

# CAUSES Research Clinic Inclusion Criteria

The CAUSES research clinic is a BC pediatric initiative. Genomics consultation referrals may be made for new patients under 17 years old and for existing BC Children's Hospital patients under 19 years old.

Note: Please ensure BOTH parents are available in the event of eventual sequencing and that the family is aware that you are pursuing a genomics referral.

## Inclusion criteria

- 1) Patient presents with a suspected single gene disorder and one or more of the following:
  - Previous genetic investigations have not identified the genetic cause
  - Genome-wide sequencing is more cost-effective than serial single gene or gene panel investigations
  - The associated condition exhibits genetic heterogeneity
  - Consanguinity is present
  - A multi-generation family history strongly suggestive of a Mendelian condition
- 2) The patient presents with a nonspecific clinical phenotype (e.g. multiple congenital anomalies or intellectual disability) of unknown cause after routine clinical evaluation and testing.

## Exclusion criteria

Clinical examples of patients who would not be appropriate include patients with a clinical presentation suggestive of a:

- Infectious, toxic or other non-genetic cause
- Multifactorial condition
- Teratogenic exposure
- Well-delineated chromosomal disorder (e.g. Trisomy 21)
- Mendelian condition for which a single or small number of molecular diagnostic tests or an appropriate and cost-effective gene panel is available



The CAUSES research clinic at BC Children's Hospital provides genome-wide sequencing for children and their parents with complex, undiagnosed disorders. It also provides families with genetic counselling and personalized treatment. Mining for Miracles, through BC Children's Hospital Foundation, committed \$3-million towards the development of CAUSES. Genome British Columbia has pledged to match Mining for Miracles' commitment up to an additional \$1-million. The research clinic is supported by the Provincial Health Services Authority and the University of British Columbia.