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White Paper findings from My Gene Counsel

Oncology Insights: The Speed and Scope of Genetic Updates

Expanding Genetic Testing to Advance Precision Medicine

One of the biggest achievements in modern medicine over the past three decades has been the integration of genetic test results in a patient's management to personalize medical care¹. Germline and somatic testing have enabled clinicians to tailor treatments based on individual genetic profiles, leading to significant advancements in areas including oncology, cardiovascular disease, renal care, and rare diseases. The rise of precision medicine has spurred pharmaceutical companies to invest heavily in targeted therapies, but these innovations are only effective if patients first undergo the necessary genetic or biomarker testing to determine eligibility.

Despite growing recognition of genetic testing's importance, adoption remains low. A June 2023 *JAMA* publication by Kurian et al. found that among cancer patients diagnosed in California and Georgia from 2013-2019, only 6.8% had undergone genetic testing by March 2021. Even among patient groups in which testing is universally recommended - such as those with male breast, ovarian, and pancreatic cancer, only 50%, 38.6%, and 5.6%, respectively, received testing. To significantly increase testing rates and enable those results to be used in clinical care, systematic workflows must be embedded into health systems to take uniform personal and family histories, run risk assessment based on national guidelines, provide pre-test information, facilitate appropriate testing and return of results in a safe, equitable manner. Busy clinicians need pre- and post-testing supports that fall into their current

workflows and take the lift off of their plates. Without such standardized measures, the vast majority of cancer patients will remain unaware of their options for precision medicines, missing opportunities for potentially life-altering therapies, such as PARP inhibitors.

“Leveraging technology in clinical workflows can help overcome barriers to efficient genetic testing so that patients and clinicians have access to the latest genetic information.”

The Challenge of Long-Term Genetic Management

For the subset of patients who do undergo genetic testing, long-term follow-up presents another major challenge. Most patients never see a genetics professional beyond their initial consultation, leaving the responsibility of ongoing management to their treating clinicians. The amount of medical knowledge generally is said to double every 73 days and this is especially true in oncology.²³ Given the rapid evolution of medical knowledge and medical management guidelines, some of which change multiple times a year (Table 1), it is unrealistic to expect clinicians to stay up to date on every development relevant to their patients.

In fact, though doctors may have the legal responsibility to inform their patients about these changes, studies have shown that general practitioners themselves feel ill-equipped to handle these regular updates and developments in genetic information due to perceived barriers such as lack of knowledge and confidence as well as lack of resources - including time - and logistical challenges to provide this information to their patients.^{15,16,19,21-25}

Some clinical updates are critical enough to change medical management for that patient - often before they are due for a routine visit - or may change recommendations for surveillance, chemoprevention, or even prophylactic surgery scheduled within that time period. The problem is magnified at a health care system level, where one high-risk patient may have as many as 5 to 10 clinicians who need accurate, in-depth information. There are additional ethical questions inherent in this dilemma, whereby patients with more resources who come to their practitioners more frequently

have the limited resources of a practitioner (or many) disproportionately directed towards them compared to a patient who may have less resources themselves.^{16, 18}

Leveraging Technology to Bridge Gaps and Barriers to Genetic Care & Information

Studies indicate that both clinicians and patients recognize digital tools as potential solutions to the logistical challenges of long-term follow-up, particularly in automating the delivery of medical management updates.^{18,19,21,24} Ensuring that both patients and clinicians receive consistent, timely, and relevant information — while providing appropriate support in an equitable and sustainable manner without excessive demands on practitioner resources - is essential.^{16, 18,21,25} Digital tools allow clinicians and patients to access information and support tailored to their respective roles and needs. Additionally, recontact can pose a significant challenge when updates arise. Automated digital solutions enable patients to retrieve updated information online, regardless of changes to their phone number or physical address.^{18,20}

The Solution: My Gene Counsel

My Gene Counsel (mGC) is a digital health company that leverages technology to address these challenges by providing a scalable, automated solution for delivering genetic testing workflows. Through its proprietary web-based platform, My Gene Counsel provides a systematic, automated system by which all patients in a system (patients presenting for annual PCP or GYN care; mammography, colonoscopy, or those diagnosed with cancer) receive a family history and guideline-based risk assessment and patients who are eligible receive pre-test support tools with best-in-class genetic counseling information. mGC delivers interactive, continuously updated genetic reports tailored to both patients and healthcare providers. These reports ensure that critical updates—such as changes in medical management guidelines, new risk estimates, or variant reclassifications—are communicated efficiently and remain accessible over time.

How Often Does Clinical Information Change?

