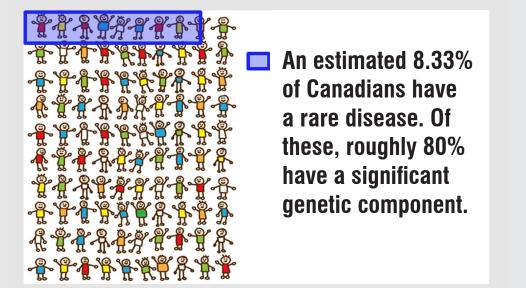


The Health of Canada's Children and Youth: A CICH Profile Genetics and Paediatric Health

Section 4 - Genetic Testing and Screening

## 4.2.3 Genetic Testing – Rare Diseases



Graphic created by CICH using a Shutterstock image and data from Canadian Organization for Rare Disorders www.raredisorders.ca

There are more than 7,000 single-gene diseases. Individually, each of these may be rare; however, when considered together, they are the cause of a significant number of childhood deaths, illnesses, and healthcare costs. Very often, rare disorders remain undiagnosed and have few therapies.

Approximately 30% of infants with a genetic disorder die before their first birthday.<sup>1</sup> Of the children who survive, many experience a comparatively high death rate over their lifetime.<sup>2,3</sup>

There are substantial costs to the healthcare system when caring for children with rare diseases. For example, approximately a third of childhood hospitalizations involve children with rare diseases.<sup>4</sup> These children also have a disproportionate number of hospital admissions and they tend to stay longer in hospital and incur larger hospital bills.

In 2013, the Canadian Institute of Health Research and Genome Canada funded "<u>CARE for RARE</u>" through its Personalized Medicine Initiative. A collaborative team from all regions of Canada, CARE for RARE is working to expand and improve the diagnosis and treatment of rare diseases.<sup>5</sup>

<sup>1</sup> Dodge JA, et al. The importance of rare diseases: from the gene to society. Arch Dis Child. 2011;96:791–2

<sup>2</sup> Dye DE, et al. The impact of single gene and chromosomal disorders on hospital admissions in an adult population. J Community Genet. 2011;2:81–90

<sup>3</sup> Yoon PW, et al. Contribution of birth defects and genetic diseases to pediatric hospitalizations. A population-based study. Arch Pediatr Adolesc Med. 1997;151:1096–103

- <sup>4</sup> McCandless SE, Brunger JW, Cassidy SB. The burden of genetic disease on inpatient care in a children's hospital. Am J Hum Genetics. 2004;74(1):121–7
- <sup>5</sup> Canadian Institute of Health Research and Genome Canada. CARE FOR RARE, <u>http://care4rare.ca/about/overview/</u>

## Implications

The relatively small number of children with rare diseases in Canada, as well as in the world, presents challenges for rare-disease research. Recent governmental initiatives have been launched in an effort to support the application and integration of rare disease research. <u>Orphanet Canada</u> and the <u>International Rare Disease Research Consortium</u> work to rapidly spread research findings regarding rare diseases around the world and to facilitate action based on the findings.



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