undiagnosed genetic conditions, predict disease progression, and guide treatment choices. This program serves the needs of the paediatric population.

ii. **The Centre for Applied Genomics**: This program provides research project support to hundreds of investigators who are involved in genomics research at local, national and international levels. An example of services provided include DNA sequencing/synthesis, microarray analysis, cytogenomics and genome resources, genetic analysis, statistical analysis, biobanking, and informatics.

iii. **Care for Rare**: As described in Section 3.1, this research initiative is focused on discovering new rare disease conditions, and developing novel and effective therapeutic approaches to treat these conditions. This program is also collaborating with multiple pharmaceutical companies for treatment discovery and recovery, and clinical translation.

iv. **Canadian Inherited Metabolic Diseases Research Network (CIMDRN)**: This program is an interdisciplinary practice-based research network designed to develop an evidence-informed approach to support health care providers and decision makers in providing the best care for paediatric patients with Inborn Errors of Metabolism (IEM). This program provides an observational research platform with which to understand patient centered, clinical, and system outcomes.
Main Gaps Identified

- Systemic barriers exist preventing research for rare diseases in Ontario, including:
  - limited availability of clinical research units and inpatient beds to facilitate clinical research projects for rare diseases; and
  - clinical research regulatory frameworks limit smaller study populations
- Clinical studies support rare disease work in the paediatric population (e.g. Inborn Errors of Metabolism). Given the ageing rare disease population, more clinical studies are required for the adult population.
- There is a lack of continuity for funding between paediatric and adult populations, and also, congruency with other rare disease programs (e.g. for diseases identified through provincial newborn screening program, continuum of support should exist for therapies).

Working Group Recommendations

12. Support the development of multi-centre REB protocols (and other such mechanisms) to facilitate exchange of clinical, scientific and regulatory information by researchers regarding clinical research in this field.

13. Continue the work of the Provincial-Territorial Working Group on Rare Disease Drugs to enable access to these therapies with evidence based approaches. Maximize opportunities for continuity between care settings, development of access pathways that provide managed access to promising therapies while addressing safety, cost-effectiveness and continuity of care with other MOHLTC funded programs (e.g., funding for therapies to treat diseases screened by NSO).

14. Explore funding support for acute care hospitals to access expensive rare disease drugs and its coordination for in-patients to allow care for patients to be undertaken at a regional level.

15. Support the development of formalized pathways to facilitate patient connection with clinical trial and research endeavors.

3.5 Promoting Innovative Research

Rare disease diagnosis and management is dependent on research both due to the continuous evolution of diagnostic and treatment modalities and disease discovery.

Assets

i. Newborn Screening Ontario (NSO): This program has been described earlier in Section 3.1. The initiative also has a mandate to perform and support innovative research and provide the available data for secondary research use. The residual dried blood samples from newborn testing for 29 rare diseases are used for research protocols that are defined by NSO policies and approved by Research Ethics Board.

ii. Better Outcomes Registry and Network (BORN): This asset has been described earlier in Section 3.1. The program facilitates research on maternal and child health issues and works with researchers requesting BORN data to ensure information privacy is regulated by Personal Health Information Protection Act, 2004 (PHIPA) including review and approval by Research Ethics Board.
iii. **Centre for Genetic Medicine, The Hospital for Sick Children**: This program has been described earlier in Section 3.4. The centre aims to boost research innovation, identify opportunities to enhance translation of research, disseminate knowledge to providers and families, and build systems for collaboration and partnership in the field of genetic medicine.

iv. **The Centre for Applied Genomics**: As described in Section 3.4, this program provides research support for genomics and genetics projects by providing services such as data generation, storage, statistical and genetic analysis and associated bioinformatics activities. The centre has a variety of computing hardware, and high-performance computing systems along with a multi-disciplinary team of scientists and staff to support the services provided.

v. **Care for Rare**: This is a nation-wide research program aimed at the development of evidence-based approaches to identify and treat rare diseases. Presently the program does not provide clinical diagnostic services, however, the approaches being developed will enable translation of these approaches future.

vi. **Academic - Industry collaborations (e.g. clinical trials, drug development)**: This asset refers to the existing relationships between healthcare providers involved with academic research and industry partners aimed at furthering research and innovation for rare diseases. These inter-disciplinary collaborations may receive financial support and/or oversight from government and initiatives involved include development of effective treatments, diagnostic services, screening assays for rare diseases.

vii. **Ontario Genomics**: This program supports and sustains genomics innovation and research in Ontario by offering competitive funding and commercialization opportunities for research projects and start-ups. Expected outcomes of this initiative include development of novel techniques and therapeutic approaches that may also benefit the rare disease patient population. Ontario Genomics is funded by the Ontario government and the federal research funding agency Genome Canada.