The field of genomics is evolving rapidly and it is now recognized that current and past patients and their families should be kept up to date on important management issues. mGC addresses this growing need for delivering scalable, accurate, and up-to-date genomic information to both patients and clinicians. In 2023 and 2024, mGC digitally delivered 232 and 307 unique updates to patients, respectively, and 123 and 254 applicable to clinicians (Table 1). These updates corresponded to 93 hereditary cancer genes and pathogenic variants within those genes (Table 3). As with every update (medical management or variant reclassification),

the change is made once in the mGC Living Lab Report and then every patient and clinician with that result receives an update. The system is infinitely scalable. Updates were categorized into four groups:

- **Medical Management** changed (the most frequent for both patients (36.21% (2023) and 54.72% (2024)) and clinicians (69.11% (2023) and 67.72% (2024))
- **New Risk Estimates** changed: 16.38% (2023) and 19.87% (2024) to patients and 20.33% (2023), 29.53% (2024) to clinicians.
- **Available Resources** for the associated genetic condition changed: 41.38% (2023) and 25.41% (2024) to patients and 0% (2023), 2.76% (2024) to clinicians.

Table 2a. 2023/2024 Notifications by Category

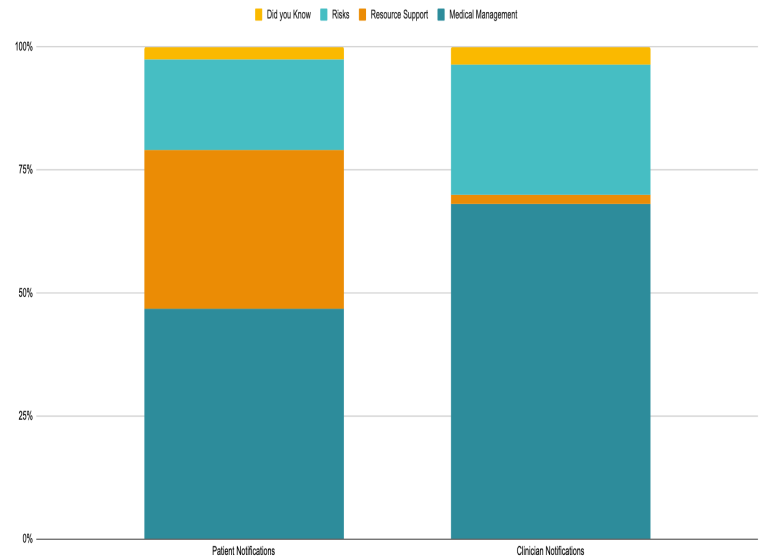


Table 1. 2023/2024 Patient and Clinician Notifications

Hereditary Cancer Notifications (93 Genes)				
Category	Patient		Clinician	
	Total		Total	
	2023 (changes/gene)	2024 (changes/ gene)	2023 (changes/gene)	2024 (changes/gene)
Medical Management	84 (0.9)	168 (1.81)	85 (0.91)	172 (1.85)
Resource: Support	96 (1.03)	78 (0.84)	0 (0)	7 (0.08)
Risks	38 (0.41)	61 (0.66)	25 (0.27)	75 (0.81)
Did You Know	14 (0.15)	0 (0)	13 (0.14)	0 (0)
TOTAL (average changes per gene)	232 (0.62)	307 (0.83)	123 (0.33)	254 (0.68)

When narrowing the list of genes to those that correspond to the American College of Medical Genetics (ACMG) list of 28 returnable genes, there were 113 patient notifications in 2023 and 137 in 2024. For clinicians, there were 68 notifications in 2023 and 119 in 2024 (Table 2). Updates were categorized into four groups:

- **Medical Management** changed (the most frequent for both patients (48.67% (2023) and 63.5% (2024)) and clinicians (82.35% (2023) and 74.79% (2024))
- **New Risk Estimates** changed: 14.16% (2023) and 28% (2024) to patients and 13.24% (2023), 24.37% (2024) to clinicians
- **Available Resources** for the associated genetic condition changed: 34.51% (2023) and 20.44% (2024) to patients and 0% (2023), 0.84% (2024) to clinicians
- **Did You Know** changed: 2.65% (2023) and 0% (2024) to patients and 4.41% (2023), 0% (2024) to clinicians.

Table 2. 2023/2024 Patient and Clinician Notifications for the ACMG Hereditary Cancer Genes

Hereditary Cancer Notifications (28 ACMG Genes)				
Category	Patient		Clinician	
	Total		Total	
	2023 (change/gene)	2024 (changes/gene)	2023 (changes/gene)	2024 (changes/gene)
Medical Management	55 (1.96)	87 (3.11)	56 (2.0)	89 (3.18)
Resource: Support	39 (1.4)	22 (0.79)	0 (0)	1 (0.04)
Risks	16 (0.57)	28 (1.0)	9 (0.32)	29 (1.04)
Did You Know	3 (0.11)	0 (0)	3 (0.11)	0 (0)
TOTAL (average changes per gene)	113 (1.01)	137 (1.23)	68 (0.61)	119 (1.07)

Each update has a multiplying impact, as it can apply to hundreds of variants and reach thousands of patients and clinicians who have received results tied to those variants.



