

Incidental Findings Policy Division of Genome Diagnostics

The Division of Genome Diagnostics at BC Children's and BC Women's Hospitals (C&W) provides clinical genetic testing for the province of BC.

Genetic test results are intended to aid the management of the presenting genetic disorder and must be used in conjunction with the clinical presentation and other markers of disease status and progression. Pre-test counseling and the consent process must include recognition and discussion of the possibility of incidental findings (IFs), which are defined as clinically relevant findings identified incidentally during analysis (i.e. not actively searched for) that are unrelated to the primary indication(s) for performing the test.

Where possible, analysis approaches minimize the identification of IFs. Thus, the failure to report an incidental finding does not suggest that these genetic changes are not present within the data. Furthermore, the likelihood of detecting IFs may differ across probands, sample type, and analysis approach.

The following guidelines are applied when IFs are identified:

1. Only IFs classified as likely pathogenic or pathogenic are reported;
2. Reportable IFs are largely confined to a pre-defined list of genes for which evidence supports benefit from interventions for pre-symptomatic patients, even when ascertainment has occurred apart from positive family history
3. Actionable childhood-onset IFs are reported, including those identified as part of a primary finding (e.g. a cancer predisposition gene within a large pathogenic deletion);
4. IFs of the following types are not generally reported:
 - a. low-penetrance genetic predispositions;
 - b. pharmacogenetic findings;
 - c. carrier status
 - i. note: carrier status may be reported when there is significant risk to the family for conceiving an affected child;
 - d. biological relationships that differ from those reported by the family, unless necessary in order to avoid an error in diagnostic interpretation and/or recurrence risk.
5. The Division does not offer cascade testing of reported IFs.