NHGRI Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

Over 250 Members: Open to individuals, organizations, and industry interested in healthcare provider genomics education; free membership

Convene Project Groups to:

- o Identify educational needs and potential solutions
- Share best practices in educational approaches
- Develop educational resources

NHGRI facilitates dissemination of products from Project Groups

Direct-to-Consumer Genetic Testing Project Group,

led by Co-Chairs Dr. Tracey Weiler and Dr. Houriya Ayoubieh

New resource on genome.gov: <u>Direct-to-Consumer</u>
 <u>Genetic Testing FAQ for Healthcare</u>
 Professionals



Please visit: genome.gov/iscc





- June 6-10, 2022. Events will include panel discussions, webinars, twitter chats and Q & As. Genomics education resources will be tweeted throughout the week #MedGeneEd22. Check out the events:
- <u>https://www.genome.gov/event-calendar/Healthcare-Professionals-Genomics-</u> Education-Week .

•Themes each day are:

June 6: Direct-to-Consumer Genetic Testing
June 7: Pharmacogenomics
June 8: Inclusive Genetics
June 9: Cancer Genomics/OB-GYN
June 10: Rare Diseases

1-page flyer/announcement.

Direct-to-Consumer Genetic Testing Case-studies

Intersociety Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

Direct-to-Consumer Genetic Testing Project Group

Panelists



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Disclosures

Houriya Ayoubieh – No conflicts to disclose Heewon Lee – No conflicts to disclose Rachel Mills – No conflicts to disclose Nguyen Park – No conflicts to disclose Christine Formea – No conflicts to disclose Tracey Weiler – No conflicts to disclose

The Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) provides a venue for individuals to collaborate and develop educational resources. The opinions expressed in this resource do not reflect the view of all ISCC-PEG members, the National Institutes of Health, the Department of Health and Human Services, or the United States government.

Link to Presentation from 2021

ISCC-PEG DTC-GT project group will host a panel of genetics professionals to discuss clinical vignettes related to DTC-GT.

For a refresher about DTC-GT, participants are encouraged to view the 2021 Healthcare Provider Education Week webinar:

DTC-GT for Healthcare Professionals

https://www.youtube.com/watch?v=fcqj6TNq9y0.

What is direct-to-consumer genetic testing (DTC-GT)?



Genetic Testing Models

	Description	Who orders the test?	How is consent obtained before the test?	Who discloses the results?
Direct-to- consumer genetic testing (DTC-GT)	Healthcare professional not involved	Consumer	Direct to consumer via written information	Report issued directly to consumer
Provider- mediated genetic testing (PM-GT)	Consumer- initiated with healthcare professional involvement	Consumer or healthcare professional	Direct to consumer with/without healthcare professional	Report issued directly to consumer with optional access to healthcare professional
Clinic-based genetic testing	Facilitated by healthcare professional	Healthcare professional	By healthcare professional	Facilitated by health- care professional; integrated into clinical care

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Benefits and Limitations of DTC-GT

Benefits

- Increases awareness about genetics
- Easy, inexpensive, no insurance or medical system involved
- Empowers, informs, entertains
 - Ancestry
 - Health

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- Paternity
- Sports/Fitness/Nutrition
- 'Benign' genetic traits

Limitations/Caveats

- Testing methods limited
 - Most often targeted segments analyzed, not full sequencing
- Results can be misleading
 - Potential false reassurance
 - Potential overburden to the healthcare system
 - Clinical confirmation needed
- Concerns about privacy and use of data
- Limited or no informed consent





DTC-GT in the Clinic

A 28-year-old patient (she/her) is establishing care in your clinic and discloses that she is **adopted** and has **no known biological family health history**.

She also relays that she had **DTC-GT** and mentions that her results were **negative** for *BRCA1/BRCA2*.

What issues should we be thinking about?

DTC-GT in the Clinic: Considerations

For adopted patients who pursued DTC-GT

- Elicit the reason for pursuing DTC-GT and determine any underlying concerns.
- Recognize:
 - A positive result/ variant detected does not mean that they will invariably go on to develop the health condition.

- Recognize:
 - Additional testing likely needed to **confirm** or refute the results.
 - Negative, absent, undetected or "reassuring" results may be a false negative.
 - Additional testing may or may **not be covered** by their health insurance.

DTC-GT Result for a Genetic Condition in the Hospital

- 50 yo man (he/him) admitted to hospital for acute exacerbation of heart failure. There is limited family history; no other CAD risk factors.
- ECG: pseudoinfarction pattern.
- Echocardiogram: LVH with preserved systolic function.
- Cardiac MRI: suspicious for amyloidosis
- The next day, the patient's daughter mentions that he had DTC-GT, showing that he has a genetic condition that could damage his heart.
- DTC-GT report Hereditary Amyloidosis (TTR-Related): Two V1121I variants
 What issues should we be thinking about?

DTC-GT for a Genetic Condition: Considerations

For patients who present with a "positive"/variant present DTC-GT result

- Evaluate the patient for signs or symptoms of the condition.
- After appropriate counseling, order confirmatory clinical genetic testing if the result requires increased screening or intervention, and genetic testing is available and feasible.
- When in doubt; refer to a genetics professional.

DTC-GT for a Pharmacogenomic Result

A 65 yo man (he/him) is admitted for myocardial infarction. The physician plans to discharge him on clopidogrel. When reviewing discharge instructions, the patient mentions that he had DTC-GT that showed he can not process certain medications:

• Poor metabolizer for CYP2C19.

You verify that this result is cleared by the FDA for clinical use without validation. So, the care team decides to use ticagrelor which is not dependent on the CYP2C19 pathway.

What issues should we be thinking about?

