THE VALUE OF DIAGNOSTIC TESTING FOR PARENTS OF CHILDREN WITH RARE GENETIC DISEASES (RD)

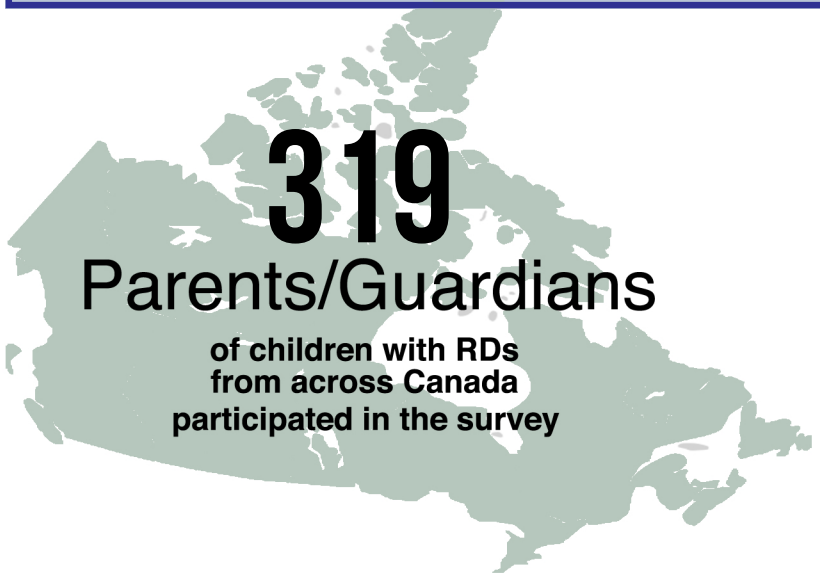
ROUGHLY
50%
of patients with a rare genetic disease (RDs) are undiagnosed

Exome Sequencing

A new technology, called whole exome sequencing (ES), is improving our ability to diagnose individuals with suspected rare genetic diseases and could have a significant impact on patients being assessed in Canadian clinics. However, before ES is incorporated routinely , there must be a clear understanding of its value to patients and families.

In 2016 researchers from the Alberta Children's Hospital at the University of Calgary and Children's Hospital of Eastern Ontario at the University of Ottawa conducted a survey to examine the value of a diagnostic test for families of children with RDs.

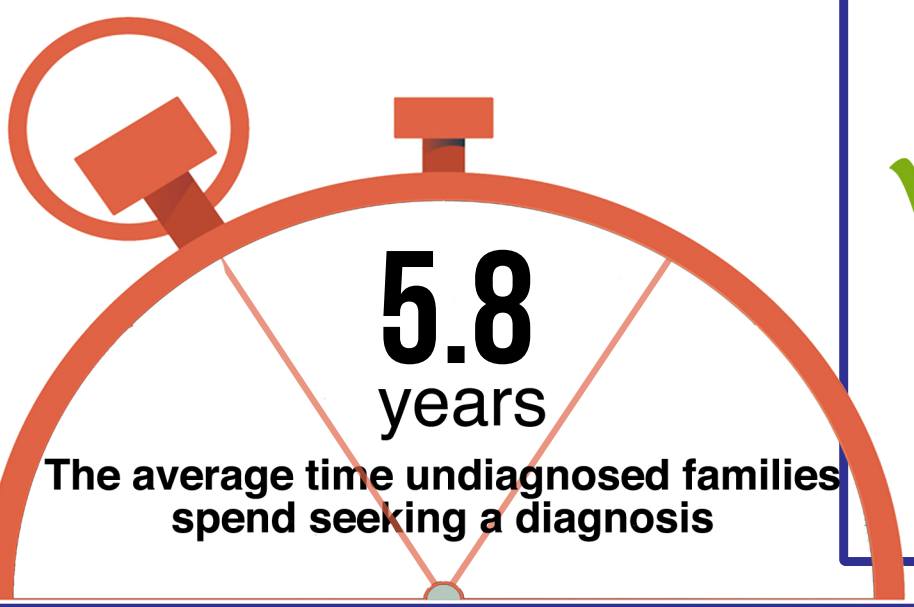
SURVEY RESULTS



319
Parents/Guardians
of children with RDs from across Canada participated in the survey

89% of parents/guardians said their child had undergone genetic testing

66% of parents/guardians said their child had received a diagnosis



5.8
years
The average time undiagnosed families spend seeking a diagnosis

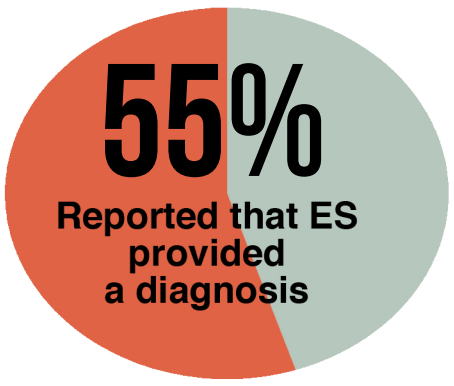
Parents value ES more than the other diagnostic tests we explored:

75% had a positive attitude towards ES

45% had heard about ES

26% had ES

Of the 26%:



Using a series of hypothetical scenarios to estimate parents/guardians preferences identified:

✓ **$6,590** CAD is the amount parents/guardians were willing to pay for ES

✓ **5.2** years is the time parents/guardians were willing to wait for ES

IMPACT

The results from our survey highlight the value of ES as part of the diagnostic process for parents/guardians of children with RDs. These results will be shared with key stakeholders to increase accessibility of this testing for Canadian children who need it.

If you would like more information about the results of this study, you can read our publication or you can contact the Study Coordinator, Karen MacDonald (karenv.macdonald@ucalgary.ca).

Alberta Children's Hospital Foundation · University of Calgary · CARE forRARE · CHEO Research Institute / Institut de recherche