Primary Care Clinicians Use of Genomics and Pharmacogenomic Testing

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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	%
Gender		
Male	166	54.1
Female	141	45.9
Age		
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
Clinical Role		
Family Medicine Physician	204	65.8
Internal medicine Physician	70	22.6
Other	36	11.6
Practice Setting		
Rural	20	10.0
Suburban	108	54.0
Urban	66	35.0
Multiple	22	11.0
Practice Type		
Academic	92	29.2
Private	106	33.7
Both	12	3.8
Neither	105	33.3
Healthcare Affiliate		
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%	**67.3%**
Build a family medical history pedigree chart	33.7%	17.8%	4.1%	**55.6%**
Assess risk of hereditary disease	42.2%	11.7%	2.5%	**56.5%**
Order genetic testing for hereditary cancers	23.8%	4.8%	1.3%	**29.8%**
Discuss risks, benefits, limits to genetic testing for hereditary cancers	32.4%	7.9%	0.6%	**41.0%**
Counsel patients about whether or not to have genetic counseling	39.4%	11.1%	1.3%	**51.7%**
Counsel patients about risk reduction strategies based on the results of genetic testing	32.1%	19.4%	7.6%	**59.0%**

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%
p-value	<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%
p-value	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%	**66.0%**
Most patients can understand the concept of risk	4.4%	34.9%	**39.4%**
In my practice, I have adequate resources to help patients understand genetic risk	28.9%	50.8%	**79.7%**
My hospital or network has adequate resources to help patients understand genetic risk	11.1%	21.3%	**32.4%**

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

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