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Christina Hansen
The University of Vermont

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Evaluation of growth in clinical genetics competency among PCPs participating in the UVMHN Genomic DNA Testing Program

Christina Hansen, LCOM 2023

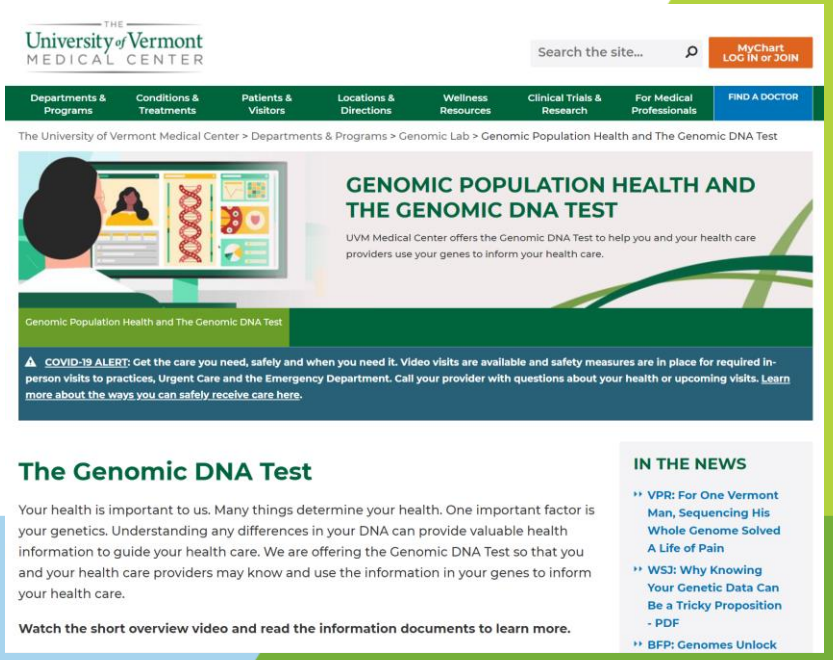
Family Medicine Clerkship
May/June 2021

Project Mentors:
Robert Wilden, MD; Aaron Reiter, MD

The University of Vermont

2: Problem Identification

- **Genomic data** is increasingly important to **patient-centered clinical care** and is now often managed by non-geneticists including **primary care providers**.[1]
    - Identifying genetic factors for disease protection or risk can support clinical decision making for both patients and their family members.
- **UVM Health Network Family Medicine** practices have recently implemented **"The Genomic DNA Test" pilot program** to begin a concerted effort in offering genetically informed primary care to all patients.
    - The program aims to increase the number of participating primary care providers stepwise over time.
    - *Some providers may find integration of genomic testing and discussion of clinical genetics issues with patients to be challenging, particularly given the relatively recent change toward emphasis on these topics in medical training curricula.*[2]



3: Public Health Cost and Considerations

- Given the lack of widespread genomic screening in the United States, it is difficult to estimate the public health opportunity cost to patients who go without receipt of diagnosis, or one that is late or incorrect.
- Among patients with previously undiagnosed disease, “omics-based” precision medicine in general may yield a diagnosis rate of ~35%, with ~1 in 5 newly diagnosed patients receiving new therapeutic recommendations.[3]
- A 2019 Australian study found population level genomic screening of young adults would be highly cost-effective compared to targeted testing. [4] Specifically reducing:
    - Variant-attributable cancers by 28.8%
    - Cancer deaths by 31.2 %
    - Combined incidence of cystic fibrosis, spinal muscular atrophy, and fragile x syndrome (CF/SMA/FXS) by 24.8%
- According to Healthy Vermonters 2020, 166 in 100,000 Vermonters will die of cancer each year. [5]
- 88% of adult Vermonters report having a usual primary care provider. [5]
- Providing better supports and education to our genetic testing PCPs could ultimately bolster efforts to expand the Genetic DNA Testing Program and allow us to offer genetic screening to more patients.

4: Community Perspectives

Dr. Robert Wilden, MD- UVMHN Genomic Medicine Program

Q: From your perspective, what challenges do you believe medical providers face as part of this initiative?

The challenges are different for each provider type. Primary care providers have a less complete grasp of genetics and genetic concepts yet are better than geneticists at perceiving opportunities for preventative care through a variety of risk assessments and their integration with other patient-specific parameters. Genetic experts are adept at distilling sometimes complex concepts and describing Mendelian risks to patients in longer sessions than is practical for PCPs. Transitioning this to primary care while making it practical will require novel partnerships that transcend traditional siloed practice paradigms. Effecting provider education during patient genomic result handoffs is one strategy that needs to be validated.

Dr. Aaron Reiter, MD- Family Physician, UVMMC South Burlington

Q: How do you perceive your role as a family physician with respect to genomics screening?

