

ISCC Direct-to-Consumer Genetic Testing

Work Group

Agenda

- Welcome
- Minutes from [Jan 19,2021 Meeting](#)
- Introduction of new student members
- [FAQ](#)
- [POC Tool](#)
- [DTC-GT Learning Modules and Vignettes](#)
- Wrap Up
 - Asks of the ISCC PEG General Membership

DTC-GT FAQ for Genome.gov

<https://www.genome.gov/health/For-Health-Professionals>

DIRECT-TO-CONSUMER GENETIC TESTING

Frequently Asked Questions for Healthcare Professionals- Authors and Reviewers

- Houriya Ayoubieh, MD, Assistant Professor, Texas Tech University Health Sciences Center, El Paso, TX
- Kathleen Blazer, EdD, MS, LCGC, Director, Cancer Genomics Education Program, City of Hope, Duarte, CA
- Dyanna Christopher, American Society of Human Genetics/ National Human Genome Research Institute Genetics and Education Fellow (2018)
- Kathryn Garber, PhD, Associate Professor, Emory University School of Medicine, Atlanta, GA
- Roseann Gammal, PharmD, BCPS, Assistant Professor, Massachusetts College of Pharmacy and Health Science, University School of Pharmacy, Boston, MA
- Linda Ho, MSPP, Presidential Management Fellow, National Center for Advancing Translational Sciences
- Katherine Hyland, PhD, Professor, University of California, San Francisco School of Medicine, San Francisco, CA
- Kimberly Jacoby Morris, PhD, National Human Genome Research Institute | National Institutes of Health | U.S. Department of Health and Human Services, Bethesda, MD
- Heewon Lee, MS, CGC, M Health Fairview- Cancer Risk Management Program, Minneapolis, MN
- Rachel Mills, MS, CGC, Assistant Professor, UNC Greensboro Master of Science in Genetic Counseling Program, Greensboro, NC
- Sarah Robbins American Society of Human Genetics/ National Human Genome Research Institute Genetics and Education Fellow (2019)
- Tracey Weiler, PhD, Associate Professor, Herbert Wertheim College of Medicine, Florida International University, Miami, FL

What is “direct-to-consumer” genetic testing?

- What is “direct-to-consumer” genetic testing?
- What are other types of genetic testing?
- What is the history of DTC-GT?
- What are the benefits of DTC-GT?
- What are the limitations of DTC-GT?
- Is the methodology used by DTC-GT different from clinic based testing?

Possible DTC-GT results

- Possible DTC-GT results
- What is DTC-GT Carrier Testing?
- What are disease risk and health results from DTC-GT?
- What are health-trait and lifestyle results from DTC-GT?

DTC-GT and Adopted Adults

- What if my patient is adopted or has little family history?
- How do I handle DTC-GT results for an adoptee?
- What if an adoptee is considering DTC-GT for health information?
- What if my patient was conceived using donor eggs or sperm and wants to use DTC-GT for health information?

Can DTC-GT provide pharmacogenomic information?

- Can DTC-GT provide pharmacogenomic information?
- Why should healthcare professionals care about pharmacogenomics?
- Can I use DTC-GT pharmacogenomic test results to select optimal medication therapy or adjust the doses of my patients' medications?
- What are the limitations of DTC-GT pharmacogenomic tests?
- How are pharmacogenomic variants named?
- What are the potential clinical effects associated with genetic variation in drug metabolizing enzymes and drug transporters?
- What genetic variants are currently approved by the FDA for DTC-GT pharmacogenomic tests?
- What resources are available to help me interpret and apply clinical pharmacogenomic test results to the care of my patients?

DTC-GT Raw Data, Third-Party Interpretation Services, and Data Privacy

- What should I tell my patient about raw data from direct-to-consumer genetic testing?
- How do direct-to-consumer genetic testing companies protect customer privacy?

DTC-GT and Insurance Coverage

- Can the results of direct-to-consumer genetic testing affect my patient's ability to get insurance?
- What insurance IS protected by GINA?
- What is NOT covered by GINA?

Costs of Genetic Testing

- What about the cost of DTC-GT testing?
- What about the cost of clinical genetic testing?

