Impact of non-clinical genetic trait insights on clinical engagement in participants after clinical genetic testing

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Introduction

Patient recall, or a patient's ability to recollect pertinent medical information, is key to effective patient-provider communication and adherence to prevention and treatment plans.[1,2] However, multiple studies report that up to 40-80% of patients are unable to recollect information from their medical encounters.[1,3]

To enhance medical information recall, Color offers 'Discovery', a platform that engages participants and provides them with non-clinical genetic insights such as ancestry, bitter taste perception, lactose intolerance, and alcohol flush response. The focal point of Discovery is to enhance the engagement with genetic information and ultimately empower clients to take relevant clinical actions. Here, we examined the impact of Color Discovery on participants' engagement with clinical results after genetic testing.

Methods

This retrospective study included data from a consecutive cohort of 16,641 Color participants who consented to participate in research. All participants were ordered a Color test by a healthcare provider. This test analyzed genes associated with hereditary risk for cancer, cardiovascular disease, and/or medication response.

Laboratory procedures were performed at the Color laboratory under CLIA and CAP compliance. Variants were classified according to the American College of Medical Genetics and Genomics 2015 guidelines for sequence variant interpretation[4] and all variant classifications were signed out by a board certified medical geneticist or pathologist. All clinical reports were shared with the ordering provider.

All phenotypic information was reported by participants through an interactive, collaborative online health and medication history tool. All participants consented to having their de-identified information and sample used in anonymized studies.

To assess the impact of the engagement platform, we compared clinical actions taken by participants who had consented into Color Discovery and those who had not. Statistical significance was evaluated using a chi-square test, and error bars in bar plots represent Wilson intervals.

Conclusions

- A personal or family history of hereditary disease, especially cancer, drives clinical actions after genetic testing.
- Color Discovery is an effective engagement tool that prompts participants to take clinical actions such as reviewing, downloading, and sharing their clinical reports with their healthcare provider.
- Our results demonstrate that this engagement platform may be an effective way to engage clients and potentially drive better medical information recall and ultimately, adherence.

References

1. Richard C, et al. *Health Expect.* 2017;20: 760–770.
2. Linn AJ, et al. *J Crohns Colitis.* 2013;7: e543–50.
3. Kessels RPC. *J R Soc Med.* 2015;17: 405–424. 2003;96: 219–222.
4. Richards S et al. *Genet Med.* 2015;17: 405–424.

Results

Figure 1. Reporting workflow

Participants are invited via email to Discovery after release of their clinical reports. Discovery includes genetic ancestry, lactose intolerance, alcohol flush response, caffeine consumption, earwax type, bitter taste perception, cilantro preference, and asparagus odor detection.

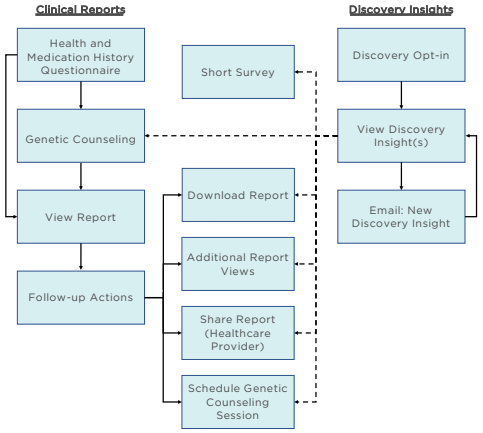

Table 1. Cohort demographics

The majority of participants (n=16,641) were female (70.9%), Caucasian (69.5%), and between 35-50 years old (35.8%).

		Individuals (n)	Population (%)
Total		16,641	100
Gender	Female	11,801	70.9
	Male	3472	29.1
Age (Years)	<35	4469	26.9
	35-50	5954	35.8
	51-60	3365	20.2
	61-70	2147	12.9
	71+	706	4.2
Ethnicity	Caucasian	11,572	69.5
	Ashkenazi Jewish	927	5.6
	Asian	779	4.7
	Hispanic	716	4.3
	Multiple Ethnicities	1128	6.8
	Other*	613	3.7
	Unknown	906	5.4
Discovery Opt-in	Yes	4952	29.8
	No	177	1.1
	No Response	11,512	69.2

*These include ethnicities that were less than 3% in this cohort: African (2.4%), Middle Eastern (1.1%), and Native American (0.2%).

Table 2. Baseline engagement by clinical history

Actions taken within 30-days of clinical report release for participants with a negative result. This analysis only included participants who provided their clinical history but did not opt-in or actively opt-out of Discovery.

	Individuals (n)	Health/ Medication History Updates (%)	First Report View (%)	Additional Report Views (%)	Report Shared/ Downloaded (%)
No Personal or Family History of Cancer or Cardiovascular Disease*	1429	71.6	91.7	42.1	11.1
Personal History of Cardiovascular Disease* *(No Cancer)*	819	70.1	93.3	45.8	22.0
Personal History of Cancer *(No Cardiovascular Disease*)*	943	72.0	94.2	56.4	40.0

*History of Cardiovascular Disease includes familial hypercholesterolemia, arrythmia, cardiomyopathy, arteriopathy, heart attack, heart failure, and stroke.

Figure 2. Actions taken in a Discovery session

Within 30 minutes of viewing a Discovery insight, 40.9% of participants responded to a short survey related to Discovery insights, 25.4% reopened a clinical report for hereditary cancer, cardiovascular disease, or medication response, and 3.1% shared or downloaded their clinical report.

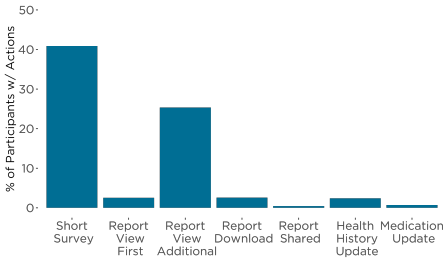

Figure 3. Additional clinical report views

Discovery participants more frequently re-reviewed their clinical report(s) after their initial view, both for negative (71.3 vs 45.5%) and positive reports (83.3 vs 51.7%).

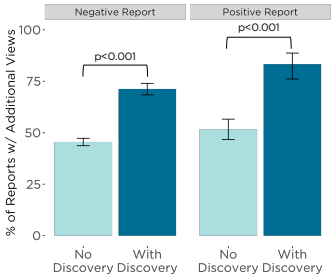

Figure 4. Clinical reports shared and downloaded

Discovery participants were more likely to download and/or share their clinical report(s) with their healthcare provider, especially in case of a positive report (60.5% vs 40.9%).

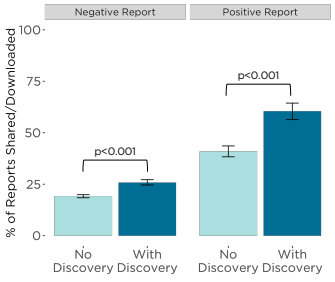

Figure 5. Health history and medication updates

Health history updates include updates to health (personal and family) and/or medication history. Discovery participants more actively updated their clinical history (70.6% vs 74.3% for negative and 76.9% vs. 83.6% for positive reports).