Canadian Organization for Rare Disorders

21 October 2015

Denise M. Perret, QC
Assistant Deputy Minister, Alberta Health
Strategic Planning and Policy Development
19th Floor, ATB North Plaza, 10025 Jasper Avenue
Edmonton, Alberta, T5J 1S6

Dear Mr. Perret,

On behalf of the Canadian Organization for Rare Disorders (CORD) and all of the patients and families living with rare diseases in Alberta, thank you for the opportunity to provide input into Alberta's initiative to assess seven conditions that are not currently screened for in Alberta: galactosemia; S, Beta-thalassemia; sickle cell anemia; hemoglobin S,C disease; homocystinuria; severe combined immunodeficiency; and tyrosinemia, type 1. In addition, we are pleased to provide comments on issues related to expanding screening programs, screening technologies and on research and policy issues.

With respect to the seven proposed conditions, we point out that these are already included in several other provinces, including Ontario and Saskatchewan. Moreover, we believe there is no stronger endorsement of their fundamental importance than the fact that these are among the 33 core conditions recommended by the USA Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC).

In terms of our comments on expanding screening, technologies, and policy issues, attached please find CORD's "Recommendations for Implementing a World-Class State-of-the-Art Canadian Newborn Screening Programme" sent to the Provincial/Territorial Working Group earlier this year.

Finally, we believe that reference to "metabolic" be removed from the name of the Alberta Newborn Metabolic Screening (NMS) Program since not all conditions to be screened are metabolic, nor indeed can they all be detected with bloodspot testing. Specifically, we urge including screening tests for hearing loss and congenital heart defects.

Sincerely,

Durhane Wong-Rieger, PhD
President
T 416-969-7435
durhane@sympatico.ca