**Main Gaps Identified**

- There are ongoing research programs for the paediatric rare disease population (e.g. Centre for Genetic Medicine, Sick Kids, CHEO). There are limited research initiatives for the adult rare disease population.
- There are only a few programs (Ontario Genomics, Canadian Institutes of Health Research) that support development of technology for genetic research and its application to human health.

**Working Group Recommendations**

16. Explore the potential for dedicated research co-ordination to conduct multi-site research, including research with a focus on rare disease throughout the lifespan into adulthood.

17. As part of a broader plan for education and knowledge translation, through networks and formal educational programs such as Project ECHO, share research findings which benefit rare disease patient population among Ontario’s medical community.

18. Facilitate government and inter-industry research collaborations to accelerate development of novel therapeutic approaches for rare diseases, and to enhance efficiency and cost effectiveness.
19. Partner with research projects for the evaluation of delivery of care for rare disease patients such as effectiveness of care delivery, care coordination, clinical management and improved patient outcomes.
Section 4.0 – Summary Recommendations

Presented below is a summary of the recommendations, with a timeframe assignment and the existing assets that should be considered when planning implementation of the recommendation. As with all provincial healthcare strategies, implementation of the recommendations will require MOHLTC support and clearly defined accountabilities. The following recommendations cross system and organizational lines and will require collaboration between multiple branches of the MOHLTC, and organizations across health sectors. Without cooperation across multiple branches of the MOHLTC and inter-sector collaboration, fragmentation across services and programs will continue to persist.

The recommendations vary in the amount of effort required. Some recommendations have interdependencies, particularly those that are related to establishing Centres of Reference and a subsequent network of program and provider connections throughout Ontario. The interdependencies have been identified and sequenced in the diagram provided in Appendix B. It was not the mandate of the Working Group to prioritize the recommendations or develop an implementation plan.