The benefits of these tools extend beyond individual patients; they support entire healthcare systems by reducing the burden on physicians while ensuring that genetic information remains up to date. With pharma companies eager to expand precision medicine, integrating technology into clinical workflows can help align industry incentives with patient care, making genetic testing more efficient and equitable.

Conclusion

The promise of precision medicine hinges on widespread access to genetic testing and the ability to keep genetic information current. While legislative efforts to improve access are a step in the right direction, systemic barriers still remain, particularly in long-term follow-up and equitable test implementation. Pharmaceutical companies have a clear incentive to drive genetic testing adoption, and their investments can play a crucial role in expanding access. However, ensuring that these efforts serve patients' best interests requires a structured, technology-driven approach that prioritizes timely, accurate, equitable, sustainable, and evolving information for both patients and clinicians.

By leveraging digital tools to streamline genetic updates and medical management, we can bridge the gap between genetic discovery and patient care and maximize the impact of precision medicine for all.

Table 3a. Notifications by Gene (ACMG Hereditary Cancer Genes)

Notifications by Gene				
Cancer - ACMG				
Patient			Clinician	
Name	2023	2024	2023	2024
APC	3	8	2	7
BMPR1A	2	2	1	4
BRCA1	9	11	7	9
BRCA2	9	7	7	8
MAX	2	1	1	1
MEN1	6	16	2	16
MLH1	2	8	0	8
MSH2	2	8	0	8
MSH6	2	9	0	8
MUTYH x2	2	1	0	1
NF2	5	8	3	5
PALB2	8	3	6	2
PMS2	3	8	1	7
PTEN	4	1	2	0
RB1	1	1	0	0
RET	8	13	6	10
SDHAF2	3	0	2	0
SDHB	4	2	3	2
SDHC	4	1	3	1
SDHD	5	0	4	0
SMAD4	2	2	1	0
STK11	4	3	1	2
TMEM127	2	1	1	1
TP53	4	3	2	3
TSC1	7	6	6	6
TSC2	7	7	6	6
VHL	2	3	1	2
WT1	1	4	0	2

Table 3b. Notifications by Gene (Non-AGMC Genes)

Notifications by Gene				
Name	Patient		Clinician	
	2023	2024	2023	2024
ABRAXAS1	0	2	0	2
AIP	0	4	1	4
ALK	1	1	0	1
ATM	5	4	3	3
ATR	0	3	0	3
AXIN2	2	1	0	0
BAP1	1	4	0	4
BLM	1	2	0	2
BARD1	4	2	3	1
BRIP1	3	3	2	3
CDC73	0	1	0	1
CDH1	4	5	1	4
CDK4	1	1	0	0
CDKN1B	3	5	2	4
CDKN2A	1	1	0	3
CEBPA	0	1	0	0
CFTR	0	2	0	1
CHEK2	8	15	6	13
CTNNA1	3	1	1	0
DICER1	1	2	0	1
EGFR	2	4	1	4
EPCAM	1	9	0	8
FANCC	2	2	0	2
FANCM	0	1	0	2
FH	1	7	0	5
FLCN	0	1	0	0
GALNT12	1	1	0	0
GREM1	1	2	0	1
HOXB13	1	1	0	0
KIF1B	0	1	0	1
KIT	1	2	0	1

LZTR1	4	3	4	1
MEN2B	1	0	0	0
MET	1	0	0	0
MITF	2	1	1	0
MRE11A	1	4	0	4
MSH2-EPCAM	2	9	0	8
MSH3	1	1	0	0
MUTYH x1	2	2	1	1
NBN	3	3	2	5
Negative	4	2	1	0
NF1	6	5	3	6
NTHL1	6	2	4	1
PDGFRA	1	1	0	0
PIK3CA	0	1	0	1
POLD1	2	4	0	3
POLE	2	3	0	3
POT1	3	2	2	1
PRKAR1A	4	6	4	4
PTCH1	0	2	0	2
RAD50	2	2	1	2
RAD51B	0	3	0	4
RAD51C	6	2	4	1
RAD51D	6	3	4	1
RECQL	2	2	1	2
RECQL4	0	2	0	2
RINT1	0	2	0	2
RNF43	1	1	0	0
RPS20	1	2	0	1
RUNX1	0	1	0	0
SDHA	4	0	3	0
SMARCA4	1	2	0	1
SMARCE1	0	1	0	1
SUFU	0	2	0	2
VUS	1	1	0	0
XRCC2	2	2	1	2