DTC-GT Information for Patients

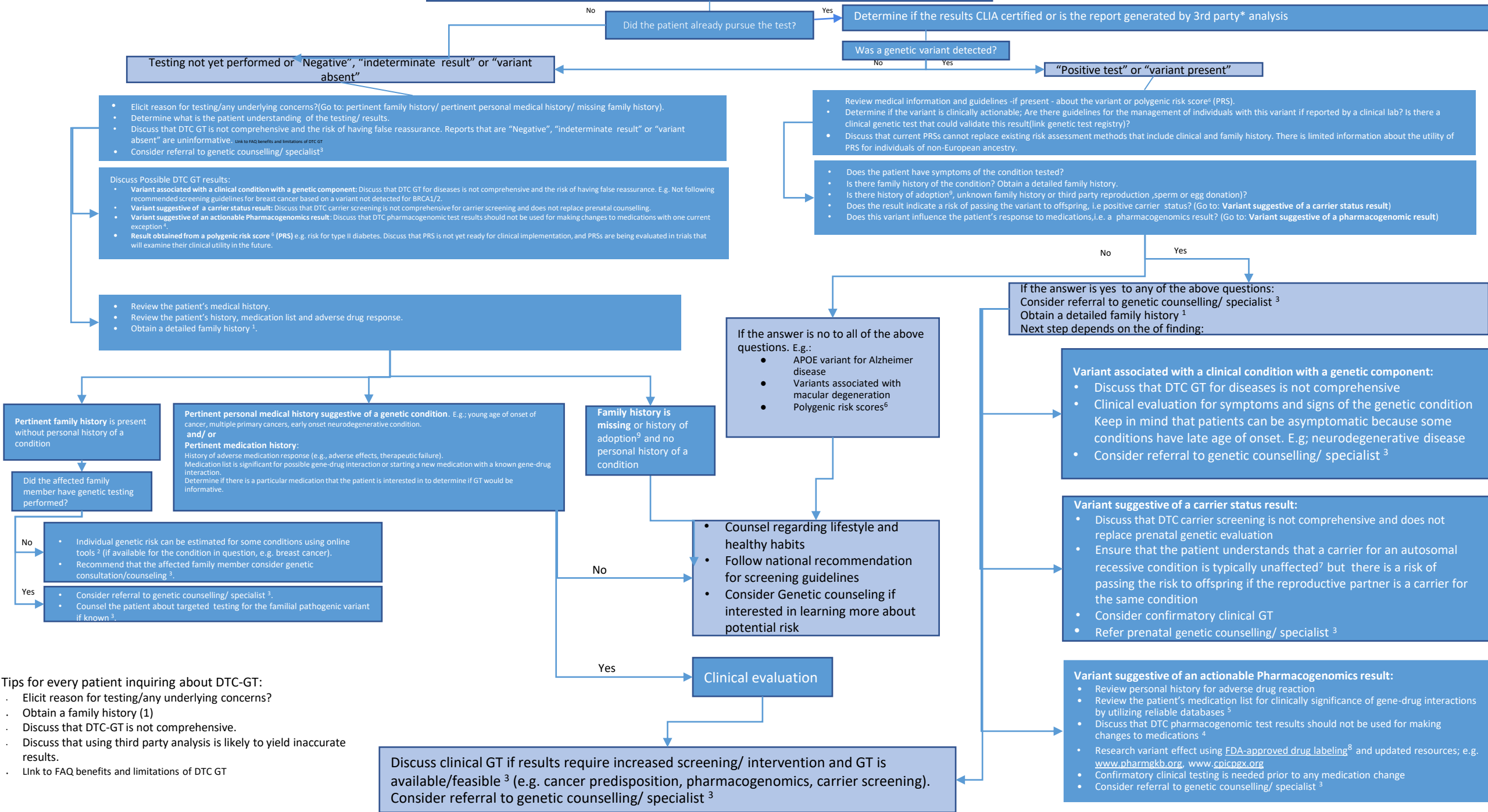
- Where can I find resources about DTC-GT that are targeted to patients?
- Where can I find resources about the diseases and traits from DTC-GT for my patients?
- Where can I find a genetics professional to refer a patient for a genetics consultation?

Tool to Analyze DTC-GT related results for Healthcare professionals

Process of modifying the tool

Aim to adapt to a user friendly format e.g. an App

Health-care professionals: Analyzing Health-DTC GT related results



Tips for every patient inquiring about DTC-GT:

- Elicit reason for testing/any underlying concerns?
- Obtain a family history (1)
- Discuss that DTC-GT is not comprehensive.
- Discuss that using third party analysis is likely to yield inaccurate results.
- Link to FAQ benefits and limitations of DTC GT

Direct to Consumer Genetic Testing for Healthcare professional

Your patient is asking
you whether or not they
should do a DTC-GT.

