

## Primary Care Clinicians Use of Genomics and Pharmacogenomic Testing

Heather Bittner-Fagan MD, MPH

Brian Stello MD  
*Lehigh Valley Health Network*, [Brian.Stello@lvhn.org](mailto:Brian.Stello@lvhn.org)

Christopher V. Chambers MD

Geoffrey Mills MD, PhD

Beth Careyva MD  
*Lehigh Valley Health Network*, [beth\\_a.careyva@lvhn.org](mailto:beth_a.careyva@lvhn.org)

*See next page for additional authors*

Follow this and additional works at: <https://scholarlyworks.lvhn.org/family-medicine>



Part of the [Medical Genetics Commons](#), [Medical Pharmacology Commons](#), and the [Primary Care Commons](#)

---

### Published In/Presented At

Bittner, H., Stello, B., Chambers, C., Mills, G., Careyva, B., Johnson, M., Axell-House, D., & Rosenthal, M. (2014, June 6-7). *Primary care clinicians use of genomics and pharmacogenomic testing*. Poster presented at: The Delaware Academy of Family Physicians (DAFP) Annual Scientific Assembly, Newark, DE.

This Poster is brought to you for free and open access by LVHN Scholarly Works. It has been accepted for inclusion in LVHN Scholarly Works by an authorized administrator. For more information, please contact [LibraryServices@lvhn.org](mailto:LibraryServices@lvhn.org).

---

## Authors

Heather Bittner-Fagan MD, MPH; Brian Stello MD; Christopher V. Chambers MD; Geoffrey Mills MD, PhD; Beth Careyva MD; Melanie B. Johnson MPA; Dierdre B. Axell-House BA; and Michael P. Rosenthal MD

# Primary Care Clinicians' Use of Genomic and Pharmacogenomic Testing

Heather Bittner Fagan, M.D., MPH<sup>1</sup>, Brian Stello, M.D.<sup>2</sup>, Christopher V. Chambers, M.D.<sup>3</sup>,  
Geoffry Mills, M.D., PhD<sup>3</sup>, Beth Careyva, M.D.<sup>2</sup>, Melanie Johnson, MPA, Dierdre Axell-House, BA<sup>3</sup>, Michael Rosenthal, MD<sup>1</sup>  
Christiana Care Health Systems<sup>1</sup>; Lehigh Valley Health Network<sup>2</sup>; Thomas Jefferson University<sup>3</sup>

## INTRODUCTION

- The Human Genome Project has led to an exponential increase in the potential use of genetic information in health care.
- The clinical utility of most of this testing is undetermined
- Healthcare professionals do not have substantial education in genetic medicine.
- Despite these barriers, genomics will continue to integrate into clinical practice.

## PURPOSE

To assess the awareness, experience, and preparedness of regional primary care providers regarding genomic tests.

## METHODS

- A convenience sample of primary care physicians at JeffCare, the Lehigh Valley Health Network, and the Christiana Care Health System. Initial postcards were sent to describing project. Two weeks later, packets with a link to the survey, a printed copy, a stamped/addressed return envelope and a \$5 gift card were sent. Surveys were anonymous. A total of 315 responses out of 833 were received; response rate was 37.89%
- A novel survey consisting of 20 questions,
- Data analysis using Excel for age, gender, practice type, specialty, affiliation and EMR use. Bivariate associations were made comparing either personal characteristics or perceived barriers to genomic testing implementation, and confidence in performing genetic testing-based tasks.

**Table 1: Characteristics of the Respondents (N=315)**

Personal Characteristics	n	%
<b>Gender</b>		
Male	166	54.1
Female	141	45.9
<b>Age</b>		
25-34	48	15.6
35-44	78	25.3
45-54	86	27.9
55-64	79	25.6
65-74	11	3.6
≥ 75	6	1.9
<b>Clinical Role</b>		
Family Medicine Physician	204	65.8
Internal medicine Physician	70	22.6
Other	36	11.6
<b>Practice Setting</b>		
Rural	20	10.0
Suburban	108	54.0
Urban	66	35.0
Multiple	22	11.0
<b>Practice Type</b>		
Academic	92	29.2
Private	106	33.7
Both	12	3.8
Neither	105	33.3
<b>Healthcare Affiliate</b>		
JeffCare	68	22.3
CCHS/DAFP	129	42.3
LVHN	108	35.4
Use EMR	275	89.0

**Table 2: Physicians' confidence in tasks related to genomic medicine (N=315)**

Confidence	Some	High	Very High	Total
Take a thorough family history	46.0%	17.8%	3.5%	<b>67.3%</b>
Build a family medical history pedigree chart	33.7%	17.8%	4.1%	<b>55.6%</b>
Assess risk of hereditary disease	42.2%	11.7%	2.5%	<b>56.5%</b>
Order genetic testing for hereditary cancers	23.8%	4.8%	1.3%	<b>29.8%</b>
Discuss risks, benefits, limits to genetic testing for hereditary cancers	32.4%	7.9%	0.6%	<b>41.0%</b>
Counsel patients about whether or not to have genetic counseling	39.4%	11.1%	1.3%	<b>51.7%</b>
Counsel patients about risk reduction strategies based on the results of genetic testing	32.1%	19.4%	7.6%	<b>59.0%</b>

Note: Confidence is defined as the percentage of respondents who had "some" high" or "very high" confidence in ability to perform tasks for genetic-based disease.

