Precision Medicine in Practice

Keeping Patients Up to Date After Genetic Testing

INTRO: Exponential growth is occurring in clinical genomics. A patient found to have a pathogenic variant 10 years ago has likely lived through dozens of updates to risk estimates and management guidelines related to that diagnosis. Most patients who undergo genetic testing never speak to a certified genetic counselor, or do so once or twice, and then never again. Few patients or clinicians have the tools, time, or knowledge to find, track, and interpret new data on genetic findings as they emerge.

METHODS: Over the course of one year, we tracked the number and type of relevant updates for the American College of Medical Genetics SFv2.0 list of 59 genes related to:

- Medical management changes
- New risk estimates
- Information for family members
- Resources
- General information related to the genetic condition

Category	Number of Notifications
Medical Management	111
Risks	66
Family Information	59
General Information	71
Resources/Support	93
TOTAL	**400**

my GENE COUNSEL

We must think beyond the one-time return of genetic test results to the integration of **continuously updating genetic counseling information** in order to bring the genomics revolution to fruition.

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

AUTHORS:
Danielle Bonadies, MS, CGC
Meagan Farmer, MS, CGC, MBA
Michelle Kempf-Weibel, MS, CGC
Ellen Matloff, MS, CGC

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Category	Cancer (25 Genes)		Cardiac (30 Genes)	
	Total	Notifications per Gene	Total	Notifications per Gene
Medical Mgmt	75	3.0	36	1.2
Risks	35	1.4	31	1.0
Family Info	27	1.1	30	1.0
General Info	35	1.4	36	1.2
Resources	61	2.4	32	1.1
Total	**233**	**9.3**	**165**	**5.5**

Note: The other 4 ACMG genes had 2 notifications (0.5 per gene) related to Family Information.

GENE	UPDATES	GENE	UPDATES	GENE	UPDATES
CANCER		*STK11*	12	*MYBPC3*	2
APC	15	*TP53*	13	*MYH11*	5
BMPR1A	8	*TSC1*	9	*MYH7*	2
BRCA1	21	*TSC2*	9	*MYL2*	2
BRCA2	19	*VHL*	11	*MYL3*	2
MEN1	6	*WT1*	5	*PCSK9*	8
MLH1	14	CARDIAC		*PKP2*	5
MSH2	15	*ACTA2*	8	*PRKAG2*	3
MSH6	13	*ACTC1*	1	*RYR2*	10
MUTYH	3	*APOB*	8	*SCN5A*	10
NF2	8	*COL3A1*	1	*SMAD3*	5
PMS2	10	*DSC2*	5	*TGFBR1*	11
PTEN	10	*DSG2*	6	*TGFBR2*	11
RB1	5	*DSP*	8	*TMEM43*	5
RET	3	*FBN1*	8	*TNNI3*	3
SDHAF2	3	*GLA*	1	*TNNT2*	3
SDHB	7	*KCNH2*	10	*TPM1*	3
SDHC	8	*KCNQ1*	9	OTHER	
SDHD	7	*LDLR*	8	*CACNA1S*	1
SMAD4	8	*LMNA*	2	*RYR1*	1