Clinic : Grady Memorial Hospital, Atlanta, GA

Gender : Female DOB : 10/21/1980

HIGH RISK

Gene	Start	Class	Mutation	CADD
Gene	Start	Variant_Classification	Amino_Acid_change	Cosmic
FAM113A	2838470	Silent	p.P201	.67177
KIAA1009	84195006	Nonsense	p.S302*	.95415
L1TD1	62210940	Silent	p.K722	.00056
MAD2L2	11676107	Deletion	p.L123fs	0
OR2B11	247451959	Deletion	p.L9fs	0
PRDM2	13779899	Insertion	p.704in_frame_insP	0
RHBG	156384556	Deletion	p.R425fs	0
TAF1B	9904895	Deletion	p.V282fs	0
TMEM37	119437075	Insertion	p.T70in_frame_insSVP	0
ZNF804A	184937484	Missense	p.N696K	.12369
TSPYL1	116279302	Insertion	$p.176in_frame_insV$	0
AKAP9	92022864	Insertion	p.1336in_frame_insQ	0
NCRNA00174	66377558	Insertion	p.T17fs	0
OR11G2	20198016	Insertion	p.G229fs	0
CYP2A7	40880515	Missense	p.S153A	.0028
TMPRSS3	42383199	Splice_Site	e7-1	.94873
TMPRSS3	42383199	Splice_Site	e7-1	.94873
BIRC6	32545808	Missense	p.A4253V	.96697
ACO1	32408614	Missense	p.V123L	.9893
FREM2	38689594	Missense	p.D750E	.74392
TP53	7675238	Splice_Site	e4-2	.99475
TP53	7675238	Splice_Site	e4-2	.99475
BRCA1	43092919	Deletion	NA	0.99
BRCA1	43093220	Insertion	NA	0.99
BRCA1	43092919	Insertion	NA	0.99
BRCA1	43092919	Indel	NA	0.99

Gene Details: MSH2 c.8C>G : NM-000251.2

Functional Significance: Deleterious

The heterozycous germline MSH2 c.8C>G is predicted to result in the pre-

mature truncation of the MSH2 protein due to abnormal protein production and/or function.

Clinical Significance: High Cancer Risk

This mutation is associated with increased cancer risk and should be regarded as clinically significant.

Additional Information

Genes Analyzed: APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD51C, SMAD4, STK11, TP53

Disclaimer: The techincal specifications summary of ICRC Gene Expose describes the analysis, method, performance, nomenclature, and interpretive criteria of this test. The classification and interpretation of variants identified reflect the current state of scientific understanding at the time of this report.