**Table 3: Confidence of PCPs based on experience.**

	Take a thorough family history	Build a family medical history pedigree chart	Assess risk of hereditary disease	Order genetic testing for hereditary cancers	Discuss risks, benefits, limits to genetic testing for hereditary cancers	Counsel patients about whether or not to have genetic counseling
Ordered GT (n= )						
Yes	78.3%	65.0%	75.0%	55.0%	56.7%	70.0%
No	64.6%	53.1%	52.0%	23.6%	37.0%	47.2%
<i>p-value</i>	<0.05	0.09	<0.001	<0.001	<0.05	<0.05
Consulted w/ genetics counselor (n= )						
Yes	75.9%	70.7%	60.3%	43.1%	53.4%	72.4%
No	65.2%	52.0%	55.5%	26.6%	37.9%	46.9%
<i>p-value</i>	0.11	<0.05	0.49	<0.05	<0.05	<0.001

**Table 4: Perceived barriers to genetic testing in primary care (N= 315)**

	Disagree	Strongly Disagree	Total
I have enough time to counsel patients about genetic risk	18.1%	47.9%	<b>66.0%</b>
Most patients can understand the concept of risk	4.4%	34.9%	<b>39.4%</b>
In my practice, I have adequate resources to help patients understand genetic risk	28.9%	50.8%	<b>79.7%</b>
My hospital or network has adequate resources to help patients understand genetic risk	11.1%	21.3%	<b>32.4%</b>

Note: Confidence is defined as the percentage of respondents who had "some" high" or "very high" confidence in ability to perform tasks for genetic-based disease if they had or had not ordered genetic testing or consulted a genetics counselor in the past 6 months.

## DISCUSSION

- Physicians have low confidence for ordering and discussing genetic testing for hereditary cancers
- Ordering genomic testing was common
- Physicians who had previously ordered genetic testing were more confident on most tasks related to genetic testing
- A similar pattern is seen for prior consultation with a genetic counselors.
- Inadequate time and resources to counsel patients related to genetic risk were strong barriers
- Most physicians felt their hospital or network has adequate resources to help patients understand genetic risk and that patients were capable of understanding risk

## FUTURE DIRECTIONS AND RESEARCH

- The predominance of respondents who ordered testing/counseling being only "Somewhat Confident" suggest the need for educational development in primary care
- Evidence-based decision support, ideally integrated in HIT, is needed
- The FDA suspended "23 and me" a Direct-to-Consumer (DTC) genetic-testing company for unsubstantiated clinical claims

## Funding and Approval:

The project was funded through a special grant through the Dean's Office of Jefferson Medical college. [CTR line here]  
The study was approved by the Institutional Review Boards of each institution. Research reported in this publication was supported by the National Institute of General Medical Sciences of the National Institutes of Health under Award Number U54GM104941. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

## Acknowledgements:

The authors would like to acknowledge the Delaware Academy of Family Physicians (DAFP) for their support their participation. We would also like to acknowledge CCHS Medical Imaging for their help in preparing this poster.

## References

- Green ED, Guyer MS. Charting a course for genomic medicine from base pairs to bedside. *Nature*. 2011;470:204-213. doi:10.1038/nature09764
- Bernhardt BA, Zayac C, Gordon ES, Wawak L, Peyeriz RE, Gollust SE. Incorporating direct-to-consumer genomic information into patient care: attitudes and experiences of primary care physicians. *Per Med*. 2012;3(7):683-692. doi:10.2217/pms.12.80
- Goldsmit L, Jackson L, O'Connor A, Skirton H. Direct-to-consumer genomic testing from the perspective of the health professional: a systematic review of the literature. *J Community Genet*. 2013;4(2):169-80. doi:10.1007/s12687-012-0135-8
- Selkirk CG, Weissman SM, Anderson A, Hulick PJ. Physicians' preparedness for integration of genomic and pharmacogenetic testing into practice within a major healthcare system. *Genet Test Mol Biomarkers*. 2013;17(3):219-25. doi:10.1089/gmb.2012.0165
- Powell KP, Christianson C a, Cogswell W a, et al. Educational Needs of Primary Care Physicians Regarding Direct-to-Consumer Genetic Testing. *J Genet Couns*. 2012;21(3):469-478. doi:10.1007/s10897-011-9471-9
- Mainous AG, Johnson SP, Chirina S, Baker R. Academic family physicians' perception of genetic testing and integration into practice: a CERA study. *Fam Med*. 2013;45(4):257-62.
- Passamani E. Educational challenges in implementing genomic medicine. *Clin Pharmacol Ther*. 2013;94(2):192-5. doi:10.1038/clpt.2013.38
- Burke W. Genetic testing in primary care. *Annu Rev Genomics Hum Genet*. 2004;5:1-14. doi:10.1146/annurev.genom.5.061903.180029
- Powell KP, Cogswell W a, Christianson C a, et al. Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing. *J Genet Couns*. 2012;21(1):113-26. doi:10.1007/s10897-011-9390-9