As a steward and educator of patient's health and with the knowledge that 1 in 4 to 1 in 5 of diagnoses being attributable to genes, it is my duty to incorporate this field into my clinical repertoire.

Q: What challenges have you encountered incorporating genomic screening into your own practice?

Challenges include electronic medical record difficulties, time constraints, patient med knowledge limitations and biases… clinic resource limitations.

5A: Intervention and Methodology

- Aims of this project:
    - Gather information from current participating providers on how participation in the genomic testing program has influenced their personal knowledge of and comfort with clinical genetics topics.
    - Identify practice patterns of patient counseling based on specific genetic test results.
- Intervention:
    - Develop a pilot survey for primary care physicians participating in Genomic DNA Testing program. Survey questions will aim to assess changes in individual's clinical genetics knowledge base as well as comfort with interpreting the program-provided genetic reports and with counseling patients on findings.
    - Review PCP feedback and incorporate into final survey structure and content.

5B: Intervention and Methodology- Pilot Survey

Genomic DNA Testing Program PCP Survey

1) For approximately how many patients have you ordered the Genomic DNA Test to date?
    a. 0-5
    b. 5-10
    c. 10-20
    d. 20-30
    e. 30+

2) I feel genomic testing is relevant to my clinical practice.
    a. Strongly disagree to strongly agree

3) Participation in the Genomic DNA Test Program has increased my knowledge of clinical genetics topics.
    a. Strongly disagree to strongly agree

4) Which elements of the Program have contributed to your increased knowledge? (Check all that apply)
    a. Participating in the program's planning and implementation activities
    b. Patient-oriented test education materials (video and patient packet)
    c. Provider-oriented education materials (training sessions, on-boarding videos)
    d. Reading the tested patients' Genomic Medicine Action Plan (GMAP) documents
    e. Provider-to-provider discussions (among PCPs or with specialists)

5) From question 4's options, what single element provided the most effective and efficient education for you?
    a b c d e none other _____

6) Specific areas/diagnoses where my knowledge has changed or evolved:

7) Participation in the Genomic Testing Program has increased my confidence in providing guidance to patients with identified significant genetic variants.
    a. Strongly disagree to strongly agree

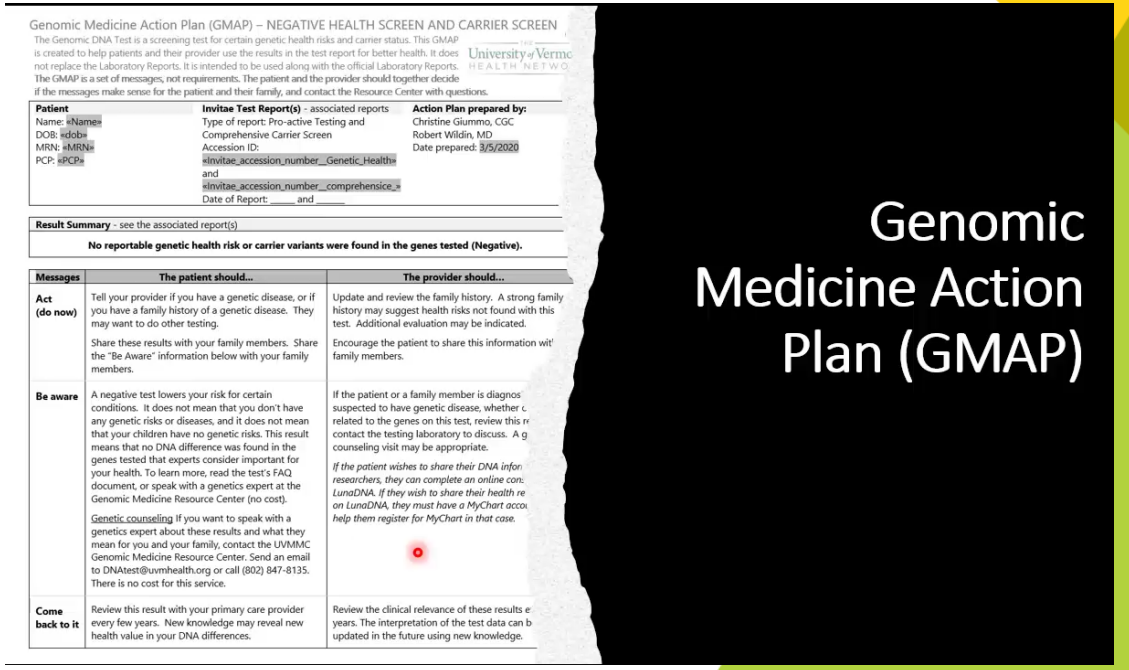
8) I feel well equipped to counsel patients on commonly identified genetic variants (e.g. Hemochromatosis, alpha-1-antitrypsin, autosomal recessive carrier status).
    a. Strongly disagree to strongly agree

9) I feel I would benefit from additional specific training on clinical genetics topics.
    a. Strongly disagree to strongly agree

10) Specific areas where I would appreciate more guidance (e.g., in-office trainings, interpretation support):

6: Results

- Among the providers asked, feedback was generally very positive. Respondents agreed that the survey will be a useful tool to expand and improve the pilot program.
- Suggested revisions largely emphasized revisions for more specific question wording.
- As there are multiple educational aspects to onboarding new participating providers, the respondents encouraged us to refer to certain learning session and materials (such as the GMAP, right) to elicit more specific feedback.