<table>
<thead>
<tr>
<th>No.</th>
<th>Recommendation</th>
<th>Short/Medium/Long Term</th>
<th>Leverage Existing Assets:</th>
</tr>
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<tbody>
<tr>
<td>1.</td>
<td>Sponsor the development of an Ontario-based Rare Disease registry for the following:</td>
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<tr>
<td></td>
<td>1. Tracking patient outcomes;</td>
<td>Long-term</td>
<td>• Better Outcomes Registry and Network</td>
</tr>
<tr>
<td></td>
<td>2. Establishing linkages to research endeavors and clinical trials;</td>
<td></td>
<td>• Newborn Screening Ontario</td>
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<td></td>
<td>3. Connecting patients and families to disease specific patient support groups;</td>
<td></td>
<td>• Care for Rare</td>
</tr>
<tr>
<td></td>
<td>4. Evaluating the program.</td>
<td></td>
<td>• MatchMaker Exchange</td>
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<td></td>
<td></td>
<td></td>
<td>• Orphanet Canada</td>
</tr>
<tr>
<td>2.</td>
<td>Develop well-defined criteria to enable access to novel genome sequencing technology (Whole-Exome Sequencing, Whole Genome Sequencing) to ensure timely access for testing and diagnosis of rare diseases.</td>
<td>Short-term</td>
<td>• Consultation and Advisory Group for Genetics in Ontario (CAGO)</td>
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<td></td>
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<td></td>
<td>• Regional Genetic Centres, including specialized centres such as The Centre for Applied Genomics (SickKids)</td>
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<td>No.</td>
<td>Recommendation</td>
<td>Short/Medium/Long Term</td>
<td>Leverage Existing Assets:</td>
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</table>
| 3.  | Identify, prepare and maintain an inventory of existing specialized clinics/services, evolving research programs and rare disease support groups and other initiatives across the province. Publish the information on government websites and other websites such as Orphanet Canada to assist patients and caregivers, along with primary care providers to support connections within and outside the health care system. | Short-term             | • Ontario Human Hereditary and Genetic Medicine Programs Inventory (included in Report as Appendix C)  
• Orphanet Canada |
| 4.  | Include rare disease education within individual curricula (with a focus on interpretation of genetic testing and management of rare disease) in undergraduate and post-graduate medical education and physician training programs. | Medium-term            | • An example is University of Toronto. Other medical education programs to be surveyed.  
• Genetics Medical Education Program (Continuing Medical Education to primary care providers and specialists for diagnosing rare diseases in their specialties) |
| 5.  | Establish criteria for “Centers of Reference” in rare diseases to facilitate improved coordination of care and access to specialized knowledge for the management of rare diseases. | Medium-term            | • Reference: European Reference Network criteria, NIH Undiagnosed Program and other jurisdictional models  
• Many/Most of the treatable rare genetic diseases are Inborn Errors of Metabolism. The existing four major pediatric metabolic diseases clinics in Ontario should be leveraged.  
• There is one adult metabolic clinic in Toronto. The London Health Sciences Centre (LHSC) and Hamilton Health Sciences Centre (HHSC) model is conducive to adult care in the same institution as the pediatric care. Ottawa lacks an adult clinic and this is a growing need. |
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<thead>
<tr>
<th>No.</th>
<th>Recommendation</th>
<th>Short/Medium/Long Term</th>
<th>Leverage Existing Assets:</th>
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</thead>
</table>
| 6.  | Develop a "hub and spoke" style between Regional Genetic Service Centres, primary care, and appropriate specialist providers, and education and capacity building programs such as Project ECHO, to improve the provision of care in suburban and rural areas. | Medium-term | • Genetics Education Canada – Knowledge Organization  
• Ontario Project ECHO |
| 7.  | Develop and disseminate accredited clinical and patient education tools such as Clinical Practice Guidelines and patient self-management tools. Ensure education tools support cross-provider knowledge transfer and patient management throughout the lifespan. | Long-term, ongoing | • Canadian College of Medical Geneticists  
• Canadian Association of Genetic Counsellors  
• Genetics Education Canada – Knowledge Organization  
• Regional Genetic Centres and specialized clinics  
• Rare Disease Patient Organizations |
| 8.  | Engage CHC and FHT networks as ‘Spokes’ of the ‘Hub and Spoke Model’ to build capacity in primary care for the co-management of rare disease patients. | Medium-term | • Genetics Education Canada – Knowledge Organization  
• Ontario Project ECHO |
| 9.  | Review policies and program criteria for complex care service delivery for organizations offering programs for patients with complex needs. Focus patient inclusion/exclusion criteria on patient’s needs, versus diagnosis. Ensure education of the health discipline providers regarding rare diseases to facilitate patient care (physiotherapist, occupational therapist), to prevent a diagnosis from becoming a barrier in care. | Medium-term | • Developmental Services Ontario  
• Community Care Access Centres  
• Ontario Disability Support Program |
| 10. | Allocate personnel as patient navigators (e.g. genetic counsellor, social worker) to assist patients with complex needs and assist caregivers to obtain adequate and appropriate supports. | Short-term (pilot initiative)/Medium-term | • Complex Care for Kids Ontario (CCKO)  
• Professional Networks e.g. Canadian Association of Genetic Counsellors (CAGC) |
<table>
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<tr>
<th>No.</th>
<th>Recommendation</th>
<th>Short/Medium/Long Term</th>
<th>Leverage Existing Assets:</th>
</tr>
</thead>
<tbody>
<tr>
<td>11.</td>
<td>For children with complex needs graduating from the paediatric care setting to the adult setting, ensure appropriate clinical transitions are in place to ensure continuity of care.</td>
<td>Medium-term</td>
<td>• Example: SickKids Good 2 Go Transition program</td>
</tr>
<tr>
<td>12.</td>
<td>Support the development of multi-centre REB protocols (and other such mechanisms) to facilitate exchange of clinical, scientific and regulatory information by researchers regarding clinical research in the field.</td>
<td>Medium-term</td>
<td></td>
</tr>
<tr>
<td>13.</td>
<td>Continue the work of the Provincial-Territorial Working Group on Rare Disease Drugs to enable access to these therapies with evidence based approaches. Maximize opportunities for continuity between care settings, development of access pathways that provide managed access to promising therapies while addressing safety, cost-effectiveness and continuity of care with other MOHLTC funded programs (e.g., funding for therapies to treat diseases screened by NSO).</td>
<td>Medium-term</td>
<td>• Ontario Public Drugs Programs and the national drug review process through the Canadian Agency for Drugs and Technologies in Health (CADTH).</td>
</tr>
<tr>
<td>14.</td>
<td>Explore funding support for acute care hospitals to access expensive rare disease drugs and its coordination for in-patients to allow care for patients to be undertaken at a regional level.</td>
<td>Medium-term</td>
<td>• Hospitals Branch, Health System Quality and Funding Division, MOHLTC</td>
</tr>
<tr>
<td>15.</td>
<td>Support the development of formalized pathways to facilitate patient connection with clinical trial and research endeavors.</td>
<td>Long-term</td>
<td>• Rare Disease Patient Organizations</td>
</tr>
<tr>
<td>16.</td>
<td>Explore the potential for dedicated research co-ordination to conduct multi-site research, including research with a focus on rare disease throughout the lifespan into adulthood.</td>
<td>Long-term</td>
<td>• Ontario Genomics • Centre for Applied Genomics • Rare Disease Specialty Clinics • Care for Rare</td>
</tr>
<tr>
<td>No.</td>
<td>Recommendation</td>
<td>Short/Medium/Long Term</td>
<td>Leverage Existing Assets:</td>
</tr>
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<td>-----</td>
<td>--------------------------------------------------------------------------------</td>
<td>------------------------</td>
<td>------------------------------------------------------------------------------------------</td>
</tr>
</tbody>
</table>
| 17. | Develop knowledge translation strategies to share beneficial research findings among Ontario’s medical community. | Medium-term            | • Professional Networks, Societies (CCMG, OMA-Genetics, CAGC)  
• Genetics Education Canada - Knowledge Organization  
• Genetics Medical Education Program (e.g. University of Toronto)  
• Continued Medical Education for physicians through OMA |
| 18. | Facilitate government and inter-industry research collaborations to accelerate development of novel therapeutic approaches for rare diseases, and to enhance efficiency and cost effectiveness. | Long-term              | • Care for Rare  
• Ontario Genomics  
• Canadian Institute of Health Research |
| 19. | Partner with research projects for evaluating effectiveness of care delivery, care coordination, clinical management and improved patient outcomes. | Medium-term            | • Centre for Genetic Medicine, The Hospital for Sick Children  
• Canadian Inherited Metabolic Diseases Research Network  
• Academic - Industry collaborations (e.g. clinical trials, drug development) |
Appendix A – Rare Diseases Working Group Members

