The value of multidisciplinary diagnostic assessment of children with unexplained intellectual disability: TIDE Complex Diagnostic Clinic at B.C. Children’s Hospital
The value of multidisciplinary diagnostic assessment of children with unexplained intellectual disability: TIDE Complex Diagnostic Clinic at B.C. Children’s Hospital

Short running title: Multidisciplinary assessment of complex intellectual disability

Clara D.M. van Karnebeek$^{1,2,3,4}$
Tyler Murphy$^{2,3,5}$
Wynona Giannasi$^{3,6}$
Marion Thomas$^{3,7}$
Mary B. Connolly$^{2,3,4,5}$
Sylvia Stockler$^{1,2,3,4}$
and the TIDE CDC Team
TIDE CDC Team:
Linlea Armstrong$^{3,7}$
Susan Baer$^{3,8}$
Anthony Bailey$^{3,8}$
Maria Boldut$^{1,2,3}$
Vann Chau$^{2,3,5}$
Jean-Paul Collet$^{2,3}$
Michelle K. Demos$^{2,3,5}$
Patrice Eydoux$^{3,9}$
Robin Friedlander$^{3,8}$
Gabriella A. Horvath$^{1,2,3}$
Osman S. Ipsioglu$^{2,3,10}$
Mir Lafek$^{1,2,3}$
Anna M. Lehman$^{3,4,7}$
M.E. Suzanne Lewis$^{3,4,7}$
Barbara McGillivray$^{3,7}$
Tanya N. Nelson$^{3,9}$
Millan S. Patel$^{3,4,7}$
Ramona Salvarinova$^{1,2,3}$
Kathryn Selby \textsuperscript{2,3,4,5}  
Christèle du Souich \textsuperscript{3,7}  
Hilary D. Vallance \textsuperscript{3,9}  
Margot I. van Allen \textsuperscript{3,7}  
Wyeth W. Wasserman \textsuperscript{3,4,7,11}  

**Affiliations:**

1. Division of Biochemical Diseases;  
2. Department of Pediatrics;  
3. Treatable Intellectual Disability Endeavour in British Columbia (www.tidebc.org);  
4. Child and Family Research Institute;  
5. Division of Neurology;  
6. President of Howegroup Public Sector Consultants Inc.;  
7. Department of Medical Genetics;  
8. Division of Child and Adolescent Psychiatry; Department of Psychiatry;  
9. Department of Pathology & Laboratory Medicine;  
10. Division of Developmental Pediatrics;  
11. Center for Molecular Medicine and Therapeutics;  

B.C. Children’s Hospital, University of British Columbia, Vancouver, B.C., Canada  

*Corresponding Author:*  
Clara D.M. van Karnebeek, MD PhD  
Clinical Assistant Professor  
Division of Biochemical Diseases, Rm K3-201  
Department of Pediatrics, B.C. Children’s Hospital  
4480 Oak Street. Vancouver B.C. V6H 3V4, Canada  
tel: 604-871-2628 / fax: 604-875-2349  
email: cvankarnebeek@cw.bc.ca
ABSTRACT

Purpose: To evaluate the yield, costs, provider and patient satisfaction of the newly established multidisciplinary Complex Diagnostic Clinic (CDC) for patients with unexplained intellectual disability (ID) at a tertiary care center.

Methods: ID patients with complex systemic and/or neurologic features remaining unexplained after thorough investigations are referred to the CDC, and evaluated by 3 medical specialists followed by multi-disciplinary rounds to formulate a differential diagnosis and testing plan. Analyses included surveys and interviews, (retrospective) chart review, costs calculations and comparison.

Results: During 7 clinics held over 16 months, 24 children (9 male) were evaluated; average age 7 years 11 months (range 9 months-18 years), and previously seen by 2 to 10 specialist services. The diagnostic yield of the CDC was higher than expected with confirmed and working diagnoses in respectively 9 (38%) and 11 (46%) children. Costs savings included fewer trips to the hospital and fewer tests via more streamlined evaluation. Positive feedback was received from both patient families, and medical professionals.

Conclusion: The CDC is an innovative model of personalized medicine and platform for translational medicine, with collaborative specialties successfully interpreting clinical, biochemical, and genomic data to end the burden of a diagnostic odyssey and improve care and outcomes.

KEYWORDS:

intellectual disability, multidisciplinary, clinic, care coordination, diagnostic evaluation, etiology

Full Manuscript submitted to Genetics in Medicine