DTC-GT for a Pharmacogenomic Result

For patients who present with a DTC-GT pharmacogenomic result

- Research variant effect using FDA-approved drug labeling and updated resources; e.g. www.pharmgkb.org, www.cpicpgx.org.
- Review the result to determine the need for additional clinical testing prior to any medication change.
- Refer to a clinical pharmacist.

Resources to help interpret and apply clinical pharmacogenomic test results to patient care

- Table of Pharmacogenomic Biomarkers in Drug Labeling
 - Food & Drug Administration (FDA)
 - <u>https://www.fda.gov/drugs/science-and-research-</u> <u>drugs/table-pharmacogenomic-biomarkers-drug-labeling</u>
- Evidence-based clinical practice guidelines for pharmacogenomics results
 - Clinical Pharmacogenetics Implementation Consortium (CPIC)
 - O <u>https://cpicpgx.org/guidelines/</u>
- Pharmacogenomics Knowledge Resource
 - PharmGKB
 - o https://www.pharmgkb.org/







A parent had direct-to-consumer genetic ancestry testing for their 2 year-old child assigned male at birth. He is generally healthy, though he has been referred to speech therapy for speech delays.

The parents decided to run DTC-GT results through a 3rd party interpretation site for health conditions. That analysis identified:

- VHL Pathogenic variant (von Hippel-Lindau tumor suppressor gene)
- *STK11* Pathogenic variant (tumor suppressor gene)
- *COL5A1* Pathogenic variant (Ehlers Danlos syndrome)
- *CFTR* pathogenic variant (cystic fibrosis)

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What issues should we be thinking about?



Please do not order DTC genetic testing for a child.

3rd Party Interpretation of DTC-GT Results

For patients who present with 3rd party DTC-GT reanalysis results

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- Recognize that third party analysis of genetic test results could yield false or uninformative results.
- Recognize that a "positive test" or variant detected or present may be false positive.
- Recognize that a "negative test," variant absent, or undetectable is uninformative.
- For a report with a "positive test," variant detected or present:
 - Determine if there is a clinical genetic test that could validate the result.
 - After appropriate counseling, order clinical genetic testing for the result.
 - When in doubt, refer to a genetics professional.

Key Takeaways

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- Elicit the reason for pursuing DTC-GT and determine any underlying concerns.
- Determine if the results were obtained directly from the DTC-GT company or is a third party generated report.
- Recognize that third party results may be false.
- Evaluate the DTC-GT result, obtain additional medical, medication and family history, and determine if there is clinical genetic testing that could validate the result.
- Remember the DTC-GT tests are not comprehensive.

Key Takeaways

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- In time sensitive situations where clinical genetic testing may not be readily available:
 - Evaluate for the genetic condition.
 - Research the effect of the DTC-GT pharmacogenomic results and discuss with a clinical pharmacist prior to medication change.
- Recognize that the ethical issue that pertains to pursuing DTC-GT in minors.

Find a genetics professional

- The National Society of Genetic Counselors (NSGC)
 - Information on how to find a genetic counselor nationwide for consultation
 - In person or telehealth
 - <u>https://www.nsgc.org/page/find-a-genetic-counselor</u>
- The <u>American College of Medical Genetics and Genomics</u> (ACMG) offers a search form by type of specialist patients may need.
 - <u>https://www.acmg.net/ACMG/Genetic Services Directory Search.aspx</u>
- The ACMG also has a Find a Genetics Clinic tool
 - <u>https://clinics.acmg.net/</u>

DTC-GT Resources



A https://medlineplus.gov/genetics/understanding/dtcgenetictesting/directtoconsumer/

 Notional Library of Medicine

MedlinePlus				Search MedlinePlus				
Truste	d Health Information for You			About MedlinePlus	What's New	Site Map	Customer	
Health Topics	Drugs & Supplements	Genetics	Medical Tests	Videos & Tools				

 $Home \rightarrow Genetics \rightarrow Help \ Me \ Understand \ Genetics \rightarrow Direct-to-Consumer \ Genetic \ Testing \rightarrow What \ is \ direct-to-consumer \ genetic \ testing?$

What is direct-to-consumer genetic testing?

Most of the time, genetic testing is done through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers determine which test is needed, order the test from a laboratory, collect and send the DNA sample, interpret the test results, and share the results with the patient. Often, a health insurance company covers part or all of the cost of testing.

Direct-to-consumer genetic testing is different: these genetic tests are marketed directly to customers via television, print advertisements, or the Internet, and the tests can be bought online or in stores. Customers send the company a DNA sample and receive their results directly from a secure website or in a written report. Direct-to-consumer genetic testing provides people access to their genetic information without necessarily involving a healthcare provider or health insurance company in the process.

Dozens of companies currently offer direct-to-consumer genetic tests for a variety of purposes. The most popular tests use genetic variations to make predictions about health, provide information about common traits, and offer clues about a person's ancestry. The number of companies providing directto-consumer genetic testing is growing, along with the range of health conditions and traits covered by these tests. Because there is currently little regulation of direct-to-consumer genetic testing services, it is important to assess the quality of available services before pursuing any testing. 🖾 f 💆 🖗 🛃

For more information about directto-consumer genetic testing:

Centers for Disease Control and Prevention (CDC) Genomics and Health Impact Blog: Direct to Consumer Genetic Testing: Think Before You Spit, 2017 Edition!

National Human Genome Research Institute: Direct-to-Consumer Genomic Testing

National Society of Genetic Counselors: What is At-Home Genetic Testing?

Questions?

Submit your questions in the Q&A box.

Please evaluate this webinar and send us your feedback

https://elpasottuhsc.co1.qualtrics.com/jfe/form/ SV_5dTFVYEIwDy77gy

