IRDiRC Task Force Proposal

Name of Task Force: **Primary Care**
Name(s) of Proposer(s): Gareth Baynam, Steve Groft, and Adam Hartman

Objective(s) of the Task Force:

1. To bring together representatives from the stakeholders required to identify the priority research areas in primary care that need to be addressed to deliver against the IRDiRC goals.
2. To bring together representatives from the stakeholders required to identify current state of play.
3. Identify challenges and opportunities in rare diseases research in primary care. This may include the following area: diagnosis, therapies, ELSI, and patient engagement.
    - **Diagnosis:**
        a) Early identification and referral pathways (first steps)
        b) Access to testing and associated pre- and post-test counselling
    - **Therapies**
        a) Care pathways and support (next steps)
        b) Hand-off back to primary care
        c) Access to non-pharmaceutical therapies e.g., Allied Health
        d) Access to rare diseases resources for primary care providers to assist in different stages of the patient journey – "galaxy guide"
    - **Interdisciplinary - ELSI**
        a) Data sharing between primary and secondary/tertiary care
        b) Primary care education
        c) Equitable access and cultural safety– e.g., Indigenous primary care organisations
        d) Role of nursing staff
    - **Patient Engagement**
        a) Direct patient phenotyping
        b) Patient resources "galaxy guide" – pre- and post-diagnosis
        c) Patient-centered outcomes

Background of the Topic:

Individuals living with rare diseases typically present first, and often recurrently, to their primary care providers (PCPs). These PCPs can also provide a medical home and coordinating centre that is central to the patients on their rare diseases journey.

PCPs are community based, may provide care to multiple family members, and have an awareness of local resources. Primary care may be provided by physicians, such as specialists in paediatrics, internal medicine, family practice, genetic counsellors, nurses, nurse practitioners, pharmacists, physical

therapists, occupational therapists, respiratory therapists, and others, involved in providing primary care services may be generalised or focused to specific groups such as indigenous populations, and patients in developing nations.

PCPs are therefore central to all aspects of patients' experiences during their rare diseases journey, be it in shortening the diagnostic odyssey, data sharing, access to approved and investigational therapies, care coordination, and social outcomes.

Therefore, this taskforce is critical to all IRDiRC goals and traverses all "IRDiRCians". To realize IRDiRC Goals, especially Goal 1, community-level providers must be engaged.

Goal 1 states: All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

Several tasks to be considered for additional emphasis and discussions of the Task Force include the following:

- Develop gateway and point of care services to provide ready access to collaborative multi- and interdisciplinary diagnostic, treatment, and research teams who communicate back to primary care.
- Provide red flag pathways, enhanced referrals, improved diagnostic access, gatekeeper services, to identify and expand pathways to and from primary care providers and tertiary care centers.
- Care coordination.
- Expand emphasis at specialty clinics at dedicated centers of excellence and provide appropriate research infrastructure for clinical evaluations and hubs of optimal patient-centered care with referrals from community hospitals and centers.
- Facilitate awareness of current rare diseases initiatives, clinical trials, patient registries, natural history studies, and information sources to utilize and participate on a global basis.
- Identify needs to meet supply chain demands of access to medicine despite poor storage conditions, short shelf life, limited size of orders, and the cost of international transport.
- Develop dedicated multidisciplinary, team approach to rare diseases at national and local community levels to address physical, mental, and psycho-social needs of patients and families living with uncertainty, isolation from mainstream medicine, and stigmatization.
- Assess expanding telemedicine and telehealth initiatives to link all communities with tertiary care centers.
- Utilize social media or mobile technologies such as smart phones to establish active partnerships throughout the research and treatment processes.
- Develop support for World Health Organization's (WHO) programs to ensure that all people including indigenous and minority populations have access to diagnostic, preventive, curative, and rehabilitative health services.
- Expand awareness, advocacy, and outreach to everyone including those with low income, poor literacy, minority ethnic status, indigenous populations, and living in underserved and marginalized populations in both urban and rural areas.
- Share access to data and bio-specimens, patient engagement activities, and sustainable and scalable research models to translate the advances in genomics, clinical phenotyping, epidemiology, functional studies, environmental research, imaging procedures,

bioinformatics, health informatics, and communications technology to deliver better prevention, diagnostics, and novel therapeutic interventions.

Other Considerations for Possible Discussions:

- Expand participation at centers of excellence, research consortia, and networks with referral from primary care health care providers with medical specialties such as family practice, internal medicine, pediatricians, physical therapists, genetic counselors, respiratory therapists, occupational therapists, pharmacists, and nurses.
- Identify necessary services at community hospital and service locations.
- Diagnostic services required for successful transfer to the community.
- Genomic sequencing capabilities, availability of interpretation of results, and transfer of results to patients and families.
- Telehealth and Tele-Medicine services in community-based hospitals and in all communities – isolated, rural, and urban – with tertiary care centers.
- Mental health services.
- Provide access to diagnosis and care to indigenous populations and minority populations worldwide. How do we provide to other countries information gained in studies of indigenous genomics in native populations?
- What information is needed?
    a) Access to ongoing, planned, and completed clinical trials
    b) Access to patient advocacy groups
    c) Referral procedures from community practices to specialty practices

Process and/or Timeline:

- Assemble Task Force team of members and Co-Chairs from recommendations of IRDiRC members and scientific and constituent committees (Date Q4 2021).
- Hold first virtual meeting (Date Q4 2021).
- Develop a scope of the challenge and state-of-play document (Q1 2022).
- Develop an asset map with the identification of the international research teams working on this challenge (Q1/Q2 2022).
- Identify a targeted workshop that address an area(s) of priority (Q2 2022).
- Develop objectives and outcomes for a workshop focused on a specific challenge(s) to be held in (Q2 2022).
- Hold workshop (Q3 2022).
- Disseminate workshop outcomes (Q3 2022).

Product/Output:

- Review paper on the scope of the problem and strategies to address the challenges.
- Collaborative groups focusing on a particular challenge or group of challenges.
- Additional workshops in other related areas may be identified as necessary.

Budget Requirement:

- Administrative support
- Workshop support for 30 participants