References

1. Ruth March. The Changing Landscape of Precision Medicine. 2023. [Website](#).
2. Mikael Dolsten and Morten Sorgaard. Pfizer's Precision Medicine Pipeline and Technology Portfolio. The Journal of Precision Medicine. 2021. 7 (4). [Website](#)
3. Kurian AW, Abrahamse P, Furgal A, et al. JAMA Germline Genetic Testing After Cancer Diagnosis. JAMA. 2023 Jul 3;330(1):43-51. [Pubmed](#)
4. Governor Newsom Signs Game-Changing Legislation Aimed at Improving Health Outcomes, Quality of Life for Californians. American Cancer Society. Accessed October 7, 2023. [Website](#)
5. Staff Reporter. Texas Law Mandating Biomarker Testing to Take Effect. Precision Medicine Online. August 30, 2023. [Website](#)
6. National Ambulatory Medical Care Survey: 2010 Summary Tables. *Kaiser Family Foundation*. [KFF.org](#).
7. My Gene Counsel. How Often Do Medical Management Guidelines Change for People with Germline Genetic Findings: A Solution for Keeping Patients and Providers Updated. Oct 2019. [MyGeneCounsel.com](#)
8. My Gene Counsel. Paradigm Shift in Genetic Testing: Genetics for a Lifetime. March 2022. [MyGeneCounsel.com](#)
9. Wendler, Ronda. Genetic testing found a variant of uncertain significance. Now what? September 29, 2021. [MDAnderson Website](#).
10. Rhem H, Alaimo JT, Aradhya S, et al. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. Genetics in Medicine. 2023 25 (12). [Website](#)
11. Esterling L, Wijayatunge R, Brown K, et al. Impact of a Cancer Gene Variant Reclassification Program Over a 20-Year Period. JCO Precis Oncol. 2020; 4. 2020 Aug 27. [PubMed](#)
12. Britt Johnson and Flavia M. Facio. Innovations in variant interpretation: Navigating the VUS landscape. Invitae Webinar viewed Aug 30, 2023. [Website](#)

13. Myriad Website: Variant Classification Request. Accessed 10.1.2023 [Website](#)
14. Kurian AW, Ward KC, Hamilton AS, et al. Uptake, Results, and Outcomes of Germline Multiple-Gene Sequencing After Diagnosis of Breast Cancer. JAMA Oncol. 2018 Aug 1;4(8):1066-1072. [Pubmed](#)
15. Practical Considerations for Reinterpretation of Individual Genetic Variants
<https://pmc.ncbi.nlm.nih.gov/articles/PMC10408279/>
16. How practice setting affects family physicians' views on genetic screening: a qualitative study
<https://bmcprimcare.biomedcentral.com/articles/10.1186/s12875-021-01492-y>
17. How do non-geneticist physicians deal with genetic tests? A qualitative analysis.
<https://www.nature.com/articles/s41431-021-00884-z>
18. Clinical guidelines - Canadian College of Medical Geneticists: clinical practice advisory document – responsibility to recontact for reinterpretation of clinical genetic testing.
<https://jmg.bmj.com/content/61/12/1123>
19. What do cancer genetic providers want us to know about variant reclassification and recontact that we are not asking? A thematic analysis of open-ended survey responses.
<https://acsjournals.onlinelibrary.wiley.com/doi/full/10.1002%2Fcncr.35429> **
20. Practices and Views of US Oncologists and Genetic Counselors Regarding Patient Recontact After Variant Reclassification: Results of a Nationwide Survey.
<https://ascopubs.org/doi/10.1200/PO.23.00079>
21. Primary care doctors need help talking to patients about genetic tests.
<https://www.fiercehealthcare.com/practices/primary-care-doctors-genetic-tests-diane-hauser>
22. Initiative helps physicians interpret genetic test results.
<https://news.vumc.org/2021/09/01/initiative-helps-physicians-interpret-genetic-test-results/>
23. Docs Struggle to Keep Up With the Flood of New Medical Knowledge. Here's Advice.
<https://www.medscape.com/viewarticle/989043>

24. Changing interpretations, stable genes: responsibilities of patients, professionals, and policy makers in the clinical interpretation of complex genetic information.

<https://www.nature.com/articles/gim2008120>

25. Bridging the gap in genetics: a progressive model for primary to specialist care.

<https://bmcmmededuc.biomedcentral.com/articles/10.1186/s12909-019-1622-y> ** - This one basically defines the solution as exactly what MGC is doing!! I will definitely incorporate this one into the white paper language.