Your patient has
completed a DTC-GT and
wants to know what to
do with the results.

Your patient is asking you whether or not they should do a DTC-GT.

Q: Elicit reasons for interest in genetic testing

- Determine what is the patient understanding of the testing/ results.
- Discuss that DTC GT is not comprehensive and the risk of having false reassurance. Reports that are “Negative”, “indeterminate result” or “variant absent” are uninformative.
- Consider referral to genetic counselling/ other genetic specialist

Discuss possible DTC-GT results

Return to beginning of tool

Return to previous section

Return to previous page

Your patient is asking you whether or not they should do a DTC-GT.

Discuss Possible DTC GT results:

- **Variant associated with a clinical condition with a genetic component:** Discuss that DTC GT for diseases is not comprehensive and the risk of having false reassurance. E.g. Not following recommended screening guidelines for breast cancer based on a variant not detected for BRCA1/2.
- **Variant suggestive of a carrier status result:** Discuss that DTC carrier screening is not comprehensive for carrier screening and does not replace prenatal counselling.
- **Variant suggestive of an actionable Pharmacogenomics result:** Discuss that DTC pharmacogenomic test results should not be used for making changes to medications with one current exception.
- **Result obtained from a polygenic risk score (PRS)** e.g. risk for type II diabetes. Discuss that PRS is not yet ready for clinical implementation, and PRSs are being evaluated in trials that will examine their clinical utility in the future.

Return to beginning of tool

Return to previous section

Return to previous page

Your patient is asking you whether or not they should do a DTC-GT.

Obtain additional history

- Review the patient's medical history.
- Review the patient's history, medication list and adverse drug response.
- Obtain a detailed family history:

There are online tools to draw a pedigree that you could ask the patient to fill out for your review, e.g.

<https://phgkb.cdc.gov/FHH/html/fhh.html?action=create>

Consider creating a three-generation pedigree: <https://phgkb.cdc.gov/FHH/html/index.html>

<https://www.progenygenetics.com/online-pedigree/>

Pertinent family history without personal history of a genetic condition

Family history is missing or history of adoption and no personal history of a condition

Pertinent personal medical history suggestive of a genetic condition. E.g.; young age of onset of cancer, multiple primary cancers, early onset neurodegenerative condition.

and/ or

Pertinent medication history:

History of adverse medication response (e.g., adverse effects, therapeutic failure). Medication list is significant for possible gene-drug interaction or starting a new medication with a known gene-drug interaction.

Determine if there is a particular medication that the patient is interested in to determine if GT would be informative.

Return to beginning of tool

Return to previous section

Return to previous page

Your patient is asking you whether or not they should do a DTC-GT.

Pertinent family history without personal history of a

Q: Did the affected family member have genetic testing performed?

Yes

No

Return to beginning of tool

Return to previous section

Return to previous page

Your patient is asking you whether or not they should do a DTC-GT.

Pertinent family history without personal history of a

Q: Did the affected family member have genetic testing performed?

No

- Individual genetic risk can be estimated for some conditions using online tools (if available for the condition in question, e.g. breast cancer).
- Recommend that the affected family member consider genetic consultation/counseling

Return to beginning of tool

Return to previous section

Return to previous page

Your patient is asking you whether or not they should do a DTC-GT.

Pertinent family history without personal history of a

Q: Did the affected family member have genetic testing performed?

Yes

- Consider referral to genetic counselling other genetic specialist
- Counsel the patient about targeted testing for the familial pathogenic variant if known.

[Return to beginning of tool](#)

[Return to previous section](#)

[Return to previous page](#)

Your patient is asking
you whether or not
they should do a DTC-
GT.

Q: Is there a limited family history, history of
adoption, third party reproduction?

Yes

No

Return to
beginning of tool

Return to previous
section

Return to previous
page

DTC-GT Learning Modules

How can we leverage the effort put into the vignettes for training healthcare professionals?

ASKS of ISCC PEG Members

- Ideas to further develop the DTC-GT point of care tool/ app
- Representatives from national organizations to help with creating a workshop for DTC GT at national meetings

Next Meeting of DTC-GT Working Group

Tuesday March 16, 2021 @ 1pm