Paradigm Shift in Genetic Testing:
Precision Medicine Over the Lifetime

INTRO: Genetic testing can be lifesaving, leading to tailored management and risk-reducing options. This is especially true when the American College of Medical Genetics and Genomics SFv2.0 list of 59 genes is considered, most of which have established interventions to prevent or significantly reduce morbidity and mortality. As new data related to these genes emerge on risks, surveillance and prevention, clinically relevant updates must be passed on to patients known to have that genetic predisposition and their clinicians. To accomplish this, the genomics industry needs to harness a technical solution.

METHODS: Over the course of one year (2021), we delivered, tracked, reviewed, and collated notifications to patients and their clinicians corresponding to the ACMG SFv2.0 list of, then, 59 genes. ACMG SFv3.0 was published mid-year, thus SFv2.0 was used.

RESULTS: Four hundred and nineteen notifications were delivered and categorized into four groups:

Category	Patient Notifications		Clinician Notifications	
	Total	per Gene	Total	per Gene
Medical Management	141	2.39	118	2.00
Risks	72	1.22	0	0
Family Information	32	0.54	2	0.03
Resource: Support	29	0.49	25	0.42
TOTAL	**274**	**4.64**	**145**	**2.46**

my GENE COUNSEL

CONCLUSION: We must move beyond one-time return of genetic test results as the end of genetic service delivery. Technical solutions can put the latest data and guidelines into the hands of patients and clinicians to support **proactive healthcare over a lifetime**.

The shift to integrate **updating genetic counseling information** will bring the genomics revolution to fruition.

Digital tools will be critical in scaling this process for patients, clinicians, and precision medicine partners.

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Notifications by Gene

Hereditary Cancer (25)			Hereditary Cardiovascular (30)		
Gene	Patient	Clinician	Gene	Patient	Clinician
BMPR1A	2	1	ACTC1	10	5
BRCA1	9	5	APOB	5	3
BRCA2	8	4	DSC2	2	1
MEN1	7	5	DSG2	2	1
MLH1	8	6	DSP	3	2
MSH2	9	6	FBN1	5	3
MSH6	8	6	GLA	3	1
MUTYH x2	1	1	KCNH2	4	3
NF2	1	0	KCNQ1	4	3
PMS2	7	6	LDLR	5	3
PTEN	3	1	LMNA	4	1
RET	4	2	MYBPC3	10	5
SDHAF2	6	1	MYH7	10	5
SDHB	7	2	MYL2	10	5
SDHC	6	1	MYL3	10	5
SDHD	7	2	PCSK9	5	3
SMAD4	2	1	PKP2	2	1
STK11	2	1	PRKAG2	7	5
TP53	8	2	RYR2	2	1
TSC1	6	3	SCN5A	4	3
TSC2	6	3	SMAD3	3	4
VHL	7	3	TGFBR1	1	1
WT1	4	2	TGFBR2	1	1
ACMG Other Genes (4)			TMEM43	2	1
Gene	Patient	Clinician	TNNI3	10	5
ATP7B	1	0	TNNT2	10	5
OTC	1	0	TPM1	10	5

*ACMG SFv3.1 includes a more extensive list of genes for which there are a greater number notifications and patients and clinicians that need to be updated.