

DIRECT TO CONSUMER GENETIC TESTING

- Direct to Consumer Genetic Testing (DTC-GT) provides testing, through direct internet sales, to consumers who wish to know more about how their genetic make-up contributes to their health.
- Various companies offer DTC-GT, with different choices of tests.
- DTC-GT often tests for variants called Single Nucleotide Polymorphisms (SNPs), where one base in the gene sequence is substituted for another. Most SNPs have no effect on health, but some are associated with disease or drug metabolism.
- DTC-GT identifies SNPs at select sites across the human genome and provides risk estimates for diseases such as cancer, heart disease and diabetes, based on findings from prior studies indicating how often that SNP was associated with a condition.



ARE YOU READY TO ANSWER YOUR PATIENT'S QUESTIONS?



RISKS OF DTC-GT

- Testing is often done without a health professional, so the consumer may not realize what they are being tested for or what the impact could be.
- Consumers may be upset to learn they are at 10x increased risk for developing an illness, even if that means the risk has changed only from 0.5% to 5%.
- While advertised to be used to improve health, most companies say DTC-GT results are not for clinical use, but for “information only.”

BENEFITS OF DTC-GT

- If individuals are well educated about the test limitations and how the risk information is reached, then DTC-GT can empower them to take an active role in their health.
- Taking responsibility for health, encourages lifestyle changes that could prevent disease.
- Many consumers find that DTC-GT information is presented in a user friendly manner, with links to further information about diseases, and the studies that the risks are derived from.
- Consumers often enjoy some of the non-health information presented by the companies, such as predicted eye color/hair texture, or information on ancestry.

WHAT ARE SOME OF THE LIMITATIONS OF DTC-GT?

- It does not take into account how lifestyle factors & family history contribute to disease risk.
- For many conditions, genetics contributes only a portion of the risk, and of this, SNPs only play a small part towards disease development.
- The scientific validity of some DTC-GT is in question, because the evidence that a particular SNP conveys a disease risk is based on only one report and has not been validated in further studies.
- Reproducibility between laboratories varies. In a study where identical samples were sent to multiple companies for the same tests, the disease risk estimates were different.
- SNPs are often specific to an ethnic population. For example, a SNP conveying an increased risk of heart disease in a European group may not alter the risk for those of Asian descent.
- DTC-GT is not regulated by Canada Health or the Food and Drug Administration and so there is no governing body to ensure the accuracy and clinical utility of the results.



CASE EXAMPLE BREAST CANCER

Only a small % of breast cancers are hereditary, and have an identifiable genetic cause. Some DTC-GT provide a risk assessment for breast cancer based on SNPs in the genes BRCA1 & 2, which only account for a small fraction of breast cancers. In some cases, the companies only test for 3 particular SNPs, which are only found in the Ashkenazi Jewish population. For women of different ethnic groups, the absence of those SNPs does not lower their risk of breast cancer. However, the average individual may believe they are not at risk for breast cancer, if the test comes back negative.

WHAT TO CONSIDER WHEN INTERPRETING DTC-GT RESULTS

- Do you have information on how the risk estimates were compiled? If not, you might have to investigate the studies done on that variant and the disease risk it conveys.
- Are the results pertinent to your patient? Was the variant studied in the same ethnic population as your patient? Are there medical interventions based on the test results?
- Does the result convey a strong risk? For example, perhaps for the disease being considered, genetics only plays a small part in disease risk.

ADDITIONAL CONSIDERATIONS

- There are many companies that offer DTC-GT. It is important to consider the reputation of the lab and whether the results are meant to be used for clinical purposes.
- If you don't feel equipped to answer all the questions, a referral to the Provincial Medical Genetics Program may be a good option: www.bcwomens.ca/services/medical+genetics/contactus.htm.
- Many DTC-GT companies offer the option of access to a genetic counsellor who can answer questions or provide more information on the test results.



BCCGN provides education for BC clinicians about genomics, access to BC's state-of-the art genomics technologies and support services. **Contact:** Shelin Adam, Network Coordinator, 604 875 2000 ext. 6063, shelin.adam@ubc.ca