Addressing the Direct-to-Consumer Genetic Testing Knowledge Gap for Non-Genetics Healthcare Professionals

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) DTC-GT Project Group

H Ayoubieh¹, K Blazer², R Mills³, K Garber⁴, H Lee⁵, RS Gammal⁶, L Ho⁷, KM Hyland⁸, K Jacoby Morris⁹, MB Massart¹⁰, E Flowers¹, SM Robbins¹, GM Kuo¹¹, DM Christopher¹², D Messersmith¹³, T. Weller¹³

¹Texas Tech Health Sciences El Paso; ²City of Hope Comprehensive Cancer Center; ³University of North Carolina - Greensboro; ⁴Emory University School of Medicine; ⁵Park Nicollet Frauenshuh Cancer Center; ⁶Massachusetts College of Pharmacy and Health Sciences; ⁷National Center for Advancing Translational Science; ⁸University of California - San Francisco; ⁹National Human Genome Research Institute; ¹⁰University of Pittsburgh; ¹¹Oregon State University - Portland; ¹²University of Colorado - Denver; ¹³Florida International University.

NIH ISCC-PEG Direct-to-Consumer Genetic Testing (DTC-GT) Project Group

- National group of geneticists, genetic counselors, physician assistants, pharmacists, educators and other healthcare professionals
- GOALS
  - Improve genomic literacy of healthcare professionals (HCPs)
  - Enhance the effective practice of clinical medicine in the area of DTC-GT

Background on DTC-GT

- >9 million Americans have used DTC-GT
- Patients are sharing their DTC-GT results with HCPs
- In the past year, 35% of physicians at Kaiser Permanente had patients share DTC-GT results with them
- Only 12% of PCPs agree that physicians have sufficient knowledge to help patients understand DTC-GT results
- 27% of consumers who shared DTC-GT results with their doctors disagreed when asked if their physician understood genetics enough to advise them on the implications of their results.
- Due to this perceived gap in understanding, many HCPs are reluctant to tackle genomic medicine in their clinical practice and would seek opportunities to enhance their knowledge.

ISCC-PEG DTC-GT Project Group Activities

- DTC-GT for Healthcare Professionals Frequently Asked Questions (FAQ)
- 2021 NHGRI Healthcare Provider Genomics Education Week - DTC-GT for healthcare professionals live webinar
- DTC-GT for HCPs - educational videos and Youtube channel
- DTC-GT point-of-care tool for HCPs
- Present DTC-GT for HCPs related topics at national scientific meetings

DTC-GT for Healthcare Professionals FAQ

- To be posted on Genome.gov
- Survey of non-genetics HCPs
  - FAQ content useful
  - Difficult to navigate during a clinical encounter.

DTC-GT Point-of-Care (POC) tool for HCPs

- Algorithm to help HCPs navigate DTC-GT questions from patients in real time.

DTC-GT HCPs - Videos and Youtube Channel

- Youtube Channel: Direct To Consumer Genetic Testing for Healthcare
- Video 1: Direct to consumer genetic testing in primary care

Find out more about ISCC-PEG & the DTC-GT Project Group by visiting https://www.genome.gov/iscc
Direct-to-consumer genetic testing (DTC-GT) is a convenient method for people to obtain genetic information. Over nine million individuals have used DTC-GT services in the U.S., and primary care and specialty physicians are now encountering patients with questions regarding their DTC-GT results. In a recent survey of Kaiser Permanente physicians, 35% of respondents reported that patients had shared their DTC-GT results with them in the past year. Despite the growing need to manage these patient questions, only 12% of surveyed primary care physicians agree that physicians have sufficient knowledge to help patients understand the results of DTC-GT. In a survey of consumers who shared DTC-GT results with their doctors, 27% disagreed with the idea that their physician understood genetics enough to advise them on the implications of their DTC-GT results. Due to this perceived lack of knowledge, many healthcare professionals are reluctant to tackle genomic medicine in their clinical practice and would seek opportunities to enhance their genomic knowledge. The National Human Genome Research Institute's Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) DTC-GT Project Group consists of a spectrum of genetics professionals who aim to improve the genomic literacy of healthcare professionals and enhance the effective practice of clinical genomic medicine in the area of DTC-GT. The ISCC-PEG DTC-GT Project Group has created a Direct-to-Consumer Genetic Testing for Healthcare Professionals Frequently Asked Questions (FAQ) to be housed on Genome.gov. Non-genetics healthcare professionals were surveyed about the FAQ. They found the FAQ content useful, but remarked that it would be difficult to navigate in real time during a clinical encounter. In addition to the FAQ, the DTC-GT Project Group has participated in the 2021 NHGRI Healthcare Provider Genomics Education Week, presenting an educational webinar and short YouTube videos about DTC-GT. The Project Group is also developing a point-of-care tool for healthcare professionals to facilitate an outpatient encounter pertaining to DTC-GT. The Project Group then created 11 DTC-GT patient scenarios, each of which were used to validate the DTC-GT point-of-care tool. The point-of-care tool has now been implemented in the Qualtrics software, as a proof of concept, with the intention to develop a mobile app/website that can support healthcare professionals as they respond to their patients queries about DTC-GT results. Products and outcomes from the DTC-GT Project Group's work will be presented.
Testing not yet performed or "Negative", "indeterminate result" or "variant absent"

