**GenePrism: Actionable Insights**

# Conditions and genes tested by category

**SOURCE:** *American College of Medical Genetics and Genomics policy statement (Kalia SS et al., 2017). Available at https://www.ncbi.nlm.nih.gov/pubmed/27854360)*

| CONDITIONS | CATEGORY(IES) | GENE(S) |
|---|---|---|
| Hereditary breast and ovarian cancer (HBOC) | Cancer | BRCA1/2 |
| Li-Fraumeni syndrome (LFS) | Cancer | TP53 |
| Peutz-Jeghers syndrome (PJS) | Cancer | STK11 |
| Lynch syndrome (LS) | Cancer | MLH1, MSH2, MSH6, PMS2 |
| Familial adenomatous polyposis (FAP) and Attenuated FAP | Cancer | APC (not including I1307K variant) |
| Hereditary colorectal cancer | Cancer | APC (I1307K variant only) |
| MUTYH-associated polyposis (MAP) | Cancer | MUTYH |
| Juvenile polyposis syndrome (JPS) | Cancer | BMPR1A, SMAD4 |
| von Hippel-Lindau syndrome (VHL) | Cancer | VHL |
| Multiple endocrine neoplasia type 1 (MEN1) | Cancer | MEN1 |
| Multiple endocrine neoplasia, type 2 (MEN2, including Familial medullary thyroid cancer or FMTC) | Cancer | RET |
| PTEN hamartoma syndrome (PHTS, including Cowden syndrome) | Cancer / Other | PTEN |
| Hereditary retinoblastoma | Cancer | RB1 |
| Hereditary paraganglioma-pheochromocytoma syndrome (PGL-PCC) | Cancer | SDHD, SDHAF2, SDHC, SDHB |
| Tuberous sclerosis complex (TSC) | Cancer / Other | TSC1, TSC2 |
| WT1-related Wilms tumor | Cancer / Other | WT1 |
| Neurofibromatosis type 2 (NF2) | Cancer / Other | NF2 |

| CONDITIONS | CATEGORY(IES) | GENE(S) |
|---|---|---|
| Vascular Ehlers-Danlos syndrome (vEDS) | Connective tissue / Heart | COL3A1 |
| Marfan syndrome (MFS) | Connective tissue / Heart | FBN1 |
| Loeys-Dietz syndrome (LDS) | Connective tissue / Heart | TGFBR1, TGFBR2, SMAD3 |
| Familial thoracic aortic aneurysms and dissections (TAAD) | Heart | ACTA2, MYH11 |
| Inherited hypertrophic cardiomyopathy (HCM) and/or dilated cardiomyopathy (DCM) | Heart | MYBPC3, MYH7, TNNT2, TNNI3, TPM1, ACTC1 |
| Inherited hypertrophic cardiomyopathy (HCM) | Heart | MYL3, PRKAG2, MYL2 |
| Dilated cardiomyopathy (DCM) and LMNA-related disorders | Heart / Other | LMNA |
| Catecholaminergic polymorphic tachycardia (CPVT) | Heart | RYR2 |
| Arrhythmogenic right ventricular cardiomyopathy (ARVC) | Heart | PKP2, DSP, DSC2, TMEM43, DSG2 |
| Inherited cardiac arrhythmias (including long QT syndrome or LQTS, Brugada syndrome or BrS, short QT syndrome or SQTS, Romano-Ward syndrome) | Heart | KCNQ1, KCNH2, SCN5A |
| Familial hypercholesterolemia (FH) | Heart / Metabolism | LDLR, APOB, PCSK9 |
| Wilson disease | Metabolism | ATP7B |
| Fabry disease | Metabolism | GLA |
| Ornithine transcarbamylase (OTC) deficiency | Metabolism | OTC |
| Malignant hyperthermia susceptibility | Metabolism | RYR1, CACNA1S |