

Table 1

Phenotype	GeneReviews	Prevalence	95%-CI PAF	Max Penetrance
APC-Polyposis	Yes	3.20e-05	3.63e-03	8.81e-03
Arrhythm RV CM	Yes	1.00e-03	5.18e-03	1.93e-01
Breast-Ovarian Cancer	No	9.62e-03	2.62e-03	1.00e+00
Brugada Syndrome	Yes	2.40e-03	6.04e-03	3.98e-01
CPV Tachycardia	Yes	1.00e-04	3.32e-06	1.00e+00
Dilated CM	Yes	4.00e-03	6.97e-03	5.74e-01
Ehlers-Danlos	Yes	5.00e-06	9.68e-05	5.16e-02
Endocrine Neoplasias 1	Yes	3.33e-05	1.86e-07	1.00e+00
Endocrine Neoplasias 2	Yes	2.86e-05	8.08e-04	3.54e-02
Fabry's Disease	Yes	6.25e-04	9.50e-04	6.58e-01
Hypercholesterolemia	Yes	5.00e-03	5.84e-03	8.55e-01
Hypertrophic CM	Yes	2.00e-03	9.14e-03	2.19e-01
Juvenile Polyposis	No	6.25e-05	2.82e-05	1.00e+00
Li-Fraumeni	No	2.00e-04	1.09e-03	1.83e-01
Loeys-Dietz	No	1.00e-05	9.55e-05	1.05e-01
Long QT Syndrome	Yes	5.00e-04	9.94e-03	5.03e-02
Lynch Syndrome	Yes	2.27e-03	1.86e-03	1.00e+00
Malignant Hyperthermia	Yes	3.00e-05	1.35e-03	2.21e-02
Marfan Syndrome	Yes	2.00e-04	1.43e-03	1.40e-01
MUTYH-Polyposis	No	5.00e-05	5.92e-06	1.00e+00
Neurofibromatosis	Yes	1.67e-05	7.98e-06	1.00e+00
OTC Deficiency	Yes	7.14e-05	2.13e-04	3.35e-01
Paragangliomas	No	1.00e-06	4.32e-04	2.32e-03
Peutz-Jeghers	Yes	4.00e-05	1.86e-07	1.00e+00
PTEN Hamartomas	Yes	5.00e-06	1.70e-04	2.93e-02
Retinoblastoma	Yes	6.67e-05	8.59e-05	7.76e-01
Thoracic Aortic	No	1.00e-04	4.55e-04	2.20e-01
Tuberous Sclerosis	Yes	1.72e-04	1.93e-04	8.92e-01
Von Hippel-Lindau	No	1.89e-05	1.26e-04	1.49e-01
Wilms Tumor	No	1.00e-04	1.86e-07	1.00e+00
Wilson Disease	Yes	1.00e-04	1.70e-06	1.00e+00