- Elicit reason for testing/any underlying concerns?
- 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 not informative.
- 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 that the risk of having false reassurance. E.g.: APOE variant for Alzheimer disease
  - Variant suggestive of a carrier status result: Discuss that DTC carrier screening is not comprehensive and does not replace genetic counseling.
  - Variant suggestive of a clinical condition: Discuss that DTC pharmacogenomic test results should not be used for making changes to medications with one current exception: Variants that are associated with increased risk or used to generate PRS are not clinically used to for medical decision making. E.g.: APOE variant for Alzheimer disease
  - Variant associated with risk for diabetes or Polycystic ovary syndrome:

Tips for every patient inquiring about DTC-GT:

- GT Genetic testing
- DTC Direct-to-consumer

* What is a CLIA certified lab? The Clinical Laboratory Improvement Amendments (CLIA) regulate laboratory testing and are enforced by CMS. Labs must be approved by CMS and have a CLIA certificate of compliance. Clinical evaluation for symptoms and signs of the genetic condition

Determine if the results CLIA certified or is the report generated by 3rd party analysis

- Review medical information and guidelines - if present - about the variant or polygenic risk score (PRS)
- Determine if the variant is clinically actionable. Are there guidelines for the management of individuals with this variant if reported by a clinical lab? Is there a clinical geno test that could validate this result (link to a genetic test registry)

- Discuss that variants associated with increased risk and PRS cannot replace existing risk assessment methods that include clinical and family history. There is limited information about the utility of PRS for individuals of non-European ancestry.

- Does the patient has symptoms of or clinical risk factors of the condition tested?
- Is there clinical laboratory testing to evaluate this condition? E.g.: In case of studies for HFE variant associated with hereditary hemochromatosis
- Is there family history of the condition? Obtain a detailed family history.
- Is there history of adoption?, unknown family history or third party reproduction (sperm or egg donation)?
- Does the result indicate a risk of passing the variant to offspring? (Go to: Variant suggestive of a carrier status result)
- Does this variant influence the patient’s response to medications, e.g.: a pharmacogenomics result? (Go to: Variant suggestive of a pharmacogenomics result)

Discusses the next step depends on the finding:

- Variant associated with a clinical condition: Discuss that DTC GT for diseases is not comprehensive
- Clinical evaluation for symptoms and signs of the genetic condition
- Keep in mind that patients can be asymptomatic because some conditions have late age of onset. E.g.: neurodegenerative disease
- Consider referral to genetic counseling/specialist

- Variant suggestive of a carrier status result:
  - Discuss that DTC carrier screening is not comprehensive and does not replace prenatal genetic evaluation
  - Ensure that the patient understands that a carrier for an autosomal recessive condition is typically unaffected but there is a risk of passing the risk to offspring if the reproductive partner is a carrier for the same condition
  - Consider confirmatory clinical GT
  - Refer prenatal genetic counseling/specialist

- Variant suggestive of an actionable Pharmacogenomics result:
  - Review personal history for adverse drug reaction
  - Review the patient’s medication list for clinically significant interactions
  - Discuss clinical GT if results require increased screening/intervention and GT is available/feasible 1 (e.g.: cancer predisposition, pharmacogenomics, carrier screening).
  - Consider referral to genetic counseling/specialist

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1 There are several tools to show a pedigran that you would not see with a fishnet and more. e.g. http://www.phylip.org/phylnet/Phylnet.html
2 If the answer is "No" to any of the above
3 Would DTC pharmacogenomics be allowed for this drug? Clinical evaluation of the condition tested with a genetic component. Clinical evaluation for symptoms and signs of the genetic condition
4 Variants that are associated with increased risk or used to generate PRS are not clinically used to for medical decision making. E.g.: APOE variant for Alzheimer disease
5 Focused variant effect using reliable databases
6 Discussion is required prior to any medication change
7 Consider referral to genetic counseling/specialist

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