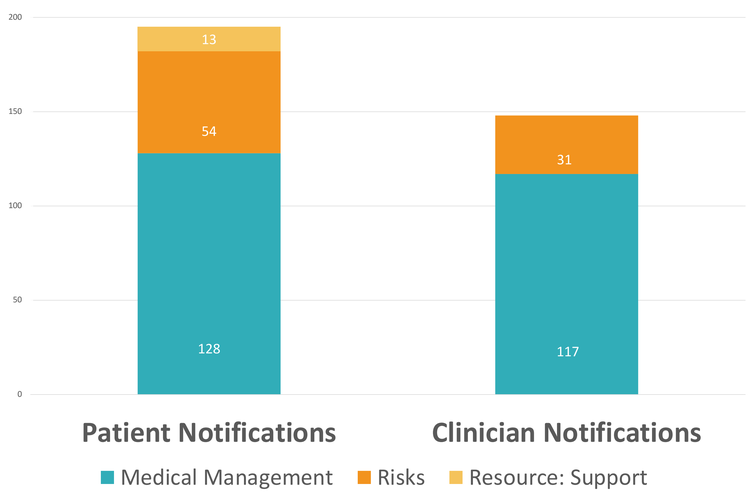
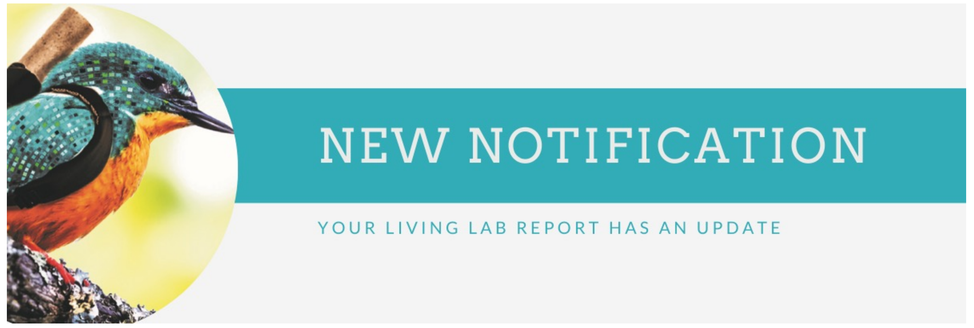
Accelerating Beyond Transactional Genetics

INTRO: The current clinical model in cancer genetics is transactional. The work-flow of risk assessment, evaluation, genetic testing and genetic counseling of a single patient occurs in rapid succession and is rarely updated or revisited. Patients who were found to have a genetic condition just one year ago, have lived through multiple updates to management guidelines, but rarely learn about them. Some of these updates are critical enough to change their medical management. A system that delivers the latest data and guidelines into the hands of patients and clinicians requires a technical solution.

METHODS: Over the course of one year (2022), My Gene Counsel tracked, reviewed, collated, and digitally delivered updates corresponding to 66 hereditary cancer genes to the patients who carried pathogenic variants within those genes and their clinicians.

RESULTS: Three hundred and forty-three unique notifications were delivered electronically, 195 to patients and 148 to clinicians and categorized into three groups:





These **critical updates** occurred within one year in the field of germline cancer genetics. The **magnitude of the update** (medical management or variant reclassification), **is compounded** by the number of patients and clinicians for which they apply.

We must stop practicing **transactional** genetic testing and provide **lifetime access** to digital genetic information that updates.

With **technical solutions**, health systems can overcome barriers to efficiently scale genetic testing and ensure that patients and clinicians have access to the most recent and relevant genetic information.

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Category	Patient Notifications		Clinician Notifications	
	Total	per Gene	Total	per Gene
Medical Management	128	2.13	117	1.95
Risks	54	0.90	31	0.52
Resource: Support	13	0.22	0	0
TOTAL	**195**	**3.25**	**148**	**2.47**

Notifications by Gene

Gene	Patient	Clinician	Gene	Patient	Clinician
APC	6	3	MUTYH x1	1	1
ATM	7	5	MUTYH x2	1	0
AXIN2	1	0	NBN	4	4
BAP1	4	4	NF1	3	4
BARD1	2	2	NTHL1	1	0
BLM	4	4	PALB2	5	5
BMPR1A	1	0	PMS2	3	2
BRCA1	4	4	POLD1	4	3
BRCA2	4	4	POLE	5	4
BRIP1	3	2	PTEN	5	3
CDH1	6	6	RAD50	2	2
CDKN2A	1	1	RAD51C	6	5
CHEK2	10	7	RAD51D	6	5
CTNNA1	3	3	RECQL	2	2
EPCAM	2	1	RET	10	7
FANCC	3	2	RNF43	1	0
FH	1	0	RPS20	1	0
FLCN	0	1	SDHA	5	3
GALNT12	2	1	SDHAF2	3	1
GREM1	1	0	SDHB	5	3
KIT	1	1	SDHC	6	4
MAX	4	2	SDHD	5	3
MEN1	1	2	SMAD4	1	0
MET	1	0	STK11	5	4
MLH1	2	1	TMEM127	2	1
MRE11A	2	2	TP53	5	5
MSH2	1	1	TSC1	5	5
MSH2-EPCAM	2	1	TSC2	5	5
MSH3	1	1	VHL	3	3
MSH6	3	2	XRCC2	2	2

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