# Prevalence and Utility of Rare Genetic Diagnoses Among Children with Physical Disabilities and/or Neurodevelopmental Disorders (NDDs)

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## Background

- Rare genetic disorders are increasingly identified in children with physical disabilities and NDDs.
- Clinical utility refers to the value of genetic testing in informing patient diagnosis, prognosis, therapeutic management, and overall well-being.[1]

## Research Question

In a population of children with physical disabilities and/or NDDs attending follow-up visits in a tertiary pediatric rehabilitation program: **1)** what proportion have undergone genetic testing and been diagnosed with a genetic condition, and **2)** how frequently has the genetic diagnosis added clinical utility regarding care planning?

## Research Aims

**1** To determine the proportion of neuromotor clients that have received genetic testing.

**2** To determine the proportion of neuromotor clients that have additional investigations, treatments, or information regarding care as a result of their genetic diagnosis.

## Methods

**Retrospective chart review** of clients aged **≤18** years seen for follow-up in the Neuromotor clinic at Holland Bloorview.

Clinical practice guidelines published on **GeneReviews** were reviewed alongside medical charts.

The **Clinician-reported Genetic testing Utility InDEx (C-GUIDE)** is a standardized measurement used to determine the clinical utility of genetic testing.[1]

Genetic diagnoses among children with physical disabilities or NDDs have the opportunity to **add clinical utility** regarding **investigations and prognosis.**

## Conclusions & Relevance

**Increased Genetic Testing** → **Informs Prognosis and Investigations** → **Tailored Clinical Experience for HB Clients and Families**

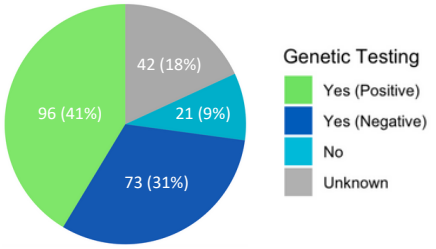
**Informs Development of Clinical Programs that Increase Support for Rare and Ultra-Rare Genetic Care Needs**

## References

1. Hayeems RZ, Luca S, Ungar WJ, Venkataramanan V, Tsiplova K, Bashir NS, et al. The Clinician-reported Genetic testing Utility InDEx (C-GUIDE): preliminary evidence of validity and reliability. Genet Med. 2022 Feb;24(2):430-8.

## Results

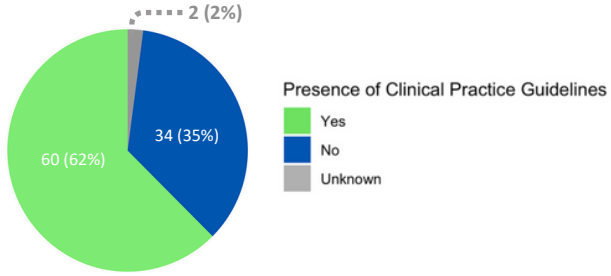
- **169 (72%)** of 232 neuromotor clients had genetic testing, **96 (41%)** had a **positive results**



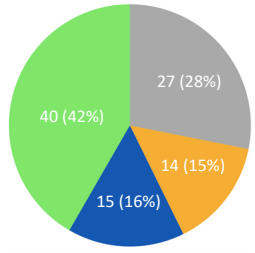

**Types of Genetic Testing:**
**101** Microarray | **33** Exome Sequencing | **24** Single Gene | **23** Gene Panel | **18** Genome Sequencing | **26** Other | **29** Unknown

**Prevalence of Genetic Conditions:**
**54** Ultra-Rare | **21** Rare | **21** Unknown

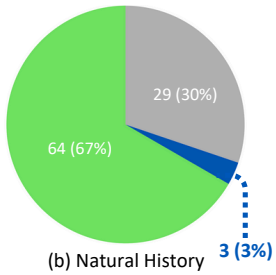
- **60 (62%)** children with positive genetic results had emerging or established **clinical practice guidelines**



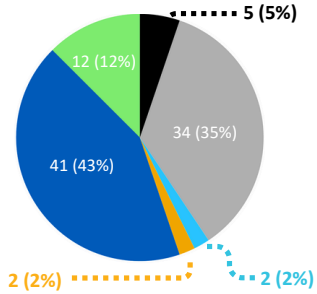

- **40 (42%)** children with positive genetic results had added **investigations**; **64 (67%)** had added discussion regarding **natural history and prognosis**; and **12 (12%)** had added **treatments** based on their diagnosis
- **14 (15%)** investigations were **potentially missed**




(a) Referrals/Investigations




(b) Natural History




(c) Treatment

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