



4.2.3 Genetic Testing – Rare Diseases



An estimated 8.33% of Canadians have a rare disease. Of these, roughly 80% have a significant genetic component.

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.¹ Of the children who survive, many experience a comparatively high death rate over their lifetime.^{2,3}

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.⁴ 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 “[CARE for RARE](#)” 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.⁵

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

² 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

³ 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

⁴ 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

⁵ Canadian Institute of Health Research and Genome Canada. CARE FOR RARE, <http://care4rare.ca/about/overview/>

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. [Orphanet Canada](#) and the [International Rare Disease Research Consortium](#) work to rapidly spread research findings regarding rare diseases around the world and to facilitate action based on the findings.