Genomic Medicine Action Plan (GMAP) – NEGATIVE HEALTH SCREEN AND CARRIER SCREEN

The Genomic DNA Test is a screening test for certain genetic health risks and carrier status. This GMAP is created to help patients and their provider use the results in the test report for better health. It does not replace the Laboratory Reports. It is intended to be used along with the official Laboratory Reports. The GMAP is a set of messages, not requirements. The patient and the provider should together decide if the messages make sense for the patient and their family, and contact the Resource Center with questions.

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Patient	Invitae Test Report(s) - associated reports	Action Plan prepared by:
Name: «Name» DOB: «dob» MRN: «MRN» PCP: «PCP»	Type of report: Pro-active Testing and Comprehensive Carrier Screen Accession ID: «Invitae_accession_number__Genetic_Health» and «Invitae_accession_number__comprehensice_» Date of Report: _____ and _____	Christine Giummo, CGC Robert Wildin, MD Date prepared: 3/5/2020

Result Summary - see the associated report(s)

No reportable genetic health risk or carrier variants were found in the genes tested (Negative).

Messages	The patient should...	The provider should...
Act (do now)	Tell your provider if you have a genetic disease, or if you have a family history of a genetic disease. They may want to do other testing. Share these results with your family members. Share the "Be Aware" information below with your family members.	Update and review the family history. A strong family history may suggest health risks not found with this test. Additional evaluation may be indicated. Encourage the patient to share this information wit family members.
Be aware	A negative test lowers your risk for certain conditions. It does not mean that you don't have any genetic risks or diseases, and it does not mean that your children have no genetic risks. This result means that no DNA difference was found in the genes tested that experts consider important for your health. To learn more, read the test's FAQ document, or speak with a genetics expert at the Genomic Medicine Resource Center (no cost). Genetic counseling If you want to speak with a genetics expert about these results and what they mean for you and your family, contact the UVMMC Genomic Medicine Resource Center. Send an email to DNAtest@uvmhealth.org or call (802) 847-8135. There is no cost for this service.	If the patient or a family member is diagnos suspected to have genetic disease, whether related to the genes on this test, review this r contact the testing laboratory to discuss. A g counseling visit may be appropriate. *If the patient wishes to share their DNA infor researchers, they can complete an online con LunaDNA. If they wish to share their health re on LunaDNA, they must have a MyChart acco help them register for MyChart in that case.*
Come back to it	Review this result with your primary care provider every few years. New knowledge may reveal new health value in your DNA differences.	Review the clinical relevance of these results e years. The interpretation of the test data can b updated in the future using new knowledge.

Image from a provider onboarding video, explaining the Genomic Medicine Action Plan (GMAP), a dual patient-provider facing document. The GMAP summarizes next steps for both patients and providers following detection of a clinically relevant genetic variant.

7: Limitations and Evaluation of Effectiveness

- **Limitations**
  - Individual PCP feedback might have been artificially varied or limited due to different levels of personal involvement with the program.
  - Patient participation has been relatively limited to date, suggesting fewer providers have so far encountered many instances of difficult genetic counseling conversations.
  - Due to the limited duration of this rotation the improved survey has not yet been formally deployed to all PCPs.
- **Effectiveness**
  - Based on positive PCP feedback, we feel this project has been successful in its goal of developing a comprehensive pilot survey to evaluate competency concerns of participating providers in the Genetic DNA Testing Program.
  - Provider feedback has been useful in improving survey question clarity and content.
  - The survey was well received, and we anticipate gathering useful information on ways to improve provider support and engagement in this program.

8: Recommendations for Future Interventions

- Deploy survey to all participating UVMHN Family Medicine providers participating in the Genomic DNA Test pilot program. Relevant insights from the survey may include:
    - Suggested improvements to educational elements such as onboarding training sessions and patient-provider facing clinical action plans.
    - Requests for further training or support around specific genetic results
    - Ideas for how to better support new PCPs joining the initiative
- Future projects will likely address other strategies for supporting program expansion, such as:
    - Assessing feasibility for inclusion of other specialties (e.g., Ob-Gyn, Outpatient Internal Med)
    - Identifying barriers to entry or concerns of providers who elect not to participate

9: References

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