Ambry Genetics®
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RESEARCH FOR YOUR PRACTICE

Follow-up after DTC Genetic Testing

CLINICAL CONFIRMATION OF DIRECT-TO-CONSUMER (DTC) REPORTED RESULTS IS CRITICAL TO APPROPRIATE PATIENT CARE

What is Direct-to-Consumer (DTC) Genetic Testing?

- Genetic tests sold directly to consumers without involvement of a physician or insurance company.[1]
- In the US, the Food and Drug Administration (FDA) restricts DTC companies from offering products that function as diagnostic tests and states that consumers and health care professionals should not use DTC results to determine any treatment and that confirmatory testing is required.

WHY THIS MATTERS TO YOU

- DTC tests provide consumers with genetic information, sometimes including variants associated with serious disease.
- Your patients may bring you DTC reports that indicate an increased risk of disease. These tests are often not validated or intended for medical use. Understanding DTC testing limitations is critical for accurate risk assessment and management.
- DTC companies may release raw genetic data and 3rd-party software may be used to interpret the data, which may be inaccurate[2,3].

AMBRY'S STUDY[4]

- 49 cases of variants previously identified by DTC testing sent were for clinical confirmation testing at Ambry.
- **40% of variants analyzed were not confirmed and deemed false positives.**
- 8 variants in *ATM, BRCA1, BRCA2, COL3A1* and *COL5A1* were designated as "increased risk" in DTC raw data files or 3rd-party interpretation, but classified as "benign" by Ambry and other clinical labs.

POINTS FOR YOUR PRACTICE

- Limitations of DTC tests include potential for false positives and incorrect classification of results in raw data interpretation.
- Clinicians should use caution when interpreting DTC test results and should proceed with clinical confirmation by a diagnostic laboratory well-versed in clinical-grade variant detection and classification prior to making medical management recommendations.

REFERENCES

1. aboutgeneticcounselors.com
2. Badalato L, Kalokairinou L & Borry P. Third party interpretation of raw genetic data: an ethical exploration. Eur J Hum Genet 2017;25:1189–1194.
3. Dorschner MO, Amendola LM, Turner EH, *et al*. Actionable, pathogenic incidental findings in 1,000 participants' exomes. Am J Hum Genet 2013;93:631–40.
4. Tandy-Connor S *et al* False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care. Genet Med. 2018 Mar 22.

DIRECT-TO-CONSUMER (DTC) GENETIC TESTING

FREQUENTLY ASKED QUESTIONS

1. Why has DTC genetic testing become so popular?

   - Era of Personalized Healthcare: Genetic testing is being considered by healthy individuals to access more personalized information about their risk for disease, so that they can pursue available preventive options.
   - Accessibility: DTC genetic testing is easily accessible. It is cheap, easy to purchase, and promoted widely.
   - Entertainment Value: Many pursue DTC genetic testing to find out "fun facts" about their family, such as ancestry information.

2. Are there any risks to a patient with DTC genetic testing?

   - Misinterpretation: Since a healthcare provider does not need to be involved in DTC testing, consumers may misinterpret their results, which could lead to medical and psychological risks. The consumer could act on results inappropriately, such as switching or discontinuing a medication or pursuing unnecessary medical tests.
   - False positives or false negative results: DTC labs offer tests that are not intended for medical use, so there is a higher risk of receiving inaccurate results.
   - Unwanted information: DTC testing could reveal unwanted information, such as undisclosed family relationships.

3. What is the difference between DTC genetic testing and clinical laboratory testing?

DTC Testing	Clinical Testing
Technology: SNP array	Technology: Full gene sequencing and deletion/duplication analysis
Not a comprehensive risk assessment: may not include all genes associated with a disease and may not test for all possible mutations within a gene	Comprehensive assessment for one or more diseases: likely to include all known genes associated with the target disease and includes a comprehensive analysis for all possible mutations
Most often not validated	Validated and designed for diagnostic purposes
Results not intended for medical use	Results are intended for medical use with guidance of a healthcare professional

4. If a patient brings me a DTC genetic report or 3rd party data interpretation, what are the best next steps?

Proceed with caution
Do not make medical management recommendations based on the information provided

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Refer to a Genetic Counselor
Find a Genetic Counselor on NSGC.org

→

Consider confirmatory testing
Through an experienced clinical laboratory, when applicable

Learn more about our research here.

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