Comparison of clinical profiles among children with and without rare genetic conditions followed in a tertiary pediatric rehabilitation program

Tailor, J. [1,2], Rafeeh, G. [1], Baribeau, D. [1,2], Kawamura, A. [1,2], Peltekova, I. [1,2], Yu, H. [1]

1) Bloorview Research Institute
2) University of Toronto

Background

Rare genetic conditions affect about 1 in 12 Canadians and are common in children with neurodevelopmental or physical disabilities, often leading to complex profiles with multiple co-occurring conditions.

Understanding these profiles can improve care planning, early intervention, and support for families and clinicians.

Research Question

Among children receiving care in a tertiary pediatric rehabilitation program, how do clinical profiles—including co-occurring conditions differ by genetic testing status and presence of a rare genetic condition?

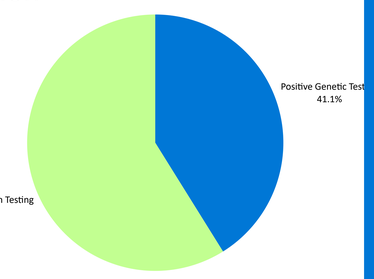
Unknown Genetic Testing → No Genetic Testing → Negative Genetic Testing Result → Positive Genetic Testing Result

Design and Methods

WHO: children/youth (≤18 years)
WHERE: Holland Bloorview Neuromotor Porgram

1. Reviewed 231 charts and collected key clinical information

2. Divided participants into two groups



3. Used Chi-square tests to compare condition prevalence between groups.

Children in the neuromotor clinic with rare genetic conditions show more delays, medical needs, and communication challenges

SCAN ME

Results and Key Themes

PGT = Positive Genetic Testing
NUT = No/Negative Genetic Testing + Unknown

Condition	PGT Yes	PGT %	NUT Yes	NUT %	Chi-square	p-value
Intellectual Disability	24	25.30%	25	18.40%	1.2	0.273
Learning Disability	3	3.20%	20	14.70%	7.08	0.008
Cerebral Palsy	8	8.40%	59	43.40%	31.52	<0.001
Other Developmental Motor	**55**	**57.90%**	**38**	**28.00%**	**19.64**	**<0.001**
Autism Spectrum Disorder	41	43.20%	62	45.60%	0.05	0.817
Global Developmental Delay	**60**	**63.20%**	**46**	**33.80%**	**18.22**	**<0.001**
Language Delay	**83**	**87.40%**	**103**	**75.70%**	**4.11**	**0.043**
Congenital Malformation	**49**	**51.60%**	**45**	**33.10%**	**7.18**	**0.007**
Tube Feeding	13	13.70%	9	6.60%	2.47	0.116
Gastroesophageal Reflux	**13**	**13.70%**	**7**	**5.10%**	**4.13**	**0.042**

5/24 clinical conditions significantly more common in PGT group ($p < 0.05$) = Global Developmental Delay, Language Delay, Congenital Malformation, GER, Other Developmental Motor Conditions

Conclusions

Rare genetic conditions are linked to **more complex developmental, medical, and communication needs** in neuromotor clinic patients.

Next Steps

- Expand the sample: 231 → 700 charts
- Chi-square and logistic regression analyses
- Use insights to support early interventions, inform family-centered planning, and enhance referral pathways

Relavence to Holland Bloorview Clients and Families

Better understanding of how rare genetic conditions affect complex clinical presentations

Help families and care teams anticipate needs, improve interdisciplinary collaboration, and facilitate timely referrals for genetic counseling and specialized services.

References: MacKenzie, A., & Boycott, K. M. (2012). The future is now for rare genetic diseases. Canadian Medical Association Journal, 184(14), 1603.1-1603. https://doi.org/10.1503/cmaj.112-2069
Acknowledgements: Thank you to the WARD Summer Summer Student Research program, Bloorview Research Institute, and the Autism Research Centre for making this research possible.

Bloorview RESEARCH INSTITUTE · UNIVERSITY OF TORONTO · Holland Bloorview Kids Rehabilitation Hospital