Co-Chairs:

- Dr. Ronald Cohn, Paediatrician-in-Chief, The Hospital for Sick Children
- Scott McIntaggart, Senior Clinical Vice President, University Health Network

Members:

- Dr. Allan Grill, Lead Physician, Markham Family Health Team
- Dr. Kym Boycott, Clinical Geneticist, Children’s Hospital of Eastern Ontario
- Dr. Pranesh Chakraborty, Director, Newborn Screening Ontario; Physician, Section of Metabolism and Newborn Screening, Children’s Hospital of Eastern Ontario
- Eriskay Liston, Genetic Counsellor, Clinical and Metabolic Genetics, The Hospital for Sick Children
- Dr. Raymond Kim, Medical Geneticist, University Health Network
- Dr. Hanna Faghfoury, Clinical and Metabolic Geneticist, University Health Network
- Dr. Richard Ward, Physician Lead, Blood Disorders Program, University Health Network
- Dr. Bernard Lawless, Provincial Lead, Critical Care
- Linda Kostrzewa, Senior Director, Strategy and System Transformation
- Durhane Wong-Rieger, President, Canadian Organization for Rare Diseases
- Guida Clozza, Patient Caregiver
- Crystal Chin, Patient

Administrative Leadership & Support:

- Anastasia Vogt, Project Manager
- Benu Sethi, Senior Business Analyst
Appendix B – Sequence of Recommendations

Prefix ‘R’ refers to the recommendations as listed in the ‘Rare Disease Draft Recommendation Summary’
Appendix C – Ontario Human Hereditary and Genetic Medicine Programs Inventory

Table 1: Human Hereditary and Genetic Medicine Programs in Ontario that offer general, metabolic and prenatal services for adult and paediatric populations

<table>
<thead>
<tr>
<th>Sr. No.</th>
<th>Centre</th>
<th>Services Offered</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>General Clinical</td>
</tr>
<tr>
<td>1.</td>
<td>Hamilton Health Sciences; McMaster Children’s Hospital</td>
<td>Y</td>
</tr>
<tr>
<td>2.</td>
<td>Kingston General Hospital (KGH)</td>
<td>Y</td>
</tr>
<tr>
<td>3.</td>
<td>London Health Sciences Centre; Children’s Hospital of Western Ontario</td>
<td>Y</td>
</tr>
<tr>
<td>4.</td>
<td>Windsor-Essex County Health Unit</td>
<td>Y</td>
</tr>
<tr>
<td>5.</td>
<td>Trillium Health Partners (Credit Valley Hospital, Mississauga)</td>
<td>Y</td>
</tr>
<tr>
<td>6.</td>
<td>North York General Hospital (NYGH)</td>
<td>Y</td>
</tr>
<tr>
<td>7.</td>
<td>Lakeridge Hospital</td>
<td>Y</td>
</tr>
<tr>
<td>8.</td>
<td>Children’s Hospital of Eastern Ontario (CHEO)</td>
<td>Y</td>
</tr>
<tr>
<td>9.</td>
<td>Peterborough Regional Health Centre</td>
<td>Y</td>
</tr>
<tr>
<td>10.</td>
<td>Scarborough Hospital</td>
<td>Y</td>
</tr>
<tr>
<td>11.</td>
<td>Algoma Public Health</td>
<td>Y</td>
</tr>
<tr>
<td>12.</td>
<td>Health Sciences North</td>
<td>Y</td>
</tr>
<tr>
<td>13.</td>
<td>Thunder Bay Regional Health Sciences Centre</td>
<td>Y</td>
</tr>
<tr>
<td>14.</td>
<td>Porcupine Health Unit</td>
<td>Y</td>
</tr>
<tr>
<td>15.</td>
<td>North Bay Parry Sound District Health Unit</td>
<td>Y</td>
</tr>
<tr>
<td>16.</td>
<td>Mackenzie Health</td>
<td>Y</td>
</tr>
<tr>
<td>17.</td>
<td>University Health Network</td>
<td>Y</td>
</tr>
<tr>
<td>18.</td>
<td>The Hospital for Sick Children</td>
<td>Y</td>
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<tr>
<td></td>
<td>Hospital Name</td>
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<tr>
<td>19.</td>
<td>Mount Sinai Hospital</td>
<td>Y Y V V</td>
</tr>
<tr>
<td>20.</td>
<td>Rouge Valley Health System</td>
<td>Y Y V V</td>
</tr>
<tr>
<td>21.</td>
<td>Sunnybrook Health Sciences Centre</td>
<td>Y V V</td>
</tr>
<tr>
<td>22.</td>
<td>Orillia Soldiers’ Memorial Hospital</td>
<td>Y V V</td>
</tr>
</tbody>
</table>

* Metabolic services at Kingston General Hospital (KGH) are supported by Children’s Hospital of Eastern Ontario (CHEO), which includes a monthly clinic, on-call support and provision of basic metabolic lab tests.
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<thead>
<tr>
<th>Sr. No.</th>
<th>Centre</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>Hamilton Health Sciences</td>
</tr>
<tr>
<td>2.</td>
<td>Juravinski Cancer Centre</td>
</tr>
<tr>
<td>3.</td>
<td>Kingston General Hospital (KGH)</td>
</tr>
<tr>
<td>4.</td>
<td>London Health Sciences Centre (London Regional Cancer Program)</td>
</tr>
<tr>
<td>5.</td>
<td>Trillium Health Partners (Credit Valley Hospital, Mississauga)</td>
</tr>
<tr>
<td>6.</td>
<td>North York General Hospital (NYGH)</td>
</tr>
<tr>
<td>7.</td>
<td>Lakeridge Hospital</td>
</tr>
<tr>
<td>8.</td>
<td>Children’s Hospital of Eastern Ontario (CHEO)</td>
</tr>
<tr>
<td>9.</td>
<td>Algoma Public Health</td>
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<tr>
<td>10.</td>
<td>Thunder Bay Regional Health Sciences Centre</td>
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<td>11.</td>
<td>Porcupine Health Unit</td>
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<tr>
<td>12.</td>
<td>North Bay Parry Sound District Health Unit</td>
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<tr>
<td>13.</td>
<td>Windsor Regional Hospital Cancer Program</td>
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<tr>
<td>14.</td>
<td>Mackenzie Health</td>
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<td>15.</td>
<td>University Health Network</td>
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<tr>
<td>16.</td>
<td>Princess Margaret Cancer Centre</td>
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<td>17.</td>
<td>The Hospital for Sick Children</td>
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<tr>
<td>18.</td>
<td>Mount Sinai Hospital</td>
</tr>
<tr>
<td>19.</td>
<td>Sunnybrook Health Sciences Centre</td>
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<tr>
<td>20.</td>
<td>Orillia Soldiers’ Memorial Hospital</td>
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</table>
Appendix D – Other – Jurisdictional Activity

There is no universally accepted definition for rare diseases and they are most commonly defined based on their prevalence, incidence or in some cases by a combination of factors based on severity and the existence or feasibility of alternative therapeutic options. A review of legislation, regulations, and policies related to rare diseases indicates that 18 OECD countries (Organization for Economic Co-operation and Development) have established national strategies to address rare disease populations. The strategies are mainly aimed at access to treatment services, creating a regulatory framework for research stimulation and patient advocacy.

In the United States, National Institutes of Health (NIH) funds and conducts the majority of basic and translational research on rare diseases, and the Food & Drug Administration (FDA) works with advancing and evaluating development of products (drugs, biologics, devices, or medical foods) that demonstrate promise for diagnosis and/or treatment of rare diseases. The Human Genome Project mapped the human genome, allowing for discovery of the genetic cause of thousands of rare diseases. The Rare Diseases Clinical Research Network (RDCRN) program at NIH supports clinical studies and facilitates collaborations with scientists from multiple disciplines and patient advocacy groups to develop treatments for rare diseases. The National Organization for Rare Disorders (NORD) is a patient advocacy organization dedicated to individuals with rare diseases and organizations serving them. NORD collaborates with NIH programs on issues pertaining to rare disease research and patient care.

The UK Strategy for Rare Diseases is an overarching framework document that focuses on improving the lives of rare disease patients and their families. The strategy makes a commitment in the following areas:

1) Empowering those affected by rare diseases
2) Identifying and preventing rare diseases
3) Diagnosis and early intervention
4) Coordination of care
5) Role of research

Each of the four UK countries has developed an implementation plan outlining activities to be undertaken to improve the services and research to achieve the goals of the strategy.

The European Commission (EC) Public Health estimates that rare diseases affect 6-8% – between 26-27 million – of their population. The European Union aims to gather resources to share expertise and information across borders and EC recently announced 23 European Reference Networks (ERNs) for rare diseases. The objectives of ERNs mainly include knowledge sharing, improving diagnosis and care in medical domains where expertise is rare, improved access of highly specialized healthcare to patients, enhancing innovative research in medical science and health technologies, and focusing on medical training and research, information dissemination and evaluation. ERNs also aim to help member states, with small size of rare disease patient populations, by providing highly specialized care. EURORDIS-Rare Diseases Europe is a non-profit alliance of rare disease patient organizations from over 60 European
countries. EURORDIS contributes to national processes, and facilitates the adoption and implementation of national plans and strategies for rare diseases in these countries.

The International Rare Diseases Research Consortium (IRDiRC) was established as a collaborative initiative between NIH and EC to accelerate medical breakthroughs to find treatments and diagnoses for rare disease patients. It was unanimously agreed that maximizing scarce resources and coordinating research activities are key to achieve success in the field of rare diseases\textsuperscript{14}. To date, IRDiRC has teamed up with researchers and organizations from over 20 countries to achieve two overarching objectives by the year 2020:

1) 200 new therapies for rare diseases: The consortium will develop all the necessary measures and policies to facilitate the development of new therapies for rare diseases, such as coordinating patient registries, enhancing clinical trials and improving the regulatory framework to facilitate development of novel therapies.

2) Means to diagnose most rare diseases: This will be achieved by using different approaches to identify biomarkers of rare diseases and stimulating development of efficient, multi-purpose diagnostic tests for rare diseases, in conjunction with industry.

Western Australia (WA) Rare Diseases Strategic Framework recognizes that further evidence and relevant information is required for rare diseases in the region. The framework is structured around 4 priorities, 12 objectives and over 50 initiatives intended to respond to needs of individuals living with rare diseases, as well as to support clinicians, researchers and policy-makers. The priorities of the framework are:

1) To advance rare diseases planning
2) To promote a person-centered approach for people living with rare diseases
3) To contribute to a high-quality health system for people living with rare diseases
4) To foster world class research on rare diseases

Rare Voice Australia (RVA) is Australia's National Alliance for rare diseases and advocates for promoting health policy and a healthcare system that works for individuals with rare diseases.
Appendix E

Literature Reviewed


References


6. Ibid